

Genetic Testing

Record ID

Genetic Testing Information

Type(s) of genetic testing done

- ☐ Sequence-based genetic testing
☐ Chromosomal microarray-based genetic testing
☐ Karyotype-based genetic testing
☐ Repeat expansion-based genetic testing
☐ Other

Number of sequence-based genetic tests

Upload report for sequence-based test 1

Is variant information obtained directly from testing report for sequence-based test 1?

- ☐ Yes
☐ No

Sequence-based test 1 company

- ☐ Ambry Genetics
☐ ARUP Laboratories
☐ Athena Diagnostics
☐ Baylor Genetics
☐ Blueprint Genetics
☐ Color Genetics
☐ Fulgent Genetics
☐ GeneDx
☐ Greenwood Genetics
☐ Invitae
☐ LabCorp/Integrated Genetics
☐ Laboratory for Molecular Medicine/Partners
☐ Mayo Clinic
☐ Medical Neurogenetics (MNG) Laboratories
☐ Myriad Genetics/Counsyl
☐ Natera Genetics
☐ Prevention Genetics
☐ Quest Diagnostics
☐ Seattle Children's Hospital Genetics Laboratories
☐ University of Chicago Genetic Services
☐ UW Lab Medicine/NCGL/CDL
☐ Variantyx
☐ Veritas Genetics
☐ Other

Specify 'Other' company for sequence-based test 1

Sequence-based test 1 name

Sequence-based test 1 report date

Is this an amended/reanalysis report for sequence-based test 1?

- ☐ Yes
☐ No

Specify the report date of the amended/reanalysis report for sequence-based test 1

Was RNA analysis included in sequence-based test 1?

- ☐ Yes
☐ No

Sequence-based test 1 type

- ☐ Genome
☐ Exome
☐ Panel
☐ Single gene
☐ Single variant
☐ Other

Specify single gene targeted in sequence based test 1

Specify single variant targeted in sequence-based test 1

If sequence-based test 1 is exome or genome, please select one of the following

- ☐ Proband-only
☐ Duo (i.e., proband and one parent)
☐ Trio (i.e., proband and both parents)
☐ Quad (i.e., proband, sibling, and both parents)
☐ Other

Specify the number of genes targeted in the panel for sequence-based test 1

Specify the names of the genes targeted in the panel for sequence-based test 1

Was sequence analysis included for at least one of the genes tested in sequence-based test 1?

- ☐ Yes
☐ No

Was deletion/duplication analysis included for at least one of the genes tested in sequence-based test 1?

- ☐ Yes
☐ No

Proband sample source for sequence-based test 1

- ☐ Blood
☐ Saliva
☐ Buccal swab
☐ Skin biopsy or fibroblast culture
☐ Cancer/tumor sample
☐ Non-malignant surgical sample
☐ Amniotic fluid
☐ Placenta/chorionic villi
☐ Cell free DNA
☐ Other

Specify the type of cancer/tumor sample used in sequence-based test 1

Specify the type of non-malignant surgical sample used in sequence-based test 1

Specify 'Other' proband sample source used in sequence based test 1

Were any variants reported for sequence-based test 1?

- ☐ Yes
☐ No

Number of variants reported in sequence-based test 1

Sequence-based test 1 variant 1 gene (i.e., LDLR)

Sequence-based test 1 variant 1 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 1 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 1 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 1 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 1 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 1 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 1 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 1 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 1

Sequence-based test 1 variant 1 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 1

Sequence-based test 1 variant 1 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 1

Is sequence-based test 1 variant 1 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 1 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 1?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 1 interpretation summary

Sequence-based test 1 variant 2 gene (i.e., LDLR)

Sequence-based test 1 variant 2 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 2 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 2 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 2 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 2 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 2 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 2 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 2

Sequence-based test 1 variant 2 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 2

Sequence-based test 1 variant 2 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 2

Is sequence-based test 1 variant 2 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 1 variant 2 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 1 variant 2?

- ☐ Yes
☐ No

Sequence-based test 1 variant 2 interpretation summary

Sequence-based test 1 variant 3 gene (i.e., LDLR)

Sequence-based test 1 variant 3 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 3 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 3 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 3 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 3 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 3 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 3 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 3 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 3

Sequence-based test 1 variant 3 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 3

Sequence-based test 1 variant 3 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 3

Is sequence-based test 1 variant 3 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 3 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 1 variant 3? ☐ Yes
☐ No

Sequence-based test 1 variant 3 interpretation summary

Sequence-based test 1 variant 4 gene (i.e., LDLR)

Sequence-based test 1 variant 4 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 1 variant 4 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 1 variant 4 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 4 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 4 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 4 rsID, if present
(i.e., rs193922570)

Sequence-based test 1 variant 4 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
1 variant 4

Sequence-based test 1 variant 4 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 4

Sequence-based test 1 variant 4 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 4

Is sequence-based test 1 variant 4 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 4 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 4?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 4 interpretation summary

Sequence-based test 1 variant 5 gene (i.e., LDLR)

Sequence-based test 1 variant 5 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 5 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 5 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 5 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 5 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 5 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 5 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 5

Sequence-based test 1 variant 5 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 5

Sequence-based test 1 variant 5 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 5

Is sequence-based test 1 variant 5 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 1 variant 5 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 1 variant 5?

- ☐ Yes
☐ No

Sequence-based test 1 variant 5 interpretation summary

Sequence-based test 1 variant 6 gene (i.e., LDLR)

Sequence-based test 1 variant 6 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 6 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 6 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 6 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 6 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 6 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 6 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 6 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 6

Sequence-based test 1 variant 6 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 6

Sequence-based test 1 variant 6 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 6

Is sequence-based test 1 variant 6 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 6 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 1 variant 6? ☐ Yes
☐ No

Sequence-based test 1 variant 6 interpretation summary

Sequence-based test 1 variant 7 gene (i.e., LDLR)

Sequence-based test 1 variant 7 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 1 variant 7 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 1 variant 7 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 7 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 7 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 7 rsID, if present
(i.e., rs193922570)

Sequence-based test 1 variant 7 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 7 mutation type

☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
1 variant 7

Sequence-based test 1 variant 7 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 7

Sequence-based test 1 variant 7 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 7

Is sequence-based test 1 variant 7 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 7 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 7?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 7 interpretation summary

Sequence-based test 1 variant 8 gene (i.e., LDLR)

Sequence-based test 1 variant 8 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 8 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 8 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 8 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 8 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 8 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 8 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 8 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 8

Sequence-based test 1 variant 8 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 8

Sequence-based test 1 variant 8 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 8

Is sequence-based test 1 variant 8 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 1 variant 8 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 1 variant 8?

- ☐ Yes
☐ No

Sequence-based test 1 variant 8 interpretation summary

Sequence-based test 1 variant 9 gene (i.e., LDLR)

Sequence-based test 1 variant 9 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 9 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 9 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 9 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 9 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 9 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 9 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 9 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 9

Sequence-based test 1 variant 9 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 9

Sequence-based test 1 variant 9 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 9

Is sequence-based test 1 variant 9 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 9 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 1 variant 9?

☐ Yes
☐ No

Sequence-based test 1 variant 9 interpretation summary

Sequence-based test 1 variant 10 gene (i.e., LDLR)

Sequence-based test 1 variant 10 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 1 variant 10 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 1 variant 10 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 10 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 10 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 10 rsID, if present
(i.e., rs193922570)

Sequence-based test 1 variant 10 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 10 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
1 variant 10

Sequence-based test 1 variant 10 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 10

Sequence-based test 1 variant 10 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 10

Is sequence-based test 1 variant 10 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 10 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 10?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 10 interpretation summary

Sequence-based test 1 variant 11 gene (i.e., LDLR)

Sequence-based test 1 variant 11 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 11 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 11 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 11 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 11 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 11 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 11 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 11 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 11

Sequence-based test 1 variant 11 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 11

Sequence-based test 1 variant 11 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 11

Is sequence-based test 1 variant 11 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 1 variant 11 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 1 variant 11?

- ☐ Yes
☐ No

Sequence-based test 1 variant 11 interpretation summary

Sequence-based test 1 variant 12 gene (i.e., LDLR)

Sequence-based test 1 variant 12 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 12 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 12 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 12 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 12 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 12 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 12 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 12 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 12

Sequence-based test 1 variant 12 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 12

Sequence-based test 1 variant 12 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 12

Is sequence-based test 1 variant 12 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 12 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 1 variant 12?

- ☐ Yes
☐ No

Sequence-based test 1 variant 12 interpretation summary

Sequence-based test 1 variant 13 gene (i.e., LDLR)

Sequence-based test 1 variant 13 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 1 variant 13 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 1 variant 13 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 13 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 13 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 13 rsID, if present
(i.e., rs193922570)

Sequence-based test 1 variant 13 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 13 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
1 variant 13

Sequence-based test 1 variant 13 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 13

Sequence-based test 1 variant 13 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 13

Is sequence-based test 1 variant 13 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 13 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 13?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 13 interpretation summary

Sequence-based test 1 variant 14 gene (i.e., LDLR)

Sequence-based test 1 variant 14 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 14 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 14 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 14 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 14 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 14 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 14 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 14 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 14

Sequence-based test 1 variant 14 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 14

Sequence-based test 1 variant 14 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 14

Is sequence-based test 1 variant 14 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 1 variant 14 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 1 variant 14?

- ☐ Yes
☐ No

Sequence-based test 1 variant 14 interpretation summary

Sequence-based test 1 variant 15 gene (i.e., LDLR)

Sequence-based test 1 variant 15 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 15 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 15 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 15 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 15 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 15 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 15 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 15 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 15

Sequence-based test 1 variant 15 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 15

Sequence-based test 1 variant 15 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 15

Is sequence-based test 1 variant 15 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 15 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 1 variant 15? ☐ Yes
☐ No

Sequence-based test 1 variant 15 interpretation summary

Sequence-based test 1 variant 16 gene (i.e., LDLR)

Sequence-based test 1 variant 16 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 1 variant 16 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 1 variant 16 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 16 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 16 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 16 rsID, if present
(i.e., rs193922570)

Sequence-based test 1 variant 16 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 16 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
1 variant 16

Sequence-based test 1 variant 16 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 16

Sequence-based test 1 variant 16 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 16

Is sequence-based test 1 variant 16 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 16 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 16?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 16 interpretation summary

Sequence-based test 1 variant 17 gene (i.e., LDLR)

Sequence-based test 1 variant 17 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 17 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 17 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 17 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 17 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 17 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 17 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 17 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 17

Sequence-based test 1 variant 17 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 17

Sequence-based test 1 variant 17 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 17

Is sequence-based test 1 variant 17 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 1 variant 17 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 1 variant 17?

- ☐ Yes
☐ No

Sequence-based test 1 variant 17 interpretation summary

Sequence-based test 1 variant 18 gene (i.e., LDLR)

Sequence-based test 1 variant 18 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 18 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 18 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 18 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 18 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 18 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 18 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 18 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 18

Sequence-based test 1 variant 18 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 18

Sequence-based test 1 variant 18 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 18

Is sequence-based test 1 variant 18 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 18 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 1 variant 18? ☐ Yes
☐ No

Sequence-based test 1 variant 18 interpretation summary

Sequence-based test 1 variant 19 gene (i.e., LDLR)

Sequence-based test 1 variant 19 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 1 variant 19 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 1 variant 19 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 19 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 19 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 19 rsID, if present
(i.e., rs193922570)

Sequence-based test 1 variant 19 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 19 mutation type

☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
1 variant 19

Sequence-based test 1 variant 19 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 19

Sequence-based test 1 variant 19 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 19

Is sequence-based test 1 variant 19 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 19 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 19?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 19 interpretation summary

Sequence-based test 1 variant 20 gene (i.e., LDLR)

Sequence-based test 1 variant 20 transcript ID (i.e., NM_000527.5)

Sequence-based test 1 variant 20 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 1 variant 20 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 1 variant 20 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 1 variant 20 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 1 variant 20 rsID, if present (i.e., rs193922570)

Sequence-based test 1 variant 20 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 1 variant 20 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 1 variant 20

Sequence-based test 1 variant 20 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 1 variant 20

Sequence-based test 1 variant 20 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 1 variant 20

Is sequence-based test 1 variant 20 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 1 variant 20 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 1 variant 20?

- ☐ Yes
- ☐ No

Sequence-based test 1 variant 20 interpretation summary

Upload report for sequence-based test 2

Is variant information obtained directly from testing report for sequence-based test 2?

- ☐ Yes
- ☐ No

Sequence-based test 2 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color Genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for sequence-based test 2

Sequence-based test 2 name

Sequence-based test 2 report date

Is this an amended/reanalysis report for
sequence-based test 2?

- ☐ Yes
- ☐ No

Specify the report date of the amended/reanalysis
report for sequence-based test 2

Was RNA analysis included in sequence-based test 2?

- ☐ Yes
- ☐ No

Sequence-based test 2 type

- ☐ Genome
- ☐ Exome
- ☐ Panel
- ☐ single Gene
- ☐ Single Variant
- ☐ Other

Specify single gene targeted in sequence based test 2

Specify single variant targeted in sequence-based test
2

If sequence-based test 2 is exome or genome, please select one of the following

- ☐ Proband-only
☐ Duo (i.e., Proband and one parent)
☐ Trio (i.e., Proband and both parents)
☐ Quad (i.e., Proband, sibling, and both parents)
☐ Other

Specify the number of genes targeted in the panel for sequence-based test 2

Specify the names of the genes targeted in the panel for sequence-based test 2

Was sequence analysis included for at least one of the genes tested in sequence-based test 2?

- ☐ Yes
☐ No

Was deletion/duplication analysis included for at least one of the genes tested in sequence-based test 2?

- ☐ Yes
☐ No

Proband sample source for sequence-based test 2

- ☐ Blood
☐ Saliva
☐ Buccal swab
☐ Skin biopsy or fibroblast culture
☐ Cancer/tumor sample
☐ Non-malignant surgical sample
☐ Amniotic fluid
☐ Placenta/chorionic villi
☐ Cell free DNA
☐ Other

Specify the type of cancer/tumor sample used in sequence-based test 2

Specify the type of non-malignant surgical sample used in sequence-based test 2

Specify 'Other' proband sample source used in sequence based test 2

Were any variants reported for sequence-based test 2?

- ☐ Yes
☐ No

Number of variants reported in sequence-based test 2

Sequence-based test 2 variant 1 gene (i.e., LDLR)

Sequence-based test 2 variant 1 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 1 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 1 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 1 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 1 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 1 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 1 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 1 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 1

Sequence-based test 2 variant 1 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 1

Sequence-based test 2 variant 1 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 1

Is sequence-based test 2 variant 1 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 1 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 1?

- ☐ Yes
☐ No

Sequence-based test 2 variant 1 interpretation summary

Sequence-based test 2 variant 2 gene (i.e., LDLR)

Sequence-based test 2 variant 2 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 2 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 2 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 2 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 2 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 2 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 2 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 2

Sequence-based test 2 variant 2 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 2

Sequence-based test 2 variant 2 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 2

Is sequence-based test 2 variant 2 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 2 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 2 variant 2? ☐ Yes
☐ No

Sequence-based test 2 variant 2 interpretation summary

Sequence-based test 2 variant 3 gene (i.e., LDLR)

Sequence-based test 2 variant 3 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 2 variant 3 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 2 variant 3 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 3 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 3 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 3 rsID, if present
(i.e., rs193922570)

Sequence-based test 2 variant 3 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 3 mutation type

☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
2 variant 3

Sequence-based test 2 variant 3 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 3

Sequence-based test 2 variant 3 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 3

Is sequence-based test 2 variant 3 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 3 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 3?

- ☐ Yes
- ☐ No

Sequence-based test 2 variant 3 interpretation summary

Sequence-based test 2 variant 4 gene (i.e., LDLR)

Sequence-based test 2 variant 4 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 4 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 4 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 4 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 4 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 4 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 4 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 4

Sequence-based test 2 variant 4 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 4

Sequence-based test 2 variant 4 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 4

Is sequence-based test 2 variant 4 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 4 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 4?

- ☐ Yes
☐ No

Sequence-based test 2 variant 4 interpretation summary

Sequence-based test 2 variant 5 gene (i.e., LDLR)

Sequence-based test 2 variant 5 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 5 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 5 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 5 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 5 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 5 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 5 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 5

Sequence-based test 2 variant 5 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 5

Sequence-based test 2 variant 5 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 5

Is sequence-based test 2 variant 5 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 5 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 5? ☐ Yes ☐ No

Sequence-based test 2 variant 5 interpretation summary

Sequence-based test 2 variant 6 gene (i.e., LDLR)

Sequence-based test 2 variant 6 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 6 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 6 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 6 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 6 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 6 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 6 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 6 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 6

Sequence-based test 2 variant 6 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 6

Sequence-based test 2 variant 6 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 6

Is sequence-based test 2 variant 6 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 6 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 6?

- ☐ Yes
- ☐ No

Sequence-based test 2 variant 6 interpretation summary

Sequence-based test 2 variant 7 gene (i.e., LDLR)

Sequence-based test 2 variant 7 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 7 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 7 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 7 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 7 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 7 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 7 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 7 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 7

Sequence-based test 2 variant 7 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 7

Sequence-based test 2 variant 7 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 7

Is sequence-based test 2 variant 7 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 7 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 7?

- ☐ Yes
☐ No

Sequence-based test 2 variant 7 interpretation summary

Sequence-based test 2 variant 8 gene (i.e., LDLR)

Sequence-based test 2 variant 8 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 8 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 8 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 8 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 8 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 8 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 8 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 8 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 8

Sequence-based test 2 variant 8 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 8

Sequence-based test 2 variant 8 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 8

Is sequence-based test 2 variant 8 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 8 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 2 variant 8?

- ☐ Yes
☐ No

Sequence-based test 2 variant 8 interpretation summary

Sequence-based test 2 variant 9 gene (i.e., LDLR)

Sequence-based test 2 variant 9 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 2 variant 9 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 2 variant 9 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 9 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 9 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 9 rsID, if present
(i.e., rs193922570)

Sequence-based test 2 variant 9 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 9 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
2 variant 9

Sequence-based test 2 variant 9 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 9

Sequence-based test 2 variant 9 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 9

Is sequence-based test 2 variant 9 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 9 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 9?

- ☐ Yes
- ☐ No

Sequence-based test 2 variant 9 interpretation summary

Sequence-based test 2 variant 10 gene (i.e., LDLR)

Sequence-based test 2 variant 10 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 10 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 10 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 10 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 10 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 10 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 10 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 10 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 10

Sequence-based test 2 variant 10 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 10

Sequence-based test 2 variant 10 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 10

Is sequence-based test 2 variant 10 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 10 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 10?

- ☐ Yes
☐ No

Sequence-based test 2 variant 10 interpretation summary

Sequence-based test 2 variant 11 gene (i.e., LDLR)

Sequence-based test 2 variant 11 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 11 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 11 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 11 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 11 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 11 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 11 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 11 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 11

Sequence-based test 2 variant 11 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 11

Sequence-based test 2 variant 11 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 11

Is sequence-based test 2 variant 11 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 11 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 2 variant 11? ☐ Yes
☐ No

Sequence-based test 2 variant 11 interpretation summary

Sequence-based test 2 variant 12 gene (i.e., LDLR)

Sequence-based test 2 variant 12 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 2 variant 12 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 2 variant 12 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 12 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 12 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 12 rsID, if present
(i.e., rs193922570)

Sequence-based test 2 variant 12 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 12 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
2 variant 12

Sequence-based test 2 variant 12 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 12

Sequence-based test 2 variant 12 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 12

Is sequence-based test 2 variant 12 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 12 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 12?

- ☐ Yes
- ☐ No

Sequence-based test 2 variant 12 interpretation summary

Sequence-based test 2 variant 13 gene (i.e., LDLR)

Sequence-based test 2 variant 13 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 13 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 13 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 13 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 13 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 13 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 13 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 13 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 13

Sequence-based test 2 variant 13 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 13

Sequence-based test 2 variant 13 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 13

Is sequence-based test 2 variant 13 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 13 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 13?

- ☐ Yes
☐ No

Sequence-based test 2 variant 13 interpretation summary

Sequence-based test 2 variant 14 gene (i.e., LDLR)

Sequence-based test 2 variant 14 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 14 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 14 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 14 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 14 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 14 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 14 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 14 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 14

Sequence-based test 2 variant 14 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 14

Sequence-based test 2 variant 14 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 14

Is sequence-based test 2 variant 14 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 14 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 2 variant 14? ☐ Yes
☐ No

Sequence-based test 2 variant 14 interpretation summary

Sequence-based test 2 variant 15 gene (i.e., LDLR)

Sequence-based test 2 variant 15 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 2 variant 15 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 2 variant 15 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 15 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 15 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 15 rsID, if present
(i.e., rs193922570)

Sequence-based test 2 variant 15 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 15 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
2 variant 15

Sequence-based test 2 variant 15 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 15

Sequence-based test 2 variant 15 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 15

Is sequence-based test 2 variant 15 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 15 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 15?

- ☐ Yes
- ☐ No

Sequence-based test 2 variant 15 interpretation summary

Sequence-based test 2 variant 16 gene (i.e., LDLR)

Sequence-based test 2 variant 16 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 16 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 16 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 16 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 16 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 16 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 16 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 16 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 16

Sequence-based test 2 variant 16 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 16

Sequence-based test 2 variant 16 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 16

Is sequence-based test 2 variant 16 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 16 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 16?

- ☐ Yes
☐ No

Sequence-based test 2 variant 16 interpretation summary

Sequence-based test 2 variant 17 gene (i.e., LDLR)

Sequence-based test 2 variant 17 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 17 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 17 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 17 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 17 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 17 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 17 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 17 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 17

Sequence-based test 2 variant 17 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 17

Sequence-based test 2 variant 17 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 17

Is sequence-based test 2 variant 17 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 17 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 17? ☐ Yes ☐ No

Sequence-based test 2 variant 17 interpretation summary

Sequence-based test 2 variant 18 gene (i.e., LDLR)

Sequence-based test 2 variant 18 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 18 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 18 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 18 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 18 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 18 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 18 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 18 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 18

Sequence-based test 2 variant 18 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 18

Sequence-based test 2 variant 18 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 18

Is sequence-based test 2 variant 18 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 18 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 2 variant 18?

- ☐ Yes
- ☐ No

Sequence-based test 2 variant 18 interpretation summary

Sequence-based test 2 variant 19 gene (i.e., LDLR)

Sequence-based test 2 variant 19 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 19 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 19 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 19 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 19 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 19 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 19 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 19 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 19

Sequence-based test 2 variant 19 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 19

Sequence-based test 2 variant 19 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 19

Is sequence-based test 2 variant 19 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 2 variant 19 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 2 variant 19?

- ☐ Yes
☐ No

Sequence-based test 2 variant 19 interpretation summary

Sequence-based test 2 variant 20 gene (i.e., LDLR)

Sequence-based test 2 variant 20 transcript ID (i.e., NM_000527.5)

Sequence-based test 2 variant 20 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 2 variant 20 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 2 variant 20 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 2 variant 20 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 2 variant 20 rsID, if present (i.e., rs193922570)

Sequence-based test 2 variant 20 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 2 variant 20 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 2 variant 20

Sequence-based test 2 variant 20 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 2 variant 20

Sequence-based test 2 variant 20 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 2 variant 20

Is sequence-based test 2 variant 20 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 2 variant 20 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 2 variant 20? ☐ Yes
☐ No

Sequence-based test 2 variant 20 interpretation summary

Upload report for sequence-based test 3

Is variant information obtained directly from testing
report for sequence-based test 3? ☐ Yes
☐ No

Sequence-based test 3 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color Genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for sequence-based test 3

Sequence-based test 3 name

Sequence-based test 3 report date

Is this an amended/reanalysis report for
sequence-based test 3? ☐ Yes
☐ No

Specify the report date of the amended/reanalysis
report for sequence-based test 3

Was RNA analysis included in sequence-based test 3? ☐ Yes
☐ No

| | |
|---|---|
| Sequence-based test 3 type | <input type="radio"/> Genome <input type="radio"/> Exome <input type="radio"/> Panel <input type="radio"/> single Gene <input type="radio"/> Single Variant <input type="radio"/> Other |
| Specify single gene targeted in sequence based test 3 | _____ |
| Specify single variant targeted in sequence-based test 3 | _____ |
| If sequence-based test 3 is exome or genome, please select one of the following | <input type="radio"/> Proband-only <input type="radio"/> Duo (i.e., Proband and one parent) <input type="radio"/> Trio (i.e., Proband and both parents) <input type="radio"/> Quad (i.e., Proband, sibling, and both parents) <input type="radio"/> Other |
| Specify the number of genes targeted in the panel for sequence-based test 3 | _____ |
| Specify the names of the genes targeted in the panel for sequence-based test 3 | _____ |
| Was sequence analysis included for at least one of the genes tested in sequence-based test 3? | <input type="radio"/> Yes <input type="radio"/> No |
| Was deletion/duplication analysis included for at least one of the genes tested in sequence-based test 3? | <input type="radio"/> Yes <input type="radio"/> No |
| Proband sample source for sequence-based test 3 | <input type="checkbox"/> Blood <input type="checkbox"/> Saliva <input type="checkbox"/> Buccal swab <input type="checkbox"/> Skin biopsy or fibroblast culture <input type="checkbox"/> Cancer/tumor sample <input type="checkbox"/> Non-malignant surgical sample <input type="checkbox"/> Amniotic fluid <input type="checkbox"/> Placenta/chorionic villi <input type="checkbox"/> Cell free DNA <input type="checkbox"/> Other |
| Specify the type of cancer/tumor sample used in sequence-based test 3 | _____ |
| Specify the type of non-malignant surgical sample used in sequence-based test 3 | _____ |
| Specify 'Other' proband sample source used in sequence based test 3 | _____ |
| Were any variants reported for sequence-based test 3? | <input type="radio"/> Yes <input type="radio"/> No |
| Number of variants reported in sequence-based test 3 | _____ |

Sequence-based test 3 variant 1 gene (i.e., LDLR)

Sequence-based test 3 variant 1 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 1 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 1 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 1 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 1 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 1 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 1 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 1 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 1

Sequence-based test 3 variant 1 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 1

Sequence-based test 3 variant 1 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 1

Is sequence-based test 3 variant 1 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 1 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 1?

- ☐ Yes
☐ No

Sequence-based test 3 variant 1 interpretation summary

Sequence-based test 3 variant 2 gene (i.e., LDLR)

Sequence-based test 3 variant 2 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 2 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 2 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 2 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 2 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 2 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 2 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 2

Sequence-based test 3 variant 2 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 2

Sequence-based test 3 variant 2 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 2

Is sequence-based test 3 variant 2 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 2 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 2? ☐ Yes ☐ No

Sequence-based test 3 variant 2 interpretation summary

Sequence-based test 3 variant 3 gene (i.e., LDLR)

Sequence-based test 3 variant 3 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 3 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 3 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 3 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 3 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 3 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 3 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 3 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 3

Sequence-based test 3 variant 3 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 3

Sequence-based test 3 variant 3 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 3

Is sequence-based test 3 variant 3 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 3 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 3?

- ☐ Yes
- ☐ No

Sequence-based test 3 variant 3 interpretation summary

Sequence-based test 3 variant 4 gene (i.e., LDLR)

Sequence-based test 3 variant 4 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 4 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 4 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 4 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 4 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 4 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 4 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 4

Sequence-based test 3 variant 4 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 4

Sequence-based test 3 variant 4 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 4

Is sequence-based test 3 variant 4 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 4 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 4?

- ☐ Yes
☐ No

Sequence-based test 3 variant 4 interpretation summary

Sequence-based test 3 variant 5 gene (i.e., LDLR)

Sequence-based test 3 variant 5 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 5 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 5 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 5 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 5 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 5 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 5 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 5

Sequence-based test 3 variant 5 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 5

Sequence-based test 3 variant 5 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 5

Is sequence-based test 3 variant 5 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 5 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 5? ☐ Yes ☐ No

Sequence-based test 3 variant 5 interpretation summary

Sequence-based test 3 variant 6 gene (i.e., LDLR)

Sequence-based test 3 variant 6 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 6 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 6 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 6 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 6 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 6 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 6 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 6 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 6

Sequence-based test 3 variant 6 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 6

Sequence-based test 3 variant 6 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 6

Is sequence-based test 3 variant 6 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 6 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 6?

- ☐ Yes
- ☐ No

Sequence-based test 3 variant 6 interpretation summary

Sequence-based test 3 variant 7 gene (i.e., LDLR)

Sequence-based test 3 variant 7 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 7 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 7 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 7 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 7 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 7 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 7 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 7 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 7

Sequence-based test 3 variant 7 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 7

Sequence-based test 3 variant 7 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 7

Is sequence-based test 3 variant 7 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 7 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 7?

- ☐ Yes
☐ No

Sequence-based test 3 variant 7 interpretation summary

Sequence-based test 3 variant 8 gene (i.e., LDLR)

Sequence-based test 3 variant 8 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 8 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 8 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 8 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 8 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 8 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 8 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 8 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 8

Sequence-based test 3 variant 8 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 8

Sequence-based test 3 variant 8 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 8

Is sequence-based test 3 variant 8 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 8 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 8? ☐ Yes ☐ No

Sequence-based test 3 variant 8 interpretation summary

Sequence-based test 3 variant 9 gene (i.e., LDLR)

Sequence-based test 3 variant 9 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 9 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 9 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 9 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 9 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 9 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 9 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 9 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 9

Sequence-based test 3 variant 9 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 9

Sequence-based test 3 variant 9 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 9

Is sequence-based test 3 variant 9 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 9 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 9?

- ☐ Yes
- ☐ No

Sequence-based test 3 variant 9 interpretation summary

Sequence-based test 3 variant 10 gene (i.e., LDLR)

Sequence-based test 3 variant 10 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 10 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 10 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 10 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 10 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 10 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 10 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 10 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 10

Sequence-based test 3 variant 10 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 10

Sequence-based test 3 variant 10 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 10

Is sequence-based test 3 variant 10 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 10 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 10?

- ☐ Yes
☐ No

Sequence-based test 3 variant 10 interpretation summary

Sequence-based test 3 variant 11 gene (i.e., LDLR)

Sequence-based test 3 variant 11 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 11 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 11 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 11 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 11 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 11 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 11 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 11 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 11

Sequence-based test 3 variant 11 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 11

Sequence-based test 3 variant 11 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 11

Is sequence-based test 3 variant 11 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 11 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 3 variant 11? ☐ Yes
☐ No

Sequence-based test 3 variant 11 interpretation summary

Sequence-based test 3 variant 12 gene (i.e., LDLR)

Sequence-based test 3 variant 12 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 3 variant 12 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 3 variant 12 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 12 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 12 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 12 rsID, if present
(i.e., rs193922570)

Sequence-based test 3 variant 12 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 12 mutation type

☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
3 variant 12

Sequence-based test 3 variant 12 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 12

Sequence-based test 3 variant 12 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 12

Is sequence-based test 3 variant 12 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 12 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 12?

- ☐ Yes
- ☐ No

Sequence-based test 3 variant 12 interpretation summary

Sequence-based test 3 variant 13 gene (i.e., LDLR)

Sequence-based test 3 variant 13 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 13 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 13 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 13 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 13 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 13 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 13 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 13 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 13

Sequence-based test 3 variant 13 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 13

Sequence-based test 3 variant 13 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 13

Is sequence-based test 3 variant 13 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 13 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 13?

- ☐ Yes
☐ No

Sequence-based test 3 variant 13 interpretation summary

Sequence-based test 3 variant 14 gene (i.e., LDLR)

Sequence-based test 3 variant 14 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 14 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 14 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 14 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 14 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 14 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 14 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 14 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 14

Sequence-based test 3 variant 14 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 14

Sequence-based test 3 variant 14 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 14

Is sequence-based test 3 variant 14 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 14 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 3 variant 14? ☐ Yes
☐ No

Sequence-based test 3 variant 14 interpretation summary

Sequence-based test 3 variant 15 gene (i.e., LDLR)

Sequence-based test 3 variant 15 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 3 variant 15 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 3 variant 15 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 15 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 15 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 15 rsID, if present
(i.e., rs193922570)

Sequence-based test 3 variant 15 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 15 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
3 variant 15

Sequence-based test 3 variant 15 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 15

Sequence-based test 3 variant 15 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 15

Is sequence-based test 3 variant 15 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 15 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 15?

- ☐ Yes
- ☐ No

Sequence-based test 3 variant 15 interpretation summary

Sequence-based test 3 variant 16 gene (i.e., LDLR)

Sequence-based test 3 variant 16 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 16 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 16 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 16 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 16 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 16 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 16 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 16 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 16

Sequence-based test 3 variant 16 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 16

Sequence-based test 3 variant 16 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 16

Is sequence-based test 3 variant 16 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 16 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 16?

- ☐ Yes
☐ No

Sequence-based test 3 variant 16 interpretation summary

Sequence-based test 3 variant 17 gene (i.e., LDLR)

Sequence-based test 3 variant 17 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 17 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 17 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 17 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 17 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 17 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 17 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 17 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 17

Sequence-based test 3 variant 17 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 17

Sequence-based test 3 variant 17 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 17

Is sequence-based test 3 variant 17 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 17 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 3 variant 17? ☐ Yes
☐ No

Sequence-based test 3 variant 17 interpretation summary

Sequence-based test 3 variant 18 gene (i.e., LDLR)

Sequence-based test 3 variant 18 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 3 variant 18 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 3 variant 18 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 18 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 18 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 18 rsID, if present
(i.e., rs193922570)

Sequence-based test 3 variant 18 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 18 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
3 variant 18

Sequence-based test 3 variant 18 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 18

Sequence-based test 3 variant 18 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 18

Is sequence-based test 3 variant 18 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 18 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 18?

- ☐ Yes
- ☐ No

Sequence-based test 3 variant 18 interpretation summary

Sequence-based test 3 variant 19 gene (i.e., LDLR)

Sequence-based test 3 variant 19 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 19 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 19 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 19 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 19 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 19 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 19 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 19 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 19

Sequence-based test 3 variant 19 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 19

Sequence-based test 3 variant 19 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 19

Is sequence-based test 3 variant 19 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 3 variant 19 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 3 variant 19?

- ☐ Yes
☐ No

Sequence-based test 3 variant 19 interpretation summary

Sequence-based test 3 variant 20 gene (i.e., LDLR)

Sequence-based test 3 variant 20 transcript ID (i.e., NM_000527.5)

Sequence-based test 3 variant 20 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 3 variant 20 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 3 variant 20 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 3 variant 20 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 3 variant 20 rsID, if present (i.e., rs193922570)

Sequence-based test 3 variant 20 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 3 variant 20 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 3 variant 20

Sequence-based test 3 variant 20 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence based test 3 variant 20

Sequence-based test 3 variant 20 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 3 variant 20

Is sequence-based test 3 variant 20 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 3 variant 20 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 3 variant 20? ☐ Yes
☐ No

Sequence-based test 3 variant 20 interpretation summary

Upload report for sequence-based test 4

Is variant information obtained directly from testing report for sequence-based test 4? ☐ Yes
☐ No

Sequence-based test 4 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color Genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for sequence-based test 4

Sequence-based test 4 name

Sequence-based test 4 report date

Is this an amended/reanalysis report for sequence-based test 4? ☐ Yes
☐ No

Specify the report date of the amended/reanalysis report for sequence-based test 4

Was RNA analysis included in sequence-based test 4? ☐ Yes
☐ No

| | |
|---|---|
| Sequence-based test 4 type | <input type="radio"/> Genome <input type="radio"/> Exome <input type="radio"/> Panel <input type="radio"/> single Gene <input type="radio"/> Single Variant <input type="radio"/> Other |
| Specify single gene targeted in sequence based test 4 | <input type="text"/> |
| Specify single variant targeted in sequence-based test 4 | <input type="text"/> |
| If sequence-based test 4 is exome or genome, please select one of the following | <input type="radio"/> Proband-only <input type="radio"/> Duo (i.e., Proband and one parent) <input type="radio"/> Trio (i.e., Proband and both parents) <input type="radio"/> Quad (i.e., Proband, sibling, and both parents) <input type="radio"/> Other |
| Specify the number of genes targeted in the panel for sequence-based test 4 | <input type="text"/> |
| Specify the names of the genes targeted in the panel for sequence-based test 4 | <input type="text"/> |
| Was sequence analysis included for at least one of the genes tested in sequence-based test 4? | <input type="radio"/> Yes <input type="radio"/> No |
| Was deletion/duplication analysis included for at least one of the genes tested in sequence-based test 4? | <input type="radio"/> Yes <input type="radio"/> No |
| Proband sample source for sequence-based test 4 | <input type="checkbox"/> Blood <input type="checkbox"/> Saliva <input type="checkbox"/> Buccal swab <input type="checkbox"/> Skin biopsy or fibroblast culture <input type="checkbox"/> Cancer/tumor sample <input type="checkbox"/> Non-malignant surgical sample <input type="checkbox"/> Amniotic fluid <input type="checkbox"/> Placenta/chorionic villi <input type="checkbox"/> Cell free DNA <input type="checkbox"/> Other |
| Specify the type of cancer/tumor sample used in sequence-based test 4 | <input type="text"/> |
| Specify the type of non-malignant surgical sample used in sequence-based test 4 | <input type="text"/> |
| Specify 'Other' proband sample source used in sequence based test 4 | <input type="text"/> |
| Were any variants reported for sequence-based test 4? | <input type="radio"/> Yes <input type="radio"/> No |
| Number of variants reported in sequence-based test 4 | <input type="text"/> |

Sequence-based test 4 variant 1 gene (i.e., LDLR)

Sequence-based test 4 variant 1 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 1 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 1 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 1 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 1 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 1 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 1 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 1 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 1

Sequence-based test 4 variant 1 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 1

Sequence-based test 4 variant 1 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 1

Is sequence-based test 4 variant 1 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 1 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 1?

- ☐ Yes
☐ No

Sequence-based test 4 variant 1 interpretation summary

Sequence-based test 4 variant 2 gene (i.e., LDLR)

Sequence-based test 4 variant 2 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 2 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 2 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 2 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 2 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 2 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 2 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 2

Sequence-based test 4 variant 2 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 2

Sequence-based test 4 variant 2 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 2

Is sequence-based test 4 variant 2 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 2 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 4 variant 2?

- ☐ Yes
☐ No

Sequence-based test 4 variant 2 interpretation summary

Sequence-based test 4 variant 3 gene (i.e., LDLR)

Sequence-based test 4 variant 3 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 4 variant 3 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 4 variant 3 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 3 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 3 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 3 rsID, if present
(i.e., rs193922570)

Sequence-based test 4 variant 3 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 3 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
4 variant 3

Sequence-based test 4 variant 3 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 3

Sequence-based test 4 variant 3 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 3

Is sequence-based test 4 variant 3 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 3 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 4 variant 3?

- ☐ Yes
- ☐ No

Sequence-based test 4 variant 3 interpretation summary

Sequence-based test 4 variant 4 gene (i.e., LDLR)

Sequence-based test 4 variant 4 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 4 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 4 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 4 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 4 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 4 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 4 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 4

Sequence-based test 4 variant 4 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 4

Sequence-based test 4 variant 4 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 4

Is sequence-based test 4 variant 4 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 4 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 4?

- ☐ Yes
☐ No

Sequence-based test 4 variant 4 interpretation summary

Sequence-based test 4 variant 5 gene (i.e., LDLR)

Sequence-based test 4 variant 5 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 5 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 5 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 5 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 5 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 5 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 5 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 5

Sequence-based test 4 variant 5 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 5

Sequence-based test 4 variant 5 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 5

Is sequence-based test 4 variant 5 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 5 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 4 variant 5? ☐ Yes ☐ No

Sequence-based test 4 variant 5 interpretation summary

Sequence-based test 4 variant 6 gene (i.e., LDLR)

Sequence-based test 4 variant 6 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 6 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 6 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 6 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 6 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 6 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 6 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 6 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 6

Sequence-based test 4 variant 6 zygosity

☐ Heterozygous
☐ Homozygous
☐ Hemizygous
☐ Somatic
☐ Heterozygous, maternally inherited
☐ Heterozygous, paternally inherited
☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
☐ Mitochondrial
☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 6

Sequence-based test 4 variant 6 classification

☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 6

Is sequence-based test 4 variant 6 de novo?

☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 6 mosaic?

☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 6?

☐ Yes
☐ No

Sequence-based test 4 variant 6 interpretation summary

Sequence-based test 4 variant 7 gene (i.e., LDLR)

Sequence-based test 4 variant 7 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 7 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 7 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 7 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 7 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 7 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 7 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 7 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 7

Sequence-based test 4 variant 7 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 7

Sequence-based test 4 variant 7 variant classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 7

Is sequence-based test 4 variant 7 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 7 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 7?

- ☐ Yes
☐ No

Sequence-based test 4 variant 7 interpretation summary

Sequence-based test 4 variant 8 gene (i.e., LDLR)

Sequence-based test 4 variant 8 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 8 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 8 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 8 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 8 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 8 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 8 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 8 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 8

Sequence-based test 4 variant 8 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 8

Sequence-based test 4 variant 8 variant classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 8

Is sequence-based test 4 variant 8 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 8 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 4 variant 8? ☐ Yes
☐ No

Sequence-based test 4 variant 8 interpretation summary

Sequence-based test 4 variant 9 gene (i.e., LDLR)

Sequence-based test 4 variant 9 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 9 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 9 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 9 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 9 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 9 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 9 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 9 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 9

Sequence-based test 4 variant 9 zygosity

☐ Heterozygous
☐ Homozygous
☐ Hemizygous
☐ Somatic
☐ Heterozygous, maternally inherited
☐ Heterozygous, paternally inherited
☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
☐ Mitochondrial
☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 9

Sequence-based test 4 variant 9 variant classification

☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 9

Is sequence-based test 4 variant 9 de novo?

☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 9 mosaic?

☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 9?

☐ Yes
☐ No

Sequence-based test 4 variant 9 interpretation summary

Sequence-based test 4 variant 10 gene (i.e., LDLR)

Sequence-based test 4 variant 10 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 10 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 10 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 10 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 10 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 10 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 10 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 10 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 10

Sequence-based test 4 variant 10 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 10

Sequence-based test 4 variant 10 variant classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 10

Is sequence-based test 4 variant 10 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 10 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 10?

- ☐ Yes
☐ No

Sequence-based test 4 variant 10 interpretation summary

Sequence-based test 4 variant 11 gene (i.e., LDLR)

Sequence-based test 4 variant 11 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 11 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 11 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 11 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 11 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 11 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 11 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 11 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 11

Sequence-based test 4 variant 11 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 11

Sequence-based test 4 variant 11 variant classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 11

Is sequence-based test 4 variant 11 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 11 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 4 variant 11? ☐ Yes ☐ No

Sequence-based test 4 variant 11 interpretation summary

Sequence-based test 4 variant 12 gene (i.e., LDLR)

Sequence-based test 4 variant 12 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 12 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 12 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 12 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 12 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 12 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 12 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 12 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 12

| | |
|--|---|
| Sequence-based test 4 variant 12 zygosity | <input type="radio"/> Heterozygous <input type="radio"/> Homozygous <input type="radio"/> Hemizygous <input type="radio"/> Somatic <input type="radio"/> Heterozygous, maternally inherited <input type="radio"/> Heterozygous, paternally inherited <input type="radio"/> Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified <input type="radio"/> Heterozygous, phased only in cis with other variant in same gene, but paternity not specified <input type="radio"/> Mitochondrial <input type="radio"/> Other |
| Specify 'Other' zygosity for sequence-based test 4 variant 12 | _____ |
| Sequence-based test 4 variant 12 variant classification | <input type="radio"/> Pathogenic <input type="radio"/> Likely pathogenic <input type="radio"/> VUS, lean pathogenic <input type="radio"/> VUS <input type="radio"/> VUS, lean benign <input type="radio"/> Likely benign <input type="radio"/> Benign <input type="radio"/> Pseudodeficiency allele <input type="radio"/> Drug response <input type="radio"/> Risk allele/Benign reportable variant <input type="radio"/> Other <input type="radio"/> Variant classification not specified |
| Specify 'Other' classification for sequence-based test 4 variant 12 | _____ |
| Is sequence-based test 4 variant 12 de novo? | <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown |
| Is sequence-based test 4 variant 12 mosaic? | <input type="radio"/> Yes <input type="radio"/> No |
| Is there another variant on the same gene as sequence-based test 4 variant 12? | <input type="radio"/> Yes <input type="radio"/> No |
| Sequence-based test 4 variant 12 interpretation summary | _____ |
| Sequence-based test 4 variant 13 gene (i.e., LDLR) | _____ |
| Sequence-based test 4 variant 13 transcript ID (i.e., NM_000527.5) | _____ |
| Sequence-based test 4 variant 13 cDNA variant location (i.e., c.2113G>C) | _____ |
| Sequence-based test 4 variant 13 protein variant location (i.e., p.Ala705Pro) | _____ |

Sequence-based test 4 variant 13 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 13 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 13 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 13 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 13 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 13

Sequence-based test 4 variant 13 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 13

Sequence-based test 4 variant 13 variant classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 13

Is sequence-based test 4 variant 13 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 13 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 13?

- ☐ Yes
☐ No

Sequence-based test 4 variant 13 interpretation summary

Sequence-based test 4 variant 14 gene (i.e., LDLR)

Sequence-based test 4 variant 14 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 14 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 14 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 14 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 14 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 14 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 14 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 14 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 14

Sequence-based test 4 variant 14 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 14

Sequence-based test 4 variant 14 variant classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 14

Is sequence-based test 4 variant 14 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 14 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 4 variant 14?

☐ Yes
☐ No

Sequence-based test 4 variant 14 interpretation summary

Sequence-based test 4 variant 15 gene (i.e., LDLR)

Sequence-based test 4 variant 15 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 4 variant 15 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 4 variant 15 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 15 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 15 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 15 rsID, if present
(i.e., rs193922570)

Sequence-based test 4 variant 15 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 15 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
4 variant 15

| | |
|--|---|
| Sequence-based test 4 variant 15 zygosity | <input type="radio"/> Heterozygous <input type="radio"/> Homozygous <input type="radio"/> Hemizygous <input type="radio"/> Somatic <input type="radio"/> Heterozygous, maternally inherited <input type="radio"/> Heterozygous, paternally inherited <input type="radio"/> Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified <input type="radio"/> Heterozygous, phased only in cis with other variant in same gene, but paternity not specified <input type="radio"/> Mitochondrial <input type="radio"/> Other |
| Specify 'Other' zygosity for sequence-based test 4 variant 15 | _____ |
| Sequence-based test 4 variant 15 variant classification | <input type="radio"/> Pathogenic <input type="radio"/> Likely pathogenic <input type="radio"/> VUS, lean pathogenic <input type="radio"/> VUS <input type="radio"/> VUS, lean benign <input type="radio"/> Likely benign <input type="radio"/> Benign <input type="radio"/> Pseudodeficiency allele <input type="radio"/> Drug response <input type="radio"/> Risk allele/Benign reportable variant <input type="radio"/> Other <input type="radio"/> Variant classification not specified |
| Specify 'Other' classification for sequence-based test 4 variant 15 | _____ |
| Is sequence-based test 4 variant 15 de novo? | <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown |
| Is sequence-based test 4 variant 15 mosaic? | <input type="radio"/> Yes <input type="radio"/> No |
| Is there another variant on the same gene as sequence-based test 4 variant 15? | <input type="radio"/> Yes <input type="radio"/> No |
| Sequence-based test 4 variant 15 interpretation summary | _____ |
| Sequence-based test 4 variant 16 gene (i.e., LDLR) | _____ |
| Sequence-based test 4 variant 16 transcript ID (i.e., NM_000527.5) | _____ |
| Sequence-based test 4 variant 16 cDNA variant location (i.e., c.2113G>C) | _____ |
| Sequence-based test 4 variant 16 protein variant location (i.e., p.Ala705Pro) | _____ |

Sequence-based test 4 variant 16 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 16 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 16 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 16 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 16 mutation type

- ☐ Missense
 - ☐ Nonsense
 - ☐ Silent
 - ☐ Inframe indel
 - ☐ Frameshift
 - ☐ Start loss
 - ☐ Stop Loss
 - ☐ Start gain
 - ☐ Promoter
 - ☐ Non-coding
 - ☐ Splice site
 - ☐ 5' UTR
 - ☐ 3' UTR
 - ☐ Deletion
 - ☐ Duplication
 - ☐ Inversion
 - ☐ Other
-

Specify 'Other' mutation type for sequence-based test 4 variant 16

Sequence-based test 4 variant 16 zygosity

- ☐ Heterozygous
 - ☐ Homozygous
 - ☐ Hemizygous
 - ☐ Somatic
 - ☐ Heterozygous, maternally inherited
 - ☐ Heterozygous, paternally inherited
 - ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
 - ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
 - ☐ Mitochondrial
 - ☐ Other
-

Specify 'Other' zygosity for sequence-based test 4 variant 16

Sequence-based test 4 variant 16 variant classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 16

Is sequence-based test 4 variant 16 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 16 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 16?

- ☐ Yes
☐ No

Sequence-based test 4 variant 16 interpretation summary

Sequence-based test 4 variant 17 gene (i.e., LDLR)

Sequence-based test 4 variant 17 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 17 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 17 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 17 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 17 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 17 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 17 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 17 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 17

Sequence-based test 4 variant 17 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 17

Sequence-based test 4 variant 17 variant classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 17

Is sequence-based test 4 variant 17 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 17 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 4 variant 17?

- ☐ Yes
☐ No

Sequence-based test 4 variant 17 interpretation summary

Sequence-based test 4 variant 18 gene (i.e., LDLR)

Sequence-based test 4 variant 18 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 4 variant 18 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 4 variant 18 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 18 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 18 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 18 rsID, if present
(i.e., rs193922570)

Sequence-based test 4 variant 18 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 18 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
4 variant 18

| | |
|--|---|
| Sequence-based test 4 variant 18 zygosity | <input type="radio"/> Heterozygous <input type="radio"/> Homozygous <input type="radio"/> Hemizygous <input type="radio"/> Somatic <input type="radio"/> Heterozygous, maternally inherited <input type="radio"/> Heterozygous, paternally inherited <input type="radio"/> Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified <input type="radio"/> Heterozygous, phased only in cis with other variant in same gene, but paternity not specified <input type="radio"/> Mitochondrial <input type="radio"/> Other |
| Specify 'Other' zygosity for sequence-based test 4 variant 18 | _____ |
| Sequence-based test 4 variant 18 variant classification | <input type="radio"/> Pathogenic <input type="radio"/> Likely pathogenic <input type="radio"/> VUS, lean pathogenic <input type="radio"/> VUS <input type="radio"/> VUS, lean benign <input type="radio"/> Likely benign <input type="radio"/> Benign <input type="radio"/> Pseudodeficiency allele <input type="radio"/> Drug response <input type="radio"/> Risk allele/Benign reportable variant <input type="radio"/> Other <input type="radio"/> Variant classification not specified |
| Specify 'Other' classification for sequence-based test 4 variant 18 | _____ |
| Is sequence-based test 4 variant 18 de novo? | <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown |
| Is sequence-based test 4 variant 18 mosaic? | <input type="radio"/> Yes <input type="radio"/> No |
| Is there another variant on the same gene as sequence-based test 4 variant 18? | <input type="radio"/> Yes <input type="radio"/> No |
| Sequence-based test 4 variant 18 interpretation summary | _____ |
| Sequence-based test 4 variant 19 gene (i.e., LDLR) | _____ |
| Sequence-based test 4 variant 19 transcript ID (i.e., NM_000527.5) | _____ |
| Sequence-based test 4 variant 19 cDNA variant location (i.e., c.2113G>C) | _____ |
| Sequence-based test 4 variant 19 protein variant location (i.e., p.Ala705Pro) | _____ |

Sequence-based test 4 variant 19 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 19 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 19 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 19 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 19 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 19

Sequence-based test 4 variant 19 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 19

Sequence-based test 4 variant 19 variant classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 19

Is sequence-based test 4 variant 19 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 4 variant 19 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 4 variant 19?

- ☐ Yes
☐ No

Sequence-based test 4 variant 19 interpretation summary

Sequence-based test 4 variant 20 gene (i.e., LDLR)

Sequence-based test 4 variant 20 transcript ID (i.e., NM_000527.5)

Sequence-based test 4 variant 20 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 4 variant 20 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 4 variant 20 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 4 variant 20 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 4 variant 20 rsID, if present (i.e., rs193922570)

Sequence-based test 4 variant 20 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 4 variant 20 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 4 variant 20

Sequence-based test 4 variant 20 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 4 variant 20

Sequence-based test 4 variant 20 variant classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 4 variant 20

Is sequence-based test 4 variant 20 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 4 variant 20 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 4 variant 20? ☐ Yes
☐ No

Sequence-based test 4 variant 20 interpretation summary

Upload report for sequence-based test 5

Is variant information obtained directly from testing report for sequence-based test 5? ☐ Yes
☐ No

Sequence-based test 5 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color Genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for sequence-based test 5

Sequence-based test 5 name

Sequence-based test 5 report date

Is this an amended/reanalysis report for sequence-based test 5? ☐ Yes
☐ No

Specify the report date of the amended/reanalysis report for sequence-based test 5

Was RNA analysis included in sequence-based test 5? ☐ Yes
☐ No

| | |
|---|---|
| Sequence-based test 5 type | <input type="radio"/> Genome <input type="radio"/> Exome <input type="radio"/> Panel <input type="radio"/> single Gene <input type="radio"/> Single Variant <input type="radio"/> Other |
| Specify single gene targeted in sequence based test 5 | <input type="text"/> |
| Specify single variant targeted in sequence-based test 5 | <input type="text"/> |
| If sequence-based test 5 is exome or genome, please select one of the following | <input type="radio"/> Proband-only <input type="radio"/> Duo (i.e., Proband and one parent) <input type="radio"/> Trio (i.e., Proband and both parents) <input type="radio"/> Quad (i.e., Proband, sibling, and both parents) <input type="radio"/> Other |
| Specify the number of genes targeted in the panel for sequence-based test 5 | <input type="text"/> |
| Specify the names of the genes targeted in the panel for sequence-based test 5 | <input type="text"/> |
| Was sequence analysis included for at least one of the genes tested in sequence-based test 5? | <input type="radio"/> Yes <input type="radio"/> No |
| Was deletion/duplication analysis included for at least one of the genes tested in sequence-based test 5? | <input type="radio"/> Yes <input type="radio"/> No |
| Proband sample source for sequence-based test 5 | <input type="checkbox"/> Blood <input type="checkbox"/> Saliva <input type="checkbox"/> Buccal swab <input type="checkbox"/> Skin biopsy or fibroblast culture <input type="checkbox"/> Cancer/tumor sample <input type="checkbox"/> Non-malignant surgical sample <input type="checkbox"/> Amniotic fluid <input type="checkbox"/> Placenta/chorionic villi <input type="checkbox"/> Cell free DNA <input type="checkbox"/> Other |
| Specify the type of cancer/tumor sample used in sequence-based test 5 | <input type="text"/> |
| Specify the type of non-malignant surgical sample used in sequence-based test 5 | <input type="text"/> |
| Specify 'Other' proband sample source used in sequence based test 5 | <input type="text"/> |
| Were any variants reported for sequence-based test 5? | <input type="radio"/> Yes <input type="radio"/> No |
| Number of variants reported in sequence-based test 5 | <input type="text"/> |

Sequence-based test 5 variant 1 gene (i.e., LDLR)

Sequence-based test 5 variant 1 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 1 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 1 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 1 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 1 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 1 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 1 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 1 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 1

Sequence-based test 5 variant 1 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 1

Sequence-based test 5 variant 1 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 1

Is sequence-based test 5 variant 1 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 1 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 1?

- ☐ Yes
☐ No

Sequence-based test 5 variant 1 interpretation summary

Sequence-based test 5 variant 2 gene (i.e., LDLR)

Sequence-based test 5 variant 2 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 2 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 2 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 2 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 2 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 2 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 2 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 2

Sequence-based test 5 variant 2 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 2

Sequence-based test 5 variant 2 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 2

Is sequence-based test 5 variant 2 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 2 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 5 variant 2?

☐ Yes
☐ No

Sequence-based test 5 variant 2 interpretation summary

Sequence-based test 5 variant 3 gene (i.e., LDLR)

Sequence-based test 5 variant 3 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 5 variant 3 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 5 variant 3 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 3 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 3 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 3 rsID, if present
(i.e., rs193922570)

Sequence-based test 5 variant 3 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 3 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other

Specify 'Other' mutation type for sequence-based test
5 variant 3

Sequence-based test 5 variant 3 zygosity

☐ Heterozygous
☐ Homozygous
☐ Hemizygous
☐ Somatic
☐ Heterozygous, maternally inherited
☐ Heterozygous, paternally inherited
☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
☐ Mitochondrial
☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 3

Sequence-based test 5 variant 3 classification

☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 3

Is sequence-based test 5 variant 3 de novo?

☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 3 mosaic?

☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 3?

☐ Yes
☐ No

Sequence-based test 5 variant 3 interpretation summary

Sequence-based test 5 variant 4 gene (i.e., LDLR)

Sequence-based test 5 variant 4 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 4 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 4 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 4 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 4 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 4 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 4 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 4

Sequence-based test 5 variant 4 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 4

Sequence-based test 5 variant 4 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 4

Is sequence-based test 5 variant 4 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 4 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 4?

- ☐ Yes
☐ No

Sequence-based test 5 variant 4 interpretation summary

Sequence-based test 5 variant 5 gene (i.e., LDLR)

Sequence-based test 5 variant 5 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 5 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 5 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 5 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 5 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 5 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 5 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 5

Sequence-based test 5 variant 5 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 5

Sequence-based test 5 variant 5 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 5

Is sequence-based test 5 variant 5 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 5 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 5 variant 5? ☐ Yes
☐ No

Sequence-based test 5 variant 5 interpretation summary

Sequence-based test 5 variant 6 gene (i.e., LDLR)

Sequence-based test 5 variant 6 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 5 variant 6 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 5 variant 6 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 6 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 6 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 6 rsID, if present
(i.e., rs193922570)

Sequence-based test 5 variant 6 ClinVar variant ID, if
present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 6 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
5 variant 6

Sequence-based test 5 variant 6 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 6

Sequence-based test 5 variant 6 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 6

Is sequence-based test 5 variant 6 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 6 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 5 variant 6?

- ☐ Yes
- ☐ No

Sequence-based test 5 variant 6 interpretation summary

Sequence-based test 5 variant 7 gene (i.e., LDLR)

Sequence-based test 5 variant 7 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 7 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 7 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 7 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 7 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 7 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 7 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 7 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 7

Sequence-based test 5 variant 7 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 7

Sequence-based test 5 variant 7 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 7

Is sequence-based test 5 variant 7 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 7 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 7?

- ☐ Yes
☐ No

Sequence-based test 5 variant 7 interpretation summary

Sequence-based test 5 variant 8 gene (i.e., LDLR)

Sequence-based test 5 variant 8 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 8 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 8 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 8 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 8 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 8 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 8 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 8 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 8

Sequence-based test 5 variant 8 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 8

Sequence-based test 5 variant 8 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 8

Is sequence-based test 5 variant 8 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 8 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 5 variant 8? ☐ Yes ☐ No

Sequence-based test 5 variant 8 interpretation summary

Sequence-based test 5 variant 9 gene (i.e., LDLR)

Sequence-based test 5 variant 9 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 9 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 9 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 9 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 9 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 9 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 9 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 9 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 9

Sequence-based test 5 variant 9 zygosity

☐ Heterozygous
☐ Homozygous
☐ Hemizygous
☐ Somatic
☐ Heterozygous, maternally inherited
☐ Heterozygous, paternally inherited
☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
☐ Mitochondrial
☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 9

Sequence-based test 5 variant 9 classification

☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 9

Is sequence-based test 5 variant 9 de novo?

☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 9 mosaic?

☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 9?

☐ Yes
☐ No

Sequence-based test 5 variant 9 interpretation summary

Sequence-based test 5 variant 10 gene (i.e., LDLR)

Sequence-based test 5 variant 10 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 10 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 10 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 10 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 10 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 10 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 10 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 10 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 10

Sequence-based test 5 variant 10 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 10

Sequence-based test 5 variant 10 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 10

Is sequence-based test 5 variant 10 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 10 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 10?

- ☐ Yes
☐ No

Sequence-based test 5 variant 10 interpretation summary

Sequence-based test 5 variant 11 gene (i.e., LDLR)

Sequence-based test 5 variant 11 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 11 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 11 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 11 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 11 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 11 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 11 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 11 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 11

Sequence-based test 5 variant 11 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 11

Sequence-based test 5 variant 11 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 11

Is sequence-based test 5 variant 11 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 11 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 5 variant 11?

- ☐ Yes
☐ No
-

Sequence-based test 5 variant 11 interpretation summary

Sequence-based test 5 variant 12 gene (i.e., LDLR)

Sequence-based test 5 variant 12 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 5 variant 12 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 5 variant 12 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 12 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 12 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 12 rsID, if present
(i.e., rs193922570)

Sequence-based test 5 variant 12 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 12 mutation type

- ☐ Missense
☐ Nonsense
☐ Silent
☐ Inframe indel
☐ Frameshift
☐ Start loss
☐ Stop Loss
☐ Start gain
☐ Promoter
☐ Non-coding
☐ Splice site
☐ 5' UTR
☐ 3' UTR
☐ Deletion
☐ Duplication
☐ Inversion
☐ Other
-

Specify 'Other' mutation type for sequence-based test
5 variant 12

Sequence-based test 5 variant 12 zygosity

☐ Heterozygous
☐ Homozygous
☐ Hemizygous
☐ Somatic
☐ Heterozygous, maternally inherited
☐ Heterozygous, paternally inherited
☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
☐ Mitochondrial
☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 12

Sequence-based test 5 variant 12 classification

☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 12

Is sequence-based test 5 variant 12 de novo?

☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 12 mosaic?

☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 12?

☐ Yes
☐ No

Sequence-based test 5 variant 12 interpretation summary

Sequence-based test 5 variant 13 gene (i.e., LDLR)

Sequence-based test 5 variant 13 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 13 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 13 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 13 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 13 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 13 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 13 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 13 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 13

Sequence-based test 5 variant 13 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 13

Sequence-based test 5 variant 13 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 13

Is sequence-based test 5 variant 13 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 13 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 13?

- ☐ Yes
☐ No

Sequence-based test 5 variant 13 interpretation summary

Sequence-based test 5 variant 14 gene (i.e., LDLR)

Sequence-based test 5 variant 14 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 14 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 14 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 14 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 14 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 14 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 14 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 14 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 14

Sequence-based test 5 variant 14 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 14

Sequence-based test 5 variant 14 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 14

Is sequence-based test 5 variant 14 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 14 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 5 variant 14? ☐ Yes
☐ No

Sequence-based test 5 variant 14 interpretation summary

Sequence-based test 5 variant 15 gene (i.e., LDLR)

Sequence-based test 5 variant 15 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 5 variant 15 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 5 variant 15 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 15 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 15 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 15 rsID, if present
(i.e., rs193922570)

Sequence-based test 5 variant 15 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 15 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
5 variant 15

| | |
|---|---|
| Sequence-based test 5 variant 15 zygosity | <input type="radio"/> Heterozygous <input type="radio"/> Homozygous <input type="radio"/> Hemizygous <input type="radio"/> Somatic <input type="radio"/> Heterozygous, maternally inherited <input type="radio"/> Heterozygous, paternally inherited <input type="radio"/> Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified <input type="radio"/> Heterozygous, phased only in cis with other variant in same gene, but paternity not specified <input type="radio"/> Mitochondrial <input type="radio"/> Other |
|---|---|

Specify 'Other' zygosity for sequence-based test 5 variant 15

| | |
|---|---|
| Sequence-based test 5 variant 15 classification | <input type="radio"/> Pathogenic <input type="radio"/> Likely pathogenic <input type="radio"/> VUS, lean pathogenic <input type="radio"/> VUS <input type="radio"/> VUS, lean benign <input type="radio"/> Likely benign <input type="radio"/> Benign <input type="radio"/> Pseudodeficiency allele <input type="radio"/> Drug response <input type="radio"/> Risk allele/Benign reportable variant <input type="radio"/> Other <input type="radio"/> Variant classification not specified |
|---|---|

Specify 'Other' classification for sequence-based test 5 variant 15

| | |
|--|--|
| Is sequence-based test 5 variant 15 de novo? | <input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown |
|--|--|

| | |
|---|---|
| Is sequence-based test 5 variant 15 mosaic? | <input type="radio"/> Yes <input type="radio"/> No |
|---|---|

| | |
|--|---|
| Is there another variant on the same gene as sequence-based test 5 variant 15? | <input type="radio"/> Yes <input type="radio"/> No |
|--|---|

Sequence-based test 5 variant 15 interpretation summary

Sequence-based test 5 variant 16 gene (i.e., LDLR)

Sequence-based test 5 variant 16 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 16 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 16 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 16 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 16 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 16 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 16 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 16 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 16

Sequence-based test 5 variant 16 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 16

Sequence-based test 5 variant 16 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 16

Is sequence-based test 5 variant 16 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 16 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 16?

- ☐ Yes
☐ No

Sequence-based test 5 variant 16 interpretation summary

Sequence-based test 5 variant 17 gene (i.e., LDLR)

Sequence-based test 5 variant 17 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 17 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 17 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 17 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 17 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 17 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 17 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 17 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 17

Sequence-based test 5 variant 17 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 17

Sequence-based test 5 variant 17 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 17

Is sequence-based test 5 variant 17 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 17 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as
sequence-based test 5 variant 17? ☐ Yes
☐ No

Sequence-based test 5 variant 17 interpretation summary

Sequence-based test 5 variant 18 gene (i.e., LDLR)

Sequence-based test 5 variant 18 transcript ID (i.e.,
NM_000527.5)

Sequence-based test 5 variant 18 cDNA variant location
(i.e., c.2113G>C)

Sequence-based test 5 variant 18 protein variant
location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 18 genomic variant
location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 18 deletion information,
if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 18 rsID, if present
(i.e., rs193922570)

Sequence-based test 5 variant 18 ClinVar variant ID,
if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 18 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test
5 variant 18

Sequence-based test 5 variant 18 zygosity

☐ Heterozygous
☐ Homozygous
☐ Hemizygous
☐ Somatic
☐ Heterozygous, maternally inherited
☐ Heterozygous, paternally inherited
☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
☐ Mitochondrial
☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 18

Sequence-based test 5 variant 18 classification

☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 18

Is sequence-based test 5 variant 18 de novo?

☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 18 mosaic?

☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 18?

☐ Yes
☐ No

Sequence-based test 5 variant 18 interpretation summary

Sequence-based test 5 variant 19 gene (i.e., LDLR)

Sequence-based test 5 variant 19 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 19 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 19 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 19 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 19 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 19 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 19 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 19 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 19

Sequence-based test 5 variant 19 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 19

Sequence-based test 5 variant 19 classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 19

Is sequence-based test 5 variant 19 de novo?

- ☐ Yes
☐ No
☐ Unknown

Is sequence-based test 5 variant 19 mosaic?

- ☐ Yes
☐ No

Is there another variant on the same gene as sequence-based test 5 variant 19?

- ☐ Yes
☐ No

Sequence-based test 5 variant 19 interpretation summary

Sequence-based test 5 variant 20 gene (i.e., LDLR)

Sequence-based test 5 variant 20 transcript ID (i.e., NM_000527.5)

Sequence-based test 5 variant 20 cDNA variant location (i.e., c.2113G>C)

Sequence-based test 5 variant 20 protein variant location (i.e., p.Ala705Pro)

Sequence-based test 5 variant 20 genomic variant location, if present (i.e., chr19:11231171 (GRCh37))

Sequence-based test 5 variant 20 deletion information, if applicable (i.e., deletion of exon 3)

Sequence-based test 5 variant 20 rsID, if present (i.e., rs193922570)

Sequence-based test 5 variant 20 ClinVar variant ID, if present (i.e., Variation ID: 36459)

Sequence-based test 5 variant 20 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for sequence-based test 5 variant 20

Sequence-based test 5 variant 20 zygosity

- ☐ Heterozygous
- ☐ Homozygous
- ☐ Hemizygous
- ☐ Somatic
- ☐ Heterozygous, maternally inherited
- ☐ Heterozygous, paternally inherited
- ☐ Heterozygous, phased in trans with at least one other variant in same gene, but paternity not specified
- ☐ Heterozygous, phased only in cis with other variant in same gene, but paternity not specified
- ☐ Mitochondrial
- ☐ Other

Specify 'Other' zygosity for sequence-based test 5 variant 20

Sequence-based test 5 variant 20 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for sequence-based test 5 variant 20

Is sequence-based test 5 variant 20 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is sequence-based test 5 variant 20 mosaic?

- ☐ Yes
- ☐ No

Is there another variant on the same gene as sequence-based test 5 variant 20? ☐ Yes
☐ No

Sequence-based test 5 variant 20 interpretation summary

Number of chromosomal microarray-based genetic tests

Upload report for chromosomal microarray-based test 1

Is variant information obtained directly from testing report for chromosomal microarray-based test 1? ☐ Yes
☐ No

Chromosomal microarray-based test 1 company

☐ Ambry Genetics
☐ ARUP Laboratories
☐ Athena Diagnostics
☐ Baylor Genetics
☐ Blueprint Genetics
☐ Color Genetics
☐ Fulgent Genetics
☐ GeneDx
☐ Greenwood Genetics
☐ Invitae
☐ LabCorp/Integrated Genetics
☐ Laboratory for Molecular Medicine/Partners
☐ Mayo Clinic
☐ Medical Neurogenetics (MNG) Laboratories
☐ Myriad Genetics/Counsyl
☐ Natera Genetics
☐ Prevention Genetics
☐ Quest Diagnostics
☐ Seattle Children's Hospital Genetics Laboratories
☐ University of Chicago Genetic Services
☐ UW Lab Medicine/NCGL/CDL
☐ Variantyx
☐ Veritas Genetics
☐ Other

Specify 'Other' company for chromosomal microarray-based test 1

Chromosomal microarray-based test 1 name

Test report date for chromosomal microarray-based test 1

Is this an amended report for chromosomal microarray-based test 1? ☐ Yes
☐ No

Specify the report date of the amended/reanalysis report for chromosomal microarray-based test 1

Chromosomal microarray-based test 1 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in chromosomal microarray-based test 1

Specify the type of non-malignant surgical sample used in chromosomal microarray-based test 1

Specify 'Other' proband source used in chromosomal microarray-based test 1

Did chromosomal microarray-based test 1 include SNP analysis?

- ☐ Yes
- ☐ No

Number of variants identified in chromosomal microarray-based test 1

ISCN interpretation for chromosomal microarray-based test 1 variant 1(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 1 variant 1 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 1 variant 1

Chromosomal microarray-based test 1 variant 1 mutation size

Chromosomal microarray-based test 1 variant 1 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 1 variant 1 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 1 variant 1

Is chromosomal microarray-based test 1 variant 1 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 1 variant 1 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 1 variant 1 interpretation summary

ISCN interpretation for chromosomal microarray-based test 1 variant 2(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 1 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 1 variant 2

Chromosomal microarray-based test 1 variant 2 mutation size

Chromosomal microarray-based test 1 variant 2 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 1 variant 2 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 1 variant 2

Is chromosomal microarray-based test 1 variant 2 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 1 variant 2 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 1 variant 2 interpretation summary

ISCN interpretation for chromosomal microarray-based test 1 variant 3(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 1 variant 3 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 1 variant 3

Chromosomal microarray-based test 1 variant 3 mutation size

Chromosomal microarray-based test 1 variant 3 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 1 variant 3 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 1 variant 3

Is chromosomal microarray-based test 1 variant 3 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 1 variant 3 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 1 variant 3 interpretation summary

ISCN interpretation for chromosomal microarray-based test 1 variant 4(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 1 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 1 variant 4

Chromosomal microarray-based test 1 variant 4 mutation size

Chromosomal microarray-based test 1 variant 4 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 1 variant 4 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 1 variant 4

Is chromosomal microarray-based test 1 variant 4 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 1 variant 4 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 1 variant 4 interpretation summary

ISCN interpretation for chromosomal microarray-based test 1 variant 5(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 1 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 1 variant 5

Chromosomal microarray-based test 1 variant 5 mutation size

Chromosomal microarray-based test 1 variant 5 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 1 variant 5 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 1 variant 5

Is chromosomal microarray-based test 1 variant 5 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 1 variant 5 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 1 variant 5 interpretation summary

Upload report for chromosomal microarray-based test 2

Is variant information obtained directly from testing report for chromosomal microarray-based test 2?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 2 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color Genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for chromosomal microarray-based test 2

Chromosomal microarray-based test 2 name

Test report date for chromosomal microarray-based test 2

Is this an amended report for chromosomal microarray-based test 2?

☐ Yes

☐ No

Specify the report date of the amended/reanalysis report for chromosomal microarray-based test 2

Chromosomal microarray-based test 2 proband sample source

- ☐ Blood
☐ Saliva
☐ Buccal swab
☐ Skin biopsy or fibroblast culture
☐ Cancer/tumor sample
☐ Non-malignant surgical sample
☐ Amniotic fluid
☐ Placenta/chorionic villi
☐ Cell free DNA
☐ Other

Specify the type of cancer/tumor sample used in chromosomal microarray-based test 2

Specify the type of non-malignant surgical sample used in chromosomal microarray-based test 2

Specify 'Other' proband source used in chromosomal microarray-based test 2

Did chromosomal microarray-based test 2 include SNP analysis?

☐ Yes

☐ No

Number of variants identified in chromosomal microarray-based test 2

ISCN interpretation for chromosomal microarray-based test 2 variant 1(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 2 variant 1 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 2 variant 1

Chromosomal microarray-based test 2 variant 1 mutation size

Chromosomal microarray-based test 2 variant 1 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 2 Variant 1 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 2 variant 1

Is chromosomal microarray-based test 2 variant 1 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 2 variant 1 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 2 variant 1 interpretation summary

ISCN interpretation for chromosomal microarray-based test 2 variant 2(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 2 variant 2 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 2 variant 2

Chromosomal microarray-based test 2 variant 2 mutation size

Chromosomal microarray-based test 2 variant 2 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 2 Variant 2 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 2 variant 2

Is chromosomal microarray-based test 2 variant 2 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 2 variant 2 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 2 variant 2 interpretation summary

ISCN interpretation for chromosomal microarray-based test 2 variant 3(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 2 variant 3 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 2 variant 3

Chromosomal microarray-based test 2 variant 3 mutation size

Chromosomal microarray-based test 2 variant 3 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 2 Variant 3 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 2 variant 3

Is chromosomal microarray-based test 2 variant 3 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 2 variant 3 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 2 variant 3 interpretation summary

ISCN interpretation for chromosomal microarray-based test 2 variant 4(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 2 variant 4 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 2 variant 4

Chromosomal microarray-based test 2 variant 4 mutation size

Chromosomal microarray-based test 2 variant 4 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 2 Variant 4 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 2 variant 4

Is chromosomal microarray-based test 2 variant 4 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 2 variant 4 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 2 variant 4 interpretation summary

ISCN interpretation for chromosomal microarray-based test 2 variant 5(i.e., 46,XY,inv(2),(p13q24))

Chromosomal microarray-based test 2 variant 5 mutation type

- ☐ Missense
- ☐ Nonsense
- ☐ Silent
- ☐ Inframe indel
- ☐ Frameshift
- ☐ Start loss
- ☐ Stop Loss
- ☐ Start gain
- ☐ Promoter
- ☐ Non-coding
- ☐ Splice site
- ☐ 5' UTR
- ☐ 3' UTR
- ☐ Deletion
- ☐ Duplication
- ☐ Inversion
- ☐ Other

Specify 'Other' mutation type for chromosomal microarray-based test 2 variant 5

Chromosomal microarray-based test 2 variant 5 mutation size

Chromosomal microarray-based test 2 variant 5 genomic variant location, if present (i.e., chr2:32432423-42432423, hg19)

Chromosomal microarray-based test 2 Variant 5 classification

- ☐ Pathogenic
- ☐ Likely pathogenic
- ☐ VUS, lean pathogenic
- ☐ VUS
- ☐ VUS, lean benign
- ☐ Likely benign
- ☐ Benign
- ☐ Pseudodeficiency allele
- ☐ Drug response
- ☐ Risk allele/Benign reportable variant
- ☐ Other
- ☐ Variant classification not specified

Specify 'Other' classification for chromosomal microarray-based test 2 variant 5

Is chromosomal microarray-based test 2 variant 5 de novo?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is chromosomal microarray-based test 2 variant 5 mosaic?

- ☐ Yes
- ☐ No

Chromosomal microarray-based test 2 variant 5 interpretation summary

Upload report for the karyotype-based test

Is variant information obtained directly from the testing report for the karyotype-based test?

- ☐ Yes
- ☐ No

Karyotype-based test company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for the karyotype-based test

Karyotype-based test name

Report date for the karyotype-based test

Karyotype-based test proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in the karyotype-based test

Specify the type of non-malignant surgical sample used in the karyotype-based test

Specify 'Other' proband sample source used in the karyotype-based test

Number of cells examined for karyotype-based test 1

Karyotype-based test variant band level/resolution

Karyotype-based test variant ISCN interpretation
(i.e., 46,XY,inv(2),(p13q24))

Karyotype-based test variant classification

- ☐ Pathogenic
☐ Likely pathogenic
☐ VUS, lean pathogenic
☐ VUS
☐ VUS, lean benign
☐ Likely benign
☐ Benign
☐ Pseudodeficiency allele
☐ Drug response
☐ Risk allele/Benign reportable variant
☐ Other
☐ Variant classification not specified
-

Specify 'Other' classification for the karyotype based
test variant

Is the karyotype-based test variant de novo?

- ☐ Yes
☐ No
☐ Unknown
-

Is the karyotype-based test variant mosaic?

- ☐ Yes
☐ No
-

Karyotype-based test variant interpretation summary

Number of repeat expansion-based genetic tests

Upload report for repeat expansion-based test 1

Is variant information obtained directly from testing
report for repeat expansion-based test 1?

- ☐ Yes
☐ No

Repeat expansion-based test 1 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for repeat expansion-based test 1

Repeat expansion-based test 1 name

Repeat expansion-based test 1 report date

Repeat expansion-based test 1 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in repeat expansion-based test 1

Specify the type of non-malignant surgical sample used in repeat expansion-based test 1

Specify 'Other' proband sample source used in repeat expansion-based test 1

Specify the number of genes targeted in panel for repeat expansion-based test 1

Specify the names of the genes targeted in repeat expansion-based test 1

Were any variants reported in repeat expansion-based test 1?

- ☐ Yes
☐ No

Number of variants reported in repeat-expansion based test 1

Repeat expansion-based test 1 variant 1 gene

Number of repeats in repeat expansion-based test 1 variant 1 allele 1

Repeat expansion-based test 1 variant 1 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 1 allele 2

Repeat expansion-based test 1 variant 1 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 1 repeat location information, if applicable

Is repeat expansion-based test 1 variant 1 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 1 variant 1 interpretation summary

Repeat expansion-based test 1 variant 2 gene

Number of repeats in repeat expansion-based test 1 variant 2 allele 1

Repeat expansion-based test 1 variant 2 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1
variant 2 allele 2

Repeat expansion-based test 1 variant 2 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 2 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 2
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 2 interpretation
summary

Repeat expansion-based test 1 variant 3 gene

Number of repeats in repeat expansion-based test 1
variant 3 allele 1

Repeat expansion-based test 1 variant 3 allele 1
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1
variant 3 allele 2

Repeat expansion-based test 1 variant 3 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 3 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 3
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 3 interpretation
summary

Repeat expansion-based test 1 variant 4 gene

Number of repeats in repeat expansion-based test 1 variant 4 allele 1

Repeat expansion-based test 1 variant 4 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 4 allele 2

Repeat expansion-based test 1 variant 4 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 4 repeat location information, if applicable

Is repeat expansion-based test 1 variant 4 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 1 variant 4 interpretation summary

Repeat expansion-based test 1 variant 5 gene

Number of repeats in repeat expansion-based test 1 variant 5 allele 1

Repeat expansion-based test 1 variant 5 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 5 allele 2

Repeat expansion-based test 1 variant 5 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 5 repeat location information, if applicable

Is repeat expansion-based test 1 variant 5 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 5 interpretation summary

Repeat expansion-based test 1 variant 6 gene

Number of repeats in repeat expansion-based test 1 variant 6 allele 1

Repeat expansion-based test 1 variant 6 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1 variant 6 allele 2

Repeat expansion-based test 1 variant 6 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 6 repeat location information, if applicable

Is repeat expansion-based test 1 variant 6 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 6 interpretation summary

Repeat expansion-based test 1 variant 7 gene

Number of repeats in repeat expansion-based test 1 variant 7 allele 1

Repeat expansion-based test 1 variant 7 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 7 allele 2

Repeat expansion-based test 1 variant 7 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 7 repeat location information, if applicable

Is repeat expansion-based test 1 variant 7 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 1 variant 7 interpretation summary

Repeat expansion-based test 1 variant 8 gene

Number of repeats in repeat expansion-based test 1 variant 8 allele 1

Repeat expansion-based test 1 variant 8 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 8 allele 2

Repeat expansion-based test 1 variant 8 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 8 repeat location information, if applicable

Is repeat expansion-based test 1 variant 8 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 8 interpretation summary

Repeat expansion-based test 1 variant 9 gene

Number of repeats in repeat expansion-based test 1 variant 9 allele 1

Repeat expansion-based test 1 variant 9 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1 variant 9 allele 2

Repeat expansion-based test 1 variant 9 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 9 repeat location information, if applicable

Is repeat expansion-based test 1 variant 9 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 9 interpretation summary

Repeat expansion-based test 1 variant 10 gene

Number of repeats in repeat expansion-based test 1 variant 10 allele 1

Repeat expansion-based test 1 variant 10 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1
variant 10 allele 2

Repeat expansion-based test 1 variant 10 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 10 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 10
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 10
interpretation summary

Repeat expansion-based test 1 variant 11 gene

Number of repeats in repeat expansion-based test 1
variant 11 allele 1

Repeat expansion-based test 1 variant 11 allele 1
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1
variant 11 allele 2

Repeat expansion-based test 1 variant 11 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 11 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 11
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 11
interpretation summary

Repeat expansion-based test 1 variant 12 gene

Number of repeats in repeat expansion-based test 1
variant 12 allele 1

Repeat expansion-based test 1 variant 12 allele 1
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 1
variant 12 allele 2

Repeat expansion-based test 1 variant 12 allele 2
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 1 variant 12 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 12
expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 1 variant 12
interpretation summary

Repeat expansion-based test 1 variant 13 gene

Number of repeats in repeat expansion-based test 1
variant 13 allele 1

Repeat expansion-based test 1 variant 13 allele 1
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 1
variant 13 allele 2

Repeat expansion-based test 1 variant 13 allele 2
classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 1 variant 13 repeat location information, if applicable

Is repeat expansion-based test 1 variant 13 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 13 interpretation summary

Repeat expansion-based test 1 variant 14 gene

Number of repeats in repeat expansion-based test 1 variant 14 allele 1

Repeat expansion-based test 1 variant 14 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1 variant 14 allele 2

Repeat expansion-based test 1 variant 14 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 14 repeat location information, if applicable

Is repeat expansion-based test 1 variant 14 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 14 interpretation summary

Repeat expansion-based test 1 variant 15 gene

Number of repeats in repeat expansion-based test 1 variant 15 allele 1

Repeat expansion-based test 1 variant 15 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 15 allele 2

Repeat expansion-based test 1 variant 15 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 15 repeat location information, if applicable

Is repeat expansion-based test 1 variant 15 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 1 variant 15 interpretation summary

Repeat expansion-based test 1 variant 16 gene

Number of repeats in repeat expansion-based test 1 variant 16 allele 1

Repeat expansion-based test 1 variant 16 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1 variant 16 allele 2

Repeat expansion-based test 1 variant 16 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 1 variant 16 repeat location information, if applicable

Is repeat expansion-based test 1 variant 16 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 16 interpretation summary

Repeat expansion-based test 1 variant 17 gene

Number of repeats in repeat expansion-based test 1 variant 17 allele 1

Repeat expansion-based test 1 variant 17 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1 variant 17 allele 2

Repeat expansion-based test 1 variant 17 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 17 repeat location information, if applicable

Is repeat expansion-based test 1 variant 17 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 17 interpretation summary

Repeat expansion-based test 1 variant 18 gene

Number of repeats in repeat expansion-based test 1 variant 18 allele 1

Repeat expansion-based test 1 variant 18 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 1
variant 18 allele 2

Repeat expansion-based test 1 variant 18 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 18 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 18
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 18
interpretation summary

Repeat expansion-based test 1 variant 19 gene

Number of repeats in repeat expansion-based test 1
variant 19 allele 1

Repeat expansion-based test 1 variant 19 allele 1
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 1
variant 19 allele 2

Repeat expansion-based test 1 variant 19 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 1 variant 19 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 19
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 1 variant 19
interpretation summary

Repeat expansion-based test 1 variant 20 gene

Number of repeats in repeat expansion-based test 1
variant 20 allele 1

Repeat expansion-based test 1 variant 20 allele 1
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 1
variant 20 allele 2

Repeat expansion-based test 1 variant 20 allele 2
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 1 variant 20 repeat
location information, if applicable

Is repeat expansion-based test 1 variant 20
expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 1 variant 20
interpretation summary

Upload report for repeat expansion-based test 2

Is variant information obtained directly from testing
report for repeat expansion-based test 2?

- ☐ Yes
- ☐ No

Repeat expansion-based test 2 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for repeat expansion-based test 2

Repeat expansion-based test 2 name

Repeat expansion-based test 2 report date

Repeat expansion-based test 2 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in repeat expansion-based test 2

Specify the type of non-malignant surgical sample used in repeat expansion-based test 2

Specify 'Other' proband sample source used in repeat expansion-based test 2

Specify the number of genes targeted in panel for repeat expansion-based test 2

Specify the names of the genes targeted in repeat expansion-based test 2

Were any variants reported in repeat expansion-based test 2?

- ☐ Yes
☐ No
-

Number of variants reported in repeat-expansion based test 2

Repeat expansion-based test 2 variant 1 gene

Number of repeats in repeat expansion-based test 2 variant 1 allele 1

Repeat expansion-based test 2 variant 1 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 1 allele 2

Repeat expansion-based test 2 variant 1 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 1 repeat location information, if applicable

Is repeat expansion-based test 2 variant 1 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 1 interpretation summary

Repeat expansion-based test 2 variant 2 gene

Number of repeats in repeat expansion-based test 2 variant 2 allele 1

Repeat expansion-based test 2 variant 2 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2
variant 2 allele 2

Repeat expansion-based test 2 variant 2 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 2 repeat
location information, if applicable

Is repeat expansion-based test 2 variant 2
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 2 variant 2 interpretation
summary

Repeat expansion-based test 2 variant 3 gene

Number of repeats in repeat expansion-based test 2
variant 3 allele 1

Repeat expansion-based test 2 variant 3 allele 1
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2
variant 3 allele 2

Repeat expansion-based test 2 variant 3 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 3 repeat
location information, if applicable

Is repeat expansion-based test 2 variant 3
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 2 variant 3 interpretation
summary

Repeat expansion-based test 2 variant 4 gene

Number of repeats in repeat expansion-based test 2 variant 4 allele 1

Repeat expansion-based test 2 variant 4 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 4 allele 2

Repeat expansion-based test 2 variant 4 allele 2 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 2 variant 4 repeat location information, if applicable

Is repeat expansion-based test 2 variant 4 expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 2 variant 4 interpretation summary

Repeat expansion-based test 2 variant 5 gene

Number of repeats in repeat expansion-based test 2 variant 5 allele 1

Repeat expansion-based test 2 variant 5 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 5 allele 2

Repeat expansion-based test 2 variant 5 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 2 variant 5 repeat location information, if applicable

Is repeat expansion-based test 2 variant 5 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 5 interpretation summary

Repeat expansion-based test 2 variant 6 gene

Number of repeats in repeat expansion-based test 2 variant 6 allele 1

Repeat expansion-based test 2 variant 6 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 6 allele 2

Repeat expansion-based test 2 variant 6 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 6 repeat location information, if applicable

Is repeat expansion-based test 2 variant 6 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 6 interpretation summary

Repeat expansion-based test 2 variant 7 gene

Number of repeats in repeat expansion-based test 2 variant 7 allele 1

Repeat expansion-based test 2 variant 7 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 7 allele 2

Repeat expansion-based test 2 variant 7 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 7 repeat location information, if applicable

Is repeat expansion-based test 2 variant 7 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 2 variant 7 interpretation summary

Repeat expansion-based test 2 variant 8 gene

Number of repeats in repeat expansion-based test 2 variant 8 allele 1

Repeat expansion-based test 2 variant 8 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 8 allele 2

Repeat expansion-based test 2 variant 8 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 8 repeat location information, if applicable

Is repeat expansion-based test 2 variant 8 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 8 interpretation summary

Repeat expansion-based test 2 variant 9 gene

Number of repeats in repeat expansion-based test 2 variant 9 allele 1

Repeat expansion-based test 2 variant 9 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 9 allele 2

Repeat expansion-based test 2 variant 9 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 9 repeat location information, if applicable

Is repeat expansion-based test 2 variant 9 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 9 interpretation summary

Repeat expansion-based test 2 variant 10 gene

Number of repeats in repeat expansion-based test 2 variant 10 allele 1

Repeat expansion-based test 2 variant 10 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 10 allele 2

Repeat expansion-based test 2 variant 10 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 10 repeat location information, if applicable

Is repeat expansion-based test 2 variant 10 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 2 variant 10 interpretation summary

Repeat expansion-based test 2 variant 11 gene

Number of repeats in repeat expansion-based test 2 variant 11 allele 1

Repeat expansion-based test 2 variant 11 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 11 allele 2

Repeat expansion-based test 2 variant 11 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 11 repeat location information, if applicable

Is repeat expansion-based test 2 variant 11 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 2 variant 11 interpretation summary

Repeat expansion-based test 2 variant 12 gene

Number of repeats in repeat expansion-based test 2 variant 12 allele 1

Repeat expansion-based test 2 variant 12 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 12 allele 2

Repeat expansion-based test 2 variant 12 allele 2 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 2 variant 12 repeat location information, if applicable

Is repeat expansion-based test 2 variant 12 expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 2 variant 12 interpretation summary

Repeat expansion-based test 2 variant 13 gene

Number of repeats in repeat expansion-based test 2 variant 13 allele 1

Repeat expansion-based test 2 variant 13 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 13 allele 2

Repeat expansion-based test 2 variant 13 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 2 variant 13 repeat location information, if applicable

Is repeat expansion-based test 2 variant 13 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 13 interpretation summary

Repeat expansion-based test 2 variant 14 gene

Number of repeats in repeat expansion-based test 2 variant 14 allele 1

Repeat expansion-based test 2 variant 14 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 14 allele 2

Repeat expansion-based test 2 variant 14 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 14 repeat location information, if applicable

Is repeat expansion-based test 2 variant 14 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 14 interpretation summary

Repeat expansion-based test 2 variant 15 gene

Number of repeats in repeat expansion-based test 2 variant 15 allele 1

Repeat expansion-based test 2 variant 15 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 15 allele 2

Repeat expansion-based test 2 variant 15 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 15 repeat location information, if applicable

Is repeat expansion-based test 2 variant 15 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 2 variant 15 interpretation summary

Repeat expansion-based test 2 variant 16 gene

Number of repeats in repeat expansion-based test 2 variant 16 allele 1

Repeat expansion-based test 2 variant 16 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 16 allele 2

Repeat expansion-based test 2 variant 16 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 2 variant 16 repeat location information, if applicable

Is repeat expansion-based test 2 variant 16 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 16 interpretation summary

Repeat expansion-based test 2 variant 17 gene

Number of repeats in repeat expansion-based test 2 variant 17 allele 1

Repeat expansion-based test 2 variant 17 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 17 allele 2

Repeat expansion-based test 2 variant 17 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 17 repeat location information, if applicable

Is repeat expansion-based test 2 variant 17 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 17 interpretation summary

Repeat expansion-based test 2 variant 18 gene

Number of repeats in repeat expansion-based test 2 variant 18 allele 1

Repeat expansion-based test 2 variant 18 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 2 variant 18 allele 2

Repeat expansion-based test 2 variant 18 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 18 repeat location information, if applicable

Is repeat expansion-based test 2 variant 18 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 18 interpretation summary

Repeat expansion-based test 2 variant 19 gene

Number of repeats in repeat expansion-based test 2 variant 19 allele 1

Repeat expansion-based test 2 variant 19 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 2 variant 19 allele 2

Repeat expansion-based test 2 variant 19 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 2 variant 19 repeat location information, if applicable

Is repeat expansion-based test 2 variant 19 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 2 variant 19 interpretation summary

Repeat expansion-based test 2 variant 20 gene

Number of repeats in repeat expansion-based test 2
variant 20 allele 1

Repeat expansion-based test 2 variant 20 allele 1
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 2
variant 20 allele 2

Repeat expansion-based test 2 variant 20 allele 2
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 2 variant 20 repeat
location information, if applicable

Is repeat expansion-based test 2 variant 20
expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 2 variant 20
interpretation summary

Upload report for repeat expansion-based test 3

Is variant information obtained directly from testing
report for repeat expansion-based test 3?

- ☐ Yes
- ☐ No

Repeat expansion-based test 3 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for repeat expansion-based test 3

Repeat expansion-based test 3 name

Repeat expansion-based test 3 report date

Repeat expansion-based test 3 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in repeat expansion-based test 3

Specify the type of non-malignant surgical sample used in repeat expansion-based test 3

Specify 'Other' proband sample source used in repeat expansion-based test 3

Specify the number of genes targeted in panel for repeat expansion-based test 3

Specify the names of the genes targeted in repeat expansion-based test 3

Were any variants reported in repeat expansion-based test 3?

- ☐ Yes
☐ No

Number of variants reported in repeat-expansion based test 3

Repeat expansion-based test 3 variant 1 gene

Number of repeats in repeat expansion-based test 3 variant 1 allele 1

Repeat expansion-based test 3 variant 1 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 1 allele 2

Repeat expansion-based test 3 variant 1 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 1 repeat location information, if applicable

Is repeat expansion-based test 3 variant 1 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 3 variant 1 interpretation summary

Repeat expansion-based test 3 variant 2 gene

Number of repeats in repeat expansion-based test 3 variant 2 allele 1

Repeat expansion-based test 3 variant 2 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 2 allele 2

Repeat expansion-based test 3 variant 2 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 2 repeat location information, if applicable

Is repeat expansion-based test 3 variant 2 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 3 variant 2 interpretation summary

Repeat expansion-based test 3 variant 3 gene

Number of repeats in repeat expansion-based test 3 variant 3 allele 1

Repeat expansion-based test 3 variant 3 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 3 allele 2

Repeat expansion-based test 3 variant 3 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 3 repeat location information, if applicable

Is repeat expansion-based test 3 variant 3 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 3 variant 3 interpretation summary

Repeat expansion-based test 3 variant 4 gene

Number of repeats in repeat expansion-based test 3 variant 4 allele 1

Repeat expansion-based test 3 variant 4 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 4 allele 2

Repeat expansion-based test 3 variant 4 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 4 repeat location information, if applicable

Is repeat expansion-based test 3 variant 4 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 3 variant 4 interpretation summary

Repeat expansion-based test 3 variant 5 gene

Number of repeats in repeat expansion-based test 3 variant 5 allele 1

Repeat expansion-based test 3 variant 5 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 5 allele 2

Repeat expansion-based test 3 variant 5 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 5 repeat location information, if applicable

Is repeat expansion-based test 3 variant 5 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 5 interpretation summary

Repeat expansion-based test 3 variant 6 gene

Number of repeats in repeat expansion-based test 3 variant 6 allele 1

Repeat expansion-based test 3 variant 6 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 6 allele 2

Repeat expansion-based test 3 variant 6 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 3 variant 6 repeat location information, if applicable

Is repeat expansion-based test 3 variant 6 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 6 interpretation summary

Repeat expansion-based test 3 variant 7 gene

Number of repeats in repeat expansion-based test 3 variant 7 allele 1

Repeat expansion-based test 3 variant 7 allele 1 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 7 allele 2

Repeat expansion-based test 3 variant 7 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 3 variant 7 repeat location information, if applicable

Is repeat expansion-based test 3 variant 7 expanded/contracted from parents?

- ☐ Expanded
- ☐ Contracted
- ☐ Same
- ☐ Unknown

Repeat expansion-based test 3 variant 7 interpretation summary

Repeat expansion-based test 3 variant 8 gene

Number of repeats in repeat expansion-based test 3 variant 8 allele 1

Repeat expansion-based test 3 variant 8 allele 1 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 8 allele 2

Repeat expansion-based test 3 variant 8 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 3 variant 8 repeat location information, if applicable

Is repeat expansion-based test 3 variant 8 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 8 interpretation summary

Repeat expansion-based test 3 variant 9 gene

Number of repeats in repeat expansion-based test 3 variant 9 allele 1

Repeat expansion-based test 3 variant 9 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 9 allele 2

Repeat expansion-based test 3 variant 9 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 3 variant 9 repeat location information, if applicable

Is repeat expansion-based test 3 variant 9 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 9 interpretation summary

Repeat expansion-based test 3 variant 10 gene

Number of repeats in repeat expansion-based test 3 variant 10 allele 1

Repeat expansion-based test 3 variant 10 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 10 allele 2

Repeat expansion-based test 3 variant 10 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 10 repeat location information, if applicable

Is repeat expansion-based test 3 variant 10 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 3 variant 10 interpretation summary

Repeat expansion-based test 3 variant 11 gene

Number of repeats in repeat expansion-based test 3 variant 11 allele 1

Repeat expansion-based test 3 variant 11 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 11 allele 2

Repeat expansion-based test 3 variant 11 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 3 variant 11 repeat location information, if applicable

Is repeat expansion-based test 3 variant 11 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 3 variant 11 interpretation summary

Repeat expansion-based test 3 variant 12 gene

Number of repeats in repeat expansion-based test 3 variant 12 allele 1

Repeat expansion-based test 3 variant 12 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 12 allele 2

Repeat expansion-based test 3 variant 12 allele 2 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 3 variant 12 repeat location information, if applicable

Is repeat expansion-based test 3 variant 12 expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 3 variant 12 interpretation summary

Repeat expansion-based test 3 variant 13 gene

Number of repeats in repeat expansion-based test 3 variant 13 allele 1

Repeat expansion-based test 3 variant 13 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 13 allele 2

Repeat expansion-based test 3 variant 13 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 3 variant 13 repeat location information, if applicable

Is repeat expansion-based test 3 variant 13 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 13 interpretation summary

Repeat expansion-based test 3 variant 14 gene

Number of repeats in repeat expansion-based test 3 variant 14 allele 1

Repeat expansion-based test 3 variant 14 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 14 allele 2

Repeat expansion-based test 3 variant 14 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 3 variant 14 repeat location information, if applicable

Is repeat expansion-based test 3 variant 14 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 14 interpretation summary

Repeat expansion-based test 3 variant 15 gene

Number of repeats in repeat expansion-based test 3 variant 15 allele 1

Repeat expansion-based test 3 variant 15 allele 1 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 15 allele 2

Repeat expansion-based test 3 variant 15 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 3 variant 15 repeat location information, if applicable

Is repeat expansion-based test 3 variant 15 expanded/contracted from parents?

- ☐ Expanded
- ☐ Contracted
- ☐ Same
- ☐ Unknown

Repeat expansion-based test 3 variant 15 interpretation summary

Repeat expansion-based test 3 variant 16 gene

Number of repeats in repeat expansion-based test 3 variant 16 allele 1

Repeat expansion-based test 3 variant 16 allele 1 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 16 allele 2

Repeat expansion-based test 3 variant 16 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 3 variant 16 repeat location information, if applicable

Is repeat expansion-based test 3 variant 16 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 16 interpretation summary

Repeat expansion-based test 3 variant 17 gene

Number of repeats in repeat expansion-based test 3 variant 17 allele 1

Repeat expansion-based test 3 variant 17 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 17 allele 2

Repeat expansion-based test 3 variant 17 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 3 variant 17 repeat location information, if applicable

Is repeat expansion-based test 3 variant 17 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 3 variant 17 interpretation summary

Repeat expansion-based test 3 variant 18 gene

Number of repeats in repeat expansion-based test 3 variant 18 allele 1

Repeat expansion-based test 3 variant 18 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 3 variant 18 allele 2

Repeat expansion-based test 3 variant 18 allele 2 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 3 variant 18 repeat location information, if applicable

Is repeat expansion-based test 3 variant 18 expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 3 variant 18 interpretation summary

Repeat expansion-based test 3 variant 19 gene

Number of repeats in repeat expansion-based test 3 variant 19 allele 1

Repeat expansion-based test 3 variant 19 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 3 variant 19 allele 2

Repeat expansion-based test 3 variant 19 allele 2 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 3 variant 19 repeat location information, if applicable

Is repeat expansion-based test 3 variant 19 expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 3 variant 19 interpretation summary

Repeat expansion-based test 3 variant 20 gene

Number of repeats in repeat expansion-based test 3
variant 20 allele 1

Repeat expansion-based test 3 variant 20 allele 1
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 3
variant 20 allele 2

Repeat expansion-based test 3 variant 20 allele 2
classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 3 variant 20 repeat
location information, if applicable

Is repeat expansion-based test 3 variant 20
expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 3 variant 20
interpretation summary

Upload report for repeat expansion-based test 4

Is variant information obtained directly from testing
report for repeat expansion-based test 4?

- ☐ Yes
- ☐ No

Repeat expansion-based test 4 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for repeat expansion-based test 4

Repeat expansion-based test 4 name

Repeat expansion-based test 4 report date

Repeat expansion-based test 4 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in repeat expansion-based test 4

Specify the type of non-malignant surgical sample used in repeat expansion-based test 4

Specify 'Other' proband sample source used in repeat expansion-based test 4

Specify the number of genes targeted in panel for repeat expansion-based test 4

Specify the names of the genes targeted in repeat expansion-based test 4

Were any variants reported in repeat expansion-based test 4?

- ☐ Yes
☐ No

Number of variants reported in repeat-expansion based test 4

Repeat expansion-based test 4 variant 1 gene

Number of repeats in repeat expansion-based test 4 variant 1 allele 1

Repeat expansion-based test 4 variant 1 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 1 allele 2

Repeat expansion-based test 4 variant 1 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 1 repeat location information, if applicable

Is repeat expansion-based test 4 variant 1 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 1 interpretation summary

Repeat expansion-based test 4 variant 2 gene

Number of repeats in repeat expansion-based test 4 variant 2 allele 1

Repeat expansion-based test 4 variant 2 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 2 allele 2

Repeat expansion-based test 4 variant 2 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 2 repeat location information, if applicable

Is repeat expansion-based test 4 variant 2 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 2 interpretation summary

Repeat expansion-based test 4 variant 3 gene

Number of repeats in repeat expansion-based test 4 variant 3 allele 1

Repeat expansion-based test 4 variant 3 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 3 allele 2

Repeat expansion-based test 4 variant 3 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 3 repeat location information, if applicable

Is repeat expansion-based test 4 variant 3 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 3 interpretation summary

Repeat expansion-based test 4 variant 4 gene

Number of repeats in repeat expansion-based test 4 variant 4 allele 1

Repeat expansion-based test 4 variant 4 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 4 allele 2

Repeat expansion-based test 4 variant 4 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 4 repeat location information, if applicable

Is repeat expansion-based test 4 variant 4 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 4 interpretation summary

Repeat expansion-based test 4 variant 5 gene

Number of repeats in repeat expansion-based test 4 variant 5 allele 1

Repeat expansion-based test 4 variant 5 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 5 allele 2

Repeat expansion-based test 4 variant 5 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 5 repeat location information, if applicable

Is repeat expansion-based test 4 variant 5 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 5 interpretation summary

Repeat expansion-based test 4 variant 6 gene

Number of repeats in repeat expansion-based test 4 variant 6 allele 1

Repeat expansion-based test 4 variant 6 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 6 allele 2

Repeat expansion-based test 4 variant 6 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 6 repeat location information, if applicable

Is repeat expansion-based test 4 variant 6 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 6 interpretation summary

Repeat expansion-based test 4 variant 7 gene

Number of repeats in repeat expansion-based test 4 variant 7 allele 1

Repeat expansion-based test 4 variant 7 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 7 allele 2

Repeat expansion-based test 4 variant 7 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 7 repeat location information, if applicable

Is repeat expansion-based test 4 variant 7 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 7 interpretation summary

Repeat expansion-based test 4 variant 8 gene

Number of repeats in repeat expansion-based test 4 variant 8 allele 1

Repeat expansion-based test 4 variant 8 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 8 allele 2

Repeat expansion-based test 4 variant 8 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 8 repeat location information, if applicable

Is repeat expansion-based test 4 variant 8 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 8 interpretation summary

Repeat expansion-based test 4 variant 9 gene

Number of repeats in repeat expansion-based test 4 variant 9 allele 1

Repeat expansion-based test 4 variant 9 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 9 allele 2

Repeat expansion-based test 4 variant 9 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 9 repeat location information, if applicable

Is repeat expansion-based test 4 variant 9 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 9 interpretation summary

Repeat expansion-based test 4 variant 10 gene

Number of repeats in repeat expansion-based test 4 variant 10 allele 1

Repeat expansion-based test 4 variant 10 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 10 allele 2

Repeat expansion-based test 4 variant 10 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 10 repeat location information, if applicable

Is repeat expansion-based test 4 variant 10 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 10 interpretation summary

Repeat expansion-based test 4 variant 11 gene

Number of repeats in repeat expansion-based test 4 variant 11 allele 1

Repeat expansion-based test 4 variant 11 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 11 allele 2

Repeat expansion-based test 4 variant 11 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 11 repeat location information, if applicable

Is repeat expansion-based test 4 variant 11 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 11 interpretation summary

Repeat expansion-based test 4 variant 12 gene

Number of repeats in repeat expansion-based test 4 variant 12 allele 1

Repeat expansion-based test 4 variant 12 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 12 allele 2

Repeat expansion-based test 4 variant 12 allele 2 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Repeat expansion-based test 4 variant 12 repeat location information, if applicable

Is repeat expansion-based test 4 variant 12 expanded/contracted from parents?

- ☐ Expanded
 - ☐ Contracted
 - ☐ Same
 - ☐ Unknown
-

Repeat expansion-based test 4 variant 12 interpretation summary

Repeat expansion-based test 4 variant 13 gene

Number of repeats in repeat expansion-based test 4 variant 13 allele 1

Repeat expansion-based test 4 variant 13 allele 1 classification

- ☐ Normal allele
 - ☐ Mutable normal allele
 - ☐ Indeterminate/Borderline
 - ☐ Abnormal allele with reduced penetrance
 - ☐ Abnormal allele
 - ☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 13 allele 2

Repeat expansion-based test 4 variant 13 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 4 variant 13 repeat location information, if applicable

Is repeat expansion-based test 4 variant 13 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 13 interpretation summary

Repeat expansion-based test 4 variant 14 gene

Number of repeats in repeat expansion-based test 4 variant 14 allele 1

Repeat expansion-based test 4 variant 14 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 14 allele 2

Repeat expansion-based test 4 variant 14 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 14 repeat location information, if applicable

Is repeat expansion-based test 4 variant 14 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 14 interpretation summary

Repeat expansion-based test 4 variant 15 gene

Number of repeats in repeat expansion-based test 4 variant 15 allele 1

Repeat expansion-based test 4 variant 15 allele 1 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 15 allele 2

Repeat expansion-based test 4 variant 15 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 4 variant 15 repeat location information, if applicable

Is repeat expansion-based test 4 variant 15 expanded/contracted from parents?

- ☐ Expanded
- ☐ Contracted
- ☐ Same
- ☐ Unknown

Repeat expansion-based test 4 variant 15 interpretation summary

Repeat expansion-based test 4 variant 16 gene

Number of repeats in repeat expansion-based test 4 variant 16 allele 1

Repeat expansion-based test 4 variant 16 allele 1 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 16 allele 2

Repeat expansion-based test 4 variant 16 allele 2 classification

- ☐ Normal allele
- ☐ Mutable normal allele
- ☐ Indeterminate/Borderline
- ☐ Abnormal allele with reduced penetrance
- ☐ Abnormal allele
- ☐ Premutation

Repeat expansion-based test 4 variant 16 repeat location information, if applicable

Is repeat expansion-based test 4 variant 16 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 16 interpretation summary

Repeat expansion-based test 4 variant 17 gene

Number of repeats in repeat expansion-based test 4 variant 17 allele 1

Repeat expansion-based test 4 variant 17 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4 variant 17 allele 2

Repeat expansion-based test 4 variant 17 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Repeat expansion-based test 4 variant 17 repeat location information, if applicable

Is repeat expansion-based test 4 variant 17 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown

Repeat expansion-based test 4 variant 17 interpretation summary

Repeat expansion-based test 4 variant 18 gene

Number of repeats in repeat expansion-based test 4 variant 18 allele 1

Repeat expansion-based test 4 variant 18 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation

Number of repeats in repeat expansion-based test 4
variant 18 allele 2

Repeat expansion-based test 4 variant 18 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 18 repeat
location information, if applicable

Is repeat expansion-based test 4 variant 18
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 18
interpretation summary

Repeat expansion-based test 4 variant 19 gene

Number of repeats in repeat expansion-based test 4
variant 19 allele 1

Repeat expansion-based test 4 variant 19 allele 1
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 4
variant 19 allele 2

Repeat expansion-based test 4 variant 19 allele 2
classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 19 repeat
location information, if applicable

Is repeat expansion-based test 4 variant 19
expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 19
interpretation summary

Repeat expansion-based test 4 variant 20 gene

Number of repeats in repeat expansion-based test 4 variant 20 allele 1

Repeat expansion-based test 4 variant 20 allele 1 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Number of repeats in repeat expansion-based test 4 variant 20 allele 2

Repeat expansion-based test 4 variant 20 allele 2 classification

- ☐ Normal allele
☐ Mutable normal allele
☐ Indeterminate/Borderline
☐ Abnormal allele with reduced penetrance
☐ Abnormal allele
☐ Premutation
-

Repeat expansion-based test 4 variant 20 repeat location information, if applicable

Is repeat expansion-based test 4 variant 20 expanded/contracted from parents?

- ☐ Expanded
☐ Contracted
☐ Same
☐ Unknown
-

Repeat expansion-based test 4 variant 20 interpretation summary

Number of other tests

Upload report for Other test 1

Is variant information obtained directly from the report for other test 1?

- ☐ Yes
☐ No

Other test 1 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for other test 1

Other test 1 name

Other test 1 report date

Other test 1 test type

- ☐ Methylation test
- ☐ FISH test
- ☐ MLPA
- ☐ Other

Specify 'Other' test type for other test 1

Other test 1 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in other test 1

Specify the type of non-malignant surgical sample used in other test 1

Specify 'Other' proband sample source for other test 1

Other test 1 variant information

Is other test 1 variant de novo?

- ☐ Yes
☐ No
☐ Unknown

Other test 1 variant interpretation

Upload report for Other test 2

Is variant information obtained directly from the report for other test 2?

- ☐ Yes
☐ No

Other test 2 company

- ☐ Ambry Genetics
☐ ARUP Laboratories
☐ Athena Diagnostics
☐ Baylor Genetics
☐ Blueprint Genetics
☐ Color genetics
☐ Fulgent Genetics
☐ GeneDx
☐ Greenwood Genetics
☐ Invitae
☐ LabCorp/Integrated Genetics
☐ Laboratory for Molecular Medicine/Partners
☐ Mayo Clinic
☐ Medical Neurogenetics (MNG) Laboratories
☐ Myriad Genetics/Counsyl
☐ Natera Genetics
☐ Prevention Genetics
☐ Quest Diagnostics
☐ Seattle Children's Hospital Genetics Laboratories
☐ University of Chicago Genetic Services
☐ UW Lab Medicine/NCGL/CDL
☐ Variantyx
☐ Veritas Genetics
☐ Other

Specify 'Other' company for other test 2

Other test 2 name

Other test 2 report date

Other test 2 test type

- ☐ Methylation test
☐ FISH test
☐ MLPA
☐ Other

Specify 'Other' test type for other test 2

Other test 2 proband sample source

- ☐ Blood
 - ☐ Saliva
 - ☐ Buccal swab
 - ☐ Skin biopsy or fibroblast culture
 - ☐ Cancer/tumor sample
 - ☐ Non-malignant surgical sample
 - ☐ Amniotic fluid
 - ☐ Placenta/chorionic villi
 - ☐ Cell free DNA
 - ☐ Other
-

Specify the type of cancer/tumor sample used in other test 2

Specify the type of non-malignant surgical sample used in other test 2

Specify 'Other' proband sample source for other test 2

Other test 2 variant information

Is other test 2 variant de novo?

- ☐ Yes
 - ☐ No
 - ☐ Unknown
-

Other test 2 variant interpretation

Upload report for Other test 3

Is variant information obtained directly from the report for other test 3?

- ☐ Yes
- ☐ No

Other test 3 company

- ☐ Ambry Genetics
- ☐ ARUP Laboratories
- ☐ Athena Diagnostics
- ☐ Baylor Genetics
- ☐ Blueprint Genetics
- ☐ Color genetics
- ☐ Fulgent Genetics
- ☐ GeneDx
- ☐ Greenwood Genetics
- ☐ Invitae
- ☐ LabCorp/Integrated Genetics
- ☐ Laboratory for Molecular Medicine/Partners
- ☐ Mayo Clinic
- ☐ Medical Neurogenetics (MNG) Laboratories
- ☐ Myriad Genetics/Counsyl
- ☐ Natera Genetics
- ☐ Prevention Genetics
- ☐ Quest Diagnostics
- ☐ Seattle Children's Hospital Genetics Laboratories
- ☐ University of Chicago Genetic Services
- ☐ UW Lab Medicine/NCGL/CDL
- ☐ Variantyx
- ☐ Veritas Genetics
- ☐ Other

Specify 'Other' company for other test 3

Other test 3 name

Other test 3 report date

Other test 3 test type

- ☐ Methylation test
- ☐ FISH test
- ☐ MLPA
- ☐ Other

Specify 'Other' test type for other test 3

Other test 3 proband sample source

- ☐ Blood
- ☐ Saliva
- ☐ Buccal swab
- ☐ Skin biopsy or fibroblast culture
- ☐ Cancer/tumor sample
- ☐ Non-malignant surgical sample
- ☐ Amniotic fluid
- ☐ Placenta/chorionic villi
- ☐ Cell free DNA
- ☐ Other

Specify the type of cancer/tumor sample used in other test 3

Specify the type of non-malignant surgical sample used in other test 3

Specify 'Other' proband sample source for other test 3

Other test 3 variant information

Is other test 3 variant de novo?

- ☐ Yes
☐ No
☐ Unknown

Other test 3 variant interpretation
