

# Demographics/ Background

Record ID

\_\_\_\_\_

## Personal/Protected Health Information

Medical record number

\_\_\_\_\_

Patient last name

\_\_\_\_\_

Date of birth

\_\_\_\_\_

City of residence

\_\_\_\_\_

State of residence

\_\_\_\_\_

Zip code of residence

\_\_\_\_\_

Email address

\_\_\_\_\_

Is this patient related to any other patients in the database?

☐ Yes  
☐ No

Specify relative's REDCap ID number

\_\_\_\_\_

## Demographics/ Background

Date patient entered into database

\_\_\_\_\_

Age when entered into database

\_\_\_\_\_

Is patient deceased?

☐ Yes  
☐ No

Date of death

\_\_\_\_\_

Age at death

\_\_\_\_\_

Sex assigned at birth

☐ Male  
☐ Female  
☐ Undetermined/Unknown/Other

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Gender

- ☐ Woman
- ☐ Man
- ☐ Transgender
- ☐ Non-binary/ non-conforming
- ☐ Prefer not to respond

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Race

- ☐ American Indian or Alaska Native
- ☐ Asian
- ☐ Black or African American
- ☐ Native Hawaiian or Other Pacific Islander
- ☐ White
- ☐ Declined to answer
- ☐ Unknown

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Specify continent of ethnic background

- ☐ Africa
- ☐ Antarctica
- ☐ Asia
- ☐ Australia
- ☐ Europe
- ☐ North America
- ☐ South America

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Specify region of ethnic background

- ☐ Central Africa
- ☐ Central America
- ☐ Central Asian
- ☐ Central European
- ☐ Eastern Africa
- ☐ Eastern Europe
- ☐ Middle Eastern or North African
- ☐ Northern Africa
- ☐ Northern Europe
- ☐ Southern Africa
- ☐ Southern European
- ☐ Western Africa
- ☐ Western European
- ☐ West Indian/Caribbean

Specify country of ethnic background

- ☐ Afghani
- ☐ Albanian
- ☐ Armenian
- ☐ Bangladeshi
- ☐ Bhutanese
- ☐ Bosnian
- ☐ Bulgarian
- ☐ Burmese
- ☐ Cambodian
- ☐ Canadian
- ☐ Chinese
- ☐ Costa Rican
- ☐ Croatian
- ☐ Cuban
- ☐ Eritrean
- ☐ Ethiopian
- ☐ Filipino
- ☐ Guatemalan
- ☐ Honduran
- ☐ Hong Kong
- ☐ Indian (South Asia)
- ☐ Indonesian
- ☐ Iraqi
- ☐ Israeli
- ☐ Japanese
- ☐ Kenyan
- ☐ Korean
- ☐ Laotian/Lao
- ☐ Malay
- ☐ Mexican
- ☐ Mongolian
- ☐ Nepalese/Nepali
- ☐ Nicaraguan
- ☐ Pakistani
- ☐ Panamanian
- ☐ Persian/Iranian
- ☐ Polish
- ☐ Puerto rican
- ☐ Romanian
- ☐ Russian
- ☐ Salvadoran
- ☐ Serbian
- ☐ Slovak
- ☐ Somali
- ☐ Sudanese
- ☐ Syrian
- ☐ Taiwanese
- ☐ Thai
- ☐ Tibetan
- ☐ Turkish
- ☐ Ukrainian
- ☐ United States
- ☐ Vietnamese

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Specify specific ethnic group of ethnic background

- ☐ Alaskan Native
- ☐ Amara/Amhara
- ☐ American Indian
- ☐ Arab
- ☐ Ashkenazi Jew
- ☐ Australian or New Zealander Indigenous
- ☐ Bengali
- ☐ Black or African American
- ☐ Central American Indian/Indigenous
- ☐ Declined to Answer
- ☐ Dinka
- ☐ Hmong
- ☐ Khmer
- ☐ Kurdish/Kurd
- ☐ Mexican American/Chicano
- ☐ Native Hawaiian or Pacific Islander
- ☐ Oromo
- ☐ Pacific Northwest Indian
- ☐ Palestinian
- ☐ Pathan/Pashtun
- ☐ South American Indian/Indigenous
- ☐ Tigre
- ☐ Unavailable or Unknown

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Is patient hispanic or latino?

- ☐ Hispanic or Latino
- ☐ Not Hispanic or Latino
- ☐ Unavailable or unknown
- ☐ Declined to answer

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Medical center of genetics provider who evaluated patient most recently

- ☐ University of Washington
- ☐ Fred Hutchinson Cancer Center
- ☐ Seattle Children's Hospital
- ☐ Other

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Specify 'Other' medical center where genetics provider evaluated patient most recently.

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Provider seen for pre/post-test counseling

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Indication for testing	<input type="checkbox"/> Audiology <input type="checkbox"/> Cancer <input type="checkbox"/> Cardiovascular <input type="checkbox"/> Carrier testing <input type="checkbox"/> Dermatology <input type="checkbox"/> Multiple congenital anomalies <input type="checkbox"/> Endocrinology <input type="checkbox"/> Hematology <input type="checkbox"/> Immunology <input type="checkbox"/> Intellectual disability/Developmental delay <input type="checkbox"/> Metabolic disorder <input type="checkbox"/> Skeletal <input type="checkbox"/> Nephrology <input type="checkbox"/> Neurology <input type="checkbox"/> Ophthalmology <input type="checkbox"/> Pulmonary <input type="checkbox"/> Suspected Syndromes <input type="checkbox"/> Family history of genetic condition <input type="checkbox"/> Other <input type="checkbox"/> Pre-cancerous/benign non-dermatological tumors
Family history of genetic condition is positive for which of the following	<input type="checkbox"/> Audiology <input type="checkbox"/> Cancer <input type="checkbox"/> Cardiovascular <input type="checkbox"/> Dermatology <input type="checkbox"/> Multiple congenital anomalies <input type="checkbox"/> Endocrinology <input type="checkbox"/> Hematology <input type="checkbox"/> Immunology <input type="checkbox"/> Intellectual disability <input type="checkbox"/> Metabolic disorder <input type="checkbox"/> Skeletal <input type="checkbox"/> Nephrology <input type="checkbox"/> Neurology <input type="checkbox"/> Ophthalmology <input type="checkbox"/> Pulmonary <input type="checkbox"/> Syndromic Disease <input type="checkbox"/> Other <input type="checkbox"/> Pre-cancerous/benign non-dermatological tumors
Specify family history of hearing loss	<input type="checkbox"/> Sensorineural hearing loss <input type="checkbox"/> Conductive hearing loss <input type="checkbox"/> Mixed hearing loss
Is there family history of specific frequency hearing loss?	<input type="radio"/> Yes <input type="radio"/> No
Specify family history of specific frequency of hearing loss	<input type="checkbox"/> High frequency hearing loss <input type="checkbox"/> Low frequency hearing loss <input type="checkbox"/> All frequency hearing loss <input type="checkbox"/> Unknown
Is there family history of structural ear abnormalities?	<input type="radio"/> Yes <input type="radio"/> No
Specify family history of structural ear abnormalities	<input type="checkbox"/> Enlarged vestibular aqueduct <input type="checkbox"/> External ear anomalies <input type="checkbox"/> Internal ear anomalies <input type="checkbox"/> Other

Specify laterality for family history of hearing loss

- ☐ Unilateral  
☐ Bilateral

Specify age of onset for family history of hearing loss

- ☐ Congenital  
☐ Childhood  
☐ Adult  
☐ Non-progressive  
☐ Progressive  
☐ Unknown

Is family history of hearing loss syndromic?

- ☐ Yes  
☐ No

	1st degree relative	2nd degree relative	3rd degree relative	More distant relative	Unknown relationship
Adrenocortical	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Bladder	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Brain	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Bone	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Breast	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Cervical	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Colorectal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Endometrial	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Esophageal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Gastrointestinal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Kidney	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Leukemia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Liver/Intrahepatic Bile Duct	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Lung	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Lymphoma	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Melanoma	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Multiple myeloma	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Non-melanoma skin cancer	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Oropharyngeal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Ovarian	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Pancreatic	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Paraganglioma	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Parathyroid	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Peritoneal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Pheochromocytoma	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Prostate	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Sarcoma	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Stomach	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Testicular	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Thymus	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Thyroid	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Upper urothelial	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Uterine	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Other	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Specify 'Other' family history of cancer

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Specify family history of pre-cancerous/benign non-dermatological tumors

- ☐ Breast lesions
- ☐ Colon polyps
- ☐ Chondrioma
- ☐ Desmoid tumor/Agressive fibromatosis
- ☐ Fibroids/Leiomyoma/Myoma
- ☐ Hemangioma
- ☐ Lipoma
- ☐ Lymphangioma
- ☐ Meningioma
- ☐ Osteoma
- ☐ Papiloma
- ☐ Pituitary adenoma
- ☐ Plexiform neurofibroma/Neurofibroma
- ☐ Schwannoma
- ☐ Other

Specify family history of breast lesions

- ☐ Ductal carcinoma in situ (DCIS)
- ☐ Lobular carcinoma in situ (LCIS)
- ☐ Other/Unspecified

Specify family history of Colon Polyps

- ☐ Juvenile/Hamartoma polyps
- ☐ Adenoma polyps
- ☐ Sessile polyps
- ☐ Unspecified/Other

Specify 'Other' family history of pre-cancerous/benign non-dermatological tumors

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Specify family history of cardiovascular disorders

- ☐ Arrhythmia
- ☐ Cardiomyopathy
- ☐ Congenital heart anomaly
- ☐ Connective tissue disease/Aneurysm/Dissection
- ☐ Hyperlipidemia
- ☐ Vascular/ lymphatic malformation
- ☐ Other

Specify family history of arrhythmia

- ☐ Sudden cardiac arrest/Ventricular fibrillation
- ☐ Atrial fibrillation/Supra-ventricular tachycardia
- ☐ Ventricular tachycardia/Premature ventricular contraction
- ☐ Cardiac conduction disease

Specify family history of cardiac conduction disease	<input type="checkbox"/> Sick sinus disease/AV block/Bradycardia <input type="checkbox"/> Long QT syndrome <input type="checkbox"/> Short QT syndrome <input type="checkbox"/> Brugada syndrome <input type="checkbox"/> Wolff-Parkinson-White (WPW) <input type="checkbox"/> Other
Specify 'Other' family history of cardiac conduction disease	_____
Specify family history of cardiomyopathy	<input type="checkbox"/> Hypertrophic cardiomyopathy <input type="checkbox"/> Dilated cardiomyopathy <input type="checkbox"/> Restrictive cardiomyopathy <input type="checkbox"/> Arrhythmogenic cardiomyopathy <input type="checkbox"/> Left ventricular non-compaction cardiomyopathy <input type="checkbox"/> Stress induced cardiomyopathy/reduced ejection fraction
Specify family history of congenital heart anomaly	<input type="checkbox"/> Syndromic <input type="checkbox"/> Non-syndromic
Specify family history of connective tissue disease/aneurysm/dissection	<input type="checkbox"/> Aortic Aneurysm <input type="checkbox"/> Aortic dissection <input type="checkbox"/> Aneurysm of other portions of the arterial tree <input type="checkbox"/> Dissection of other portions of the arterial tree <input type="checkbox"/> Marfan syndrome <input type="checkbox"/> Loeys-Dietz syndrome <input type="checkbox"/> Ehlers-Danlos Syndrome <input type="checkbox"/> Non-syndromic familial thoracic aortic aneurysm and dissection (TAAD) <input type="checkbox"/> Other
Specify family history of aortic aneurysm	<input type="checkbox"/> Root <input type="checkbox"/> Ascending <input type="checkbox"/> Descending <input type="checkbox"/> Abdominal
Specify family history of aortic dissection	<input type="checkbox"/> Root <input type="checkbox"/> Ascending <input type="checkbox"/> Descending <input type="checkbox"/> Abdominal
Specify family history of aneurysms of other portions of the arterial tree	<input type="checkbox"/> Cerebral artery <input type="checkbox"/> Cervical artery <input type="checkbox"/> Coronary artery <input type="checkbox"/> Iliac/Mesenteric artery <input type="checkbox"/> Renal artery <input type="checkbox"/> Other
Specify 'Other' family history of aneurysms of other portions of the arterial tree	_____
Specify family history of dissection of other portions of the arterial tree	<input type="checkbox"/> Cerebral artery <input type="checkbox"/> Cervical artery <input type="checkbox"/> Coronary artery <input type="checkbox"/> Iliac/Mesenteric artery <input type="checkbox"/> Renal artery <input type="checkbox"/> Other



Specify 'Other' family history of dissection of other portions of the arterial tree

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Specify family history of Ehlers-Danlos syndrome

- ☐ Classical
- ☐ Hypermobile/Type III
- ☐ Vascular EDS/Type IV
- ☐ Arthrochalasis/Type VII
- ☐ Other

Specify 'Other' family history of Ehlers-Danlos syndrome

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Specify 'Other' family history of connective tissue disease/aneurysm/dissection

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Specify family history of hyperlipidemia

- ☐ Hypercholesterolemia
- ☐ Hypertriglyceridemia

Specify family history of vascular/lymphatic malformations

- ☐ Arteriovenous malformations
- ☐ Venous malformations
- ☐ Capillary malformations
- ☐ Lymphatic malformations
- ☐ Telangiectasias

Specify 'Other' family history of cardiovascular disorders.

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Specify family history of dermatological disorders

- ☐ Hyperkeratosis or scaling skin
- ☐ Epidermolysis bullosa
- ☐ Abnormal skin/epidermal components
- ☐ Abnormal skin pigment
- ☐ Cutis laxa
- ☐ Collagen disorders
- ☐ Dermal vascular malformations
- ☐ Lipomas
- ☐ Lipodystrophy
- ☐ Lymphedema
- ☐ Urticaria
- ☐ Abnormal skin growth
- ☐ Photosensitivity
- ☐ Premature aging/Progeroid disorder
- ☐ Eczema/Abnormal rash
- ☐ Other

Specify family history of hyperkeratosis or scaling skin

- ☐ Ichthyosis
- ☐ Erythrokeratoderma
- ☐ Acrokeratoderma
- ☐ Palmoplantar keratoderma
- ☐ Other

Specify 'Other' family history of hyperkeratosis or scaling skin

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Specify family history of epidermolysis bullosa

- ☐ Simplex
- ☐ Junctional
- ☐ Dystrophic
- ☐ Kindler syndrome
- ☐ Unspecified
- ☐ Other

Specify 'Other' family history of epidermolysis bullosa

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Specify family history of abnormal skin/epidermal components

- ☐ Alopecia
- ☐ Hair shaft abnormality
- ☐ Nail disorders
- ☐ Ectodermal dysplasia
- ☐ Hirsutism
- ☐ Sweat gland disorders
- ☐ Sebaceous gland disorders
- ☐ Other

Specify 'Other' family history of abnormal skin/epidermal components

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Specify family history of abnormal skin pigment

- ☐ Hypopigmentation
- ☐ Patterned hyperpigmentation
- ☐ Cafe-au-lait macules
- ☐ Suspected neurofibromatosis
- ☐ Suspected tuberous sclerosis complex
- ☐ Other

Specify family history of hypopigmentation

- ☐ Albinism
- ☐ Oculocutaneous albinism
- ☐ Piebaldism
- ☐ Vitiligo
- ☐ Waardenburg syndrome
- ☐ Other

Specify 'Other' family history of hypopigmentation

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Specify 'Other' family history of abnormal skin pigment

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Specify family history of collagen disorders

- ☐ Skin hyperextensibility
- ☐ Skin fragility
- ☐ Poor wound healing

Specify family history of dermal vascular malformations

- ☐ Port-wine stain
- ☐ Telangiectasia
- ☐ Angiokeratoma
- ☐ Venous malformations
- ☐ Lymphovenous malformation
- ☐ Other

Specify 'Other' family history of dermal vascular malformations

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Specify family history of abnormal skin growth

- ☐ Atypical nevus/nevi
- ☐ Fibrofolliculomas
- ☐ Steatocystoma
- ☐ Angiofibroma
- ☐ Other

Specify 'Other' family history of abnormal skin growth

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Specify family history of photosensitivity

- ☐ Suspected porphyria  
☐ Xeroderma pigmentosum  
☐ Other

Specify 'Other' family history of photosensitivity

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Specify 'Other' family history of dermatological disorder

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Specify family history of endocrine disorders

- ☐ Adrenal  
☐ Calcium/phosphate homeostasis  
☐ Pituitary  
☐ Thyroid  
☐ Glucose/Insulin  
☐ Sex hormones  
☐ Multiple endocrine organ autoimmune disease  
☐ Obesity  
☐ Other

Specify family history of adrenal disorder

- ☐ Cushing syndrome  
☐ Adrenal insufficiency  
☐ Hyperaldosteronism  
☐ Other

Specify 'Other' family history of adrenal disorder

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Specify family history of calcium/phosphate homeostasis disorder

- ☐ Hypercalcemia  
☐ Hyperparathyroidism  
☐ Hypoparathyroidism/Pseudohypoparathyroidism  
☐ Hypophosphatemia  
☐ Low alkaline phosphatase  
☐ Other

Specify 'Other' family history of calcium/phosphate homeostasis disorder

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Specify family history of pituitary disorder

- ☐ Combined pituitary hormone deficiency  
☐ Isolated growth hormone deficiency  
☐ Pituitary adenoma  
☐ Other

Specify 'Other' family history of pituitary disorder

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Specify family history of thyroid disorder

- ☐ Hypothyroidism  
☐ Hyperthyroidism  
☐ Other

Specify 'Other' family history of thyroid disorder

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Specify family history of glucose/insulin disorder

- ☐ Diabetes  
☐ Hyperinsulinemia  
☐ Other

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Specify 'Other' family history of glucose/insulin disorder

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Specify family history of sex hormone disorder

- ☐ Androgen insensitivity
- ☐ Hypogonadism
- ☐ Precocious puberty
- ☐ Premature ovarian insufficiency
- ☐ Male infertility
- ☐ Female infertility
- ☐ Other

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Specify 'Other' family history of sex hormone disorder

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Specify 'Other' family history of endocrine disorder

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Specify family history of hematologic disorders

- ☐ Bone marrow failure
- ☐ Clotting disorder
- ☐ Platelet disorder
- ☐ Red blood cell disorder
- ☐ White blood cell disorder

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Specify family history of bone marrow failure

- ☐ Fanconi anemia
- ☐ Short telomere syndrome
- ☐ Other

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Specify 'Other' family history of bone marrow failure

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Specify family history of clotting disorder

- ☐ Frequent bleeding
- ☐ Unusual venous blood clotting
- ☐ Unusual arterial blood clotting
- ☐ Antithrombin III deficiency
- ☐ Factor V Leiden
- ☐ Protein C deficiency
- ☐ Protein S deficiency
- ☐ Thrombophilia
- ☐ Other

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Specify 'Other' family history of clotting disorder

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Specify family history of platelet disorder

- ☐ Platelet dysfunction
- ☐ Thrombocythemia
- ☐ Thrombocytopenia
- ☐ Other

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Specify 'Other' family history of platelet disorder

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Specify family history of red blood cell disorders

- ☐ Aplastic anemia
- ☐ Erythrocytosis
- ☐ Globin disorders
- ☐ Glycolysis-related hemolytic anemia
- ☐ Megaloblastic anemia
- ☐ Red blood cell membrane disorders
- ☐ Sideroblastic anemia
- ☐ Thrombotic microangiopathies/Atypical hemolytic uremic syndrome
- ☐ Other anemia
- ☐ Other red blood cell disorder

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Specify family history of globin disorder

- ☐ Beta-thalassemia
- ☐ Alpha-thalassemia
- ☐ Sickle-cell anemia
- ☐ Abnormal serum electrophoresis
- ☐ Other

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Specify 'Other' family history of globin disorder

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Specify family history of glycolysis-related hemolytic anemia

- ☐ G6PD deficiency
- ☐ Hexokinase deficiency
- ☐ Pyruvate kinase (PK) deficiency
- ☐ Other

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Specify 'Other' family history of glycolysis-related hemolytic anemia

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Specify family history of red blood cell membrane disorder

- ☐ Spherocytosis
- ☐ Elliptocytosis
- ☐ Stomatocytosis
- ☐ Other

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Specify 'Other' family history of red blood cell membrane disorders

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Specify 'Other' family history of anemia

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Specify 'Other' family history of red blood cell disorders

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Specify family history of white blood cell disorder

- ☐ Neutropenia
- ☐ Severe combined immunodeficiency
- ☐ Other

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Specify 'Other' family history of white blood cell disorder

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Specify family history of immune system disorders	<input type="checkbox"/> Immunodeficiencies affecting cellular and humoral immunity <input type="checkbox"/> Combined immune deficiencies with associated or syndromic features <input type="checkbox"/> Predominantly antibody deficiency <input type="checkbox"/> Disease of immune dysregulation <input type="checkbox"/> Congenital defects of phagocytic number or function <input type="checkbox"/> Defects of intrinsic and innate immunity <input type="checkbox"/> Autoinflammatory disorder <input type="checkbox"/> Complement deficiency <input type="checkbox"/> Recurrent fever <input type="checkbox"/> Phenocopies of inborn errors of immunity <input type="checkbox"/> Multiple sclerosis/Neuroinflammatory disorders
Specify family history of immunodeficiencies affecting cellular and humoral immunity	<input type="checkbox"/> T Cell (-) B Cell (+) severe combined immune deficiencies (SCID) <input type="checkbox"/> T Cell (-) B Cell (-) severe combined immune deficiencies (SCID) <input type="checkbox"/> Combined immune deficiencies (CID)
Specify family history of combined immune deficiencies with associated or syndromic features	<input type="checkbox"/> Immunodeficiency with congenital thrombocytopenia <input type="checkbox"/> DNA repair defects other than those classified by Immunodeficiencies affecting cellular and humoral immunity <input type="checkbox"/> Thymic defects with additional congenital anomalies <input type="checkbox"/> Immuno-osseous dysplasias <input type="checkbox"/> Hyper IgE syndrome (HIES) <input type="checkbox"/> Other
Specify family history of predominantly antibody deficiencies	<input type="checkbox"/> Severe reduction in all serum immunoglobulin isotypes with profoundly decreased or absent B cells, agammaglobulinemia <input type="checkbox"/> Severe reduction in at least 2 serum immunoglobulin isotypes with normal or low number of B cells, CVID phenotype <input type="checkbox"/> Severe reduction in serum IgG and IgA with normal/elevated IgM and normal numbers of B cells, hyper IgM <input type="checkbox"/> Isotype, light chain, or functional deficiencies with generally normal numbers of B cells
Specify family history of disease of immune dysregulation	<input type="checkbox"/> Familial hemophagocytic lymphohistiocytosis (FHL syndromes) <input type="checkbox"/> FHL syndromes with hypopigmentation <input type="checkbox"/> Regulatory T cell defects <input type="checkbox"/> Autoimmunity with or without lymphoproliferation <input type="checkbox"/> Immune dysregulation with colitis <input type="checkbox"/> Autoimmune lymphoproliferative syndrome (ALPS, Canale-Smith syndrome) <input type="checkbox"/> Susceptibility to EBV and lymphoproliferative conditions
Specify family history of congenital defects of phagocytic number or function	<input type="checkbox"/> Congenital neutropenias <input type="checkbox"/> Defects of motility <input type="checkbox"/> Defects of respiratory burst <input type="checkbox"/> Other non-lymphoid defects

Specify family history of defects in intrinsic and innate immunity

- ☐ Mendelian susceptibility to mycobacterial disease (MSMID)
- ☐ Epidermodysplasia verruciformis (HPV)
- ☐ Predisposition to severe viral infection
- ☐ Herpes simplex encephalitis (HSE)
- ☐ Predisposition to invasive fungal diseases
- ☐ Predisposition to mucocutaneous candidiasis
- ☐ TLR signaling pathway deficiency with bacterial susceptibility
- ☐ Other inborn errors of immunity related to non-hematopoietic tissues
- ☐ Other inborn errors of immunity related to leukocytes

Specify family history of autoinflammatory disorders

- ☐ Type 1 interferonopathies affecting the inflammasome
- ☐ Non-inflammasome-related conditions

Specify family history of intellectual disability/developmental delay

- ☐ Intellectual disability
- ☐ Developmental delay
- ☐ Autism
- ☐ Cerebral palsy
- ☐ Other

Specify family history of developmental delay

- ☐ Gross motor
- ☐ Fine motor
- ☐ Speech
- ☐ Global developmental delay

Specify 'Other' intellectual disability/developmental delay

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Specify family history of metabolic disorders

- ☐ Abnormal amino acid Levels
- ☐ Abnormal carnitine levels
- ☐ Abnormal carbohydrate levels
- ☐ Abnormal copper metabolism
- ☐ Abnormal fatty acid oxidation
- ☐ Abnormal glycosylation
- ☐ Abnormal iron metabolism
- ☐ Abnormal liver enzymes
- ☐ Abnormal organic acid metabolism
- ☐ Abnormal urea cycle/Hyperammonemia
- ☐ Abnormal very long chain fatty acids
- ☐ Abnormal zinc metabolism
- ☐ Alpha-1-antitrypsin deficiency
- ☐ Glycogen storage disease
- ☐ Lysosomal storage disorder
- ☐ Macrocytic anemia/Abnormal B12/folate/homocysteine
- ☐ Mitochondrial disorder
- ☐ Porphyrria
- ☐ Rhabdomyolysis
- ☐ Other

Specify family history of abnormal carbohydrate levels

- ☐ Hypoglycemia
- ☐ Glycosuria
- ☐ Other

Specify 'Other' family history of abnormal carbohydrate levels

---

Specify family history of abnormal copper metabolism

- ☐ Menkes disease
- ☐ Wilson disease
- ☐ Low ceruloplasmin/serum copper/urine copper
- ☐ Elevated ceruloplasmin/serum copper/urine copper
- ☐ Other

Specify 'Other' family history of abnormal copper metabolism

\_\_\_\_\_

Specify family history of abnormal iron metabolism

- ☐ Hemochromatosis/Elevated ferritin
- ☐ Aceroplasminemia
- ☐ Other

Specify 'Other' family history of abnormal iron metabolism

\_\_\_\_\_

Specify family history of abnormal very long chain fatty acids

- ☐ X-linked adrenoleukodystrophy
- ☐ Refsum disease
- ☐ Zellweger spectrum disorder
- ☐ Rhizomelic chondrodysplasia punctata
- ☐ Other

Specify 'Other' family history of abnormal very long chain fatty acids

\_\_\_\_\_

Specify family history of lysosomal storage disorder

- ☐ Gaucher disease
- ☐ Fabry disease
- ☐ Pompe disease
- ☐ Morquio Syndrome
- ☐ Metachromatic leukodystrophy
- ☐ Other

Specify 'Other' family history of lysosomal storage disorder

\_\_\_\_\_

Specify family history of macrocytic anemia/abnormal B12/folate/homocysteine

- ☐ Methylmalonic aciduria
- ☐ Homocystinuria
- ☐ Other

Specify 'Other' family history of macrocytic anemia/abnormal B12/Folate/Homocysteine

\_\_\_\_\_

Specify family history of mitochondrial disorder

- ☐ Abnormal mitochondrial testing
- ☐ Chronic progressive external ophthalmoplegia
- ☐ Lactic acidemia
- ☐ Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)
- ☐ Severe gastrointestinal dysmotility
- ☐ Unexplained fatigue and exercise intolerance
- ☐ Other

Specify 'Other' family history of mitochondrial disorder

\_\_\_\_\_

Specify 'Other' family history of metabolic disorders

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Specify family history of kidney disorders	<input type="checkbox"/> Congenital anomalies of the kidney and urinary tract <input type="checkbox"/> Cystic disease <input type="checkbox"/> Glomerular and tubular disease <input type="checkbox"/> Tubulopathies <input type="checkbox"/> Nephrolithiasis
Specify family history of congenital anomalies of kidney and urinary tract (CAKUT)	<input type="checkbox"/> Structural defects of kidney <input type="checkbox"/> Structural defects of urinary tract <input type="checkbox"/> Syndromic CAKUT
Specify family history of cystic disease	<input type="checkbox"/> Renal cysts <input type="checkbox"/> Liver cysts <input type="checkbox"/> Nephronophthisis
Specify family history of glomerular tubular disease	<input type="checkbox"/> Atypical hemolytic uremic syndrome (aHUS) <input type="checkbox"/> Biopsy-suggestive genetic condition <input type="checkbox"/> Chronic kidney disease <input type="checkbox"/> Microscopic hematuria <input type="checkbox"/> Nephritic syndrome <input type="checkbox"/> Nephrotic syndrome <input type="checkbox"/> Other
Specify family history of chronic kidney disease	<input type="checkbox"/> Childhood-onset chronic kidney disease (CKD)/end stage renal disease (ESRD) <input type="checkbox"/> Adult-onset chronic kidney disease (CKD)/end stage renal disease (ESRD)
Specify "Other" family history of glomerular tubular disease	_____
Specify family history of tubulopathies	<input type="checkbox"/> Chronic hypokalemia with metabolic alkalosis (Bartter/Gittelman syndrome) <input type="checkbox"/> Chronic hypokalemia with metabolic acidosis/Renal tubular acidosis <input type="checkbox"/> Other
Specify "Other" family history of tubulopathies	_____
Specify family history of nephrolithiasis	<input type="checkbox"/> Calcium oxalate stones/Hyperoxaluria <input type="checkbox"/> Calcium phosphate stones/Renal tubular acidosis <input type="checkbox"/> Uric acid renal stones/Hyperuricemia <input type="checkbox"/> Cystine renal stones/Cystinuria
Specify family history of neurological/neuromuscular disorders	<input type="checkbox"/> Neuromuscular/Muscular <input type="checkbox"/> Brain iron accumulation <input type="checkbox"/> Movement disorders <input type="checkbox"/> Charcot Marie Tooth (CMT)/Neuropathy <input type="checkbox"/> Cerebrovascular disease <input type="checkbox"/> Dementia <input type="checkbox"/> Neurocutaneous <input type="checkbox"/> Epilepsy <input type="checkbox"/> Leukodystrophies/Abnormal white matter <input type="checkbox"/> Encephalopathy <input type="checkbox"/> Headache <input type="checkbox"/> Other

Specify family history of neuromuscular/muscular disease

- ☐ Myopathy/Muscle weakness
- ☐ Congenital myasthenia
- ☐ Myotonia
- ☐ Periodic paralysis
- ☐ Muscular dystrophy
- ☐ Motor neuron disease
- ☐ Hypotonia

Specify family history of myopathy/muscle weakness

- ☐ Proximal muscle weakness
- ☐ Distal muscle weakness
- ☐ Bulbar muscle weakness
- ☐ Elevated creatinine kinase

Specify family history of muscular dystrophy

- ☐ Becker
- ☐ Congenital
- ☐ Distal
- ☐ Duchene
- ☐ Emery-Dreifuss
- ☐ Facioscapulohumeral (FSHD)
- ☐ Limb-girdle
- ☐ Myotonic
- ☐ Oculopharyngeal
- ☐ Other

Specify 'Other' family history of muscular dystrophy

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Specify family history of motor neuron disease

- ☐ Amyotrophic lateral sclerosis (ALS)
- ☐ Progressive bulbar palsy (PBP)
- ☐ Pseudobulbar palsy
- ☐ Primary lateral sclerosis (PLS)
- ☐ Progressive muscular atrophy (PMA)
- ☐ Spinal muscular atrophy (SMA)
- ☐ Monomelic amyotrophy (MMA)
- ☐ Other

Specify 'Other' family history of motor neuron disease

---

Specify family history of movement disorder

- ☐ Ataxia
- ☐ Generalized dystonia
- ☐ Hereditary spastic paraplegia (HSP)
- ☐ Huntington's disease/Chorea
- ☐ Parkinsonism
- ☐ Paroxysmal kinesigenic dyskinesia
- ☐ Other

Specify 'Other' family history of movement disorder

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Specify family history of Charcot Marie Tooth (CMT)/neuropathy

- ☐ Autonomic neuropathy
- ☐ Motor neuropathy
- ☐ Small fiber neuropathy
- ☐ Sensory neuropathy
- ☐ Mixed
- ☐ Other

Specify 'Other' family history of Charcot Marie Tooth (CMT)/Neuropathy

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Specify family history of cerebrovascular disease

- ☐ Ischemic stroke
- ☐ Hemorrhagic stroke
- ☐ Cerebral aneurysm
- ☐ Cerebral artery amyloidosis
- ☐ CADASIL
- ☐ Moya moya
- ☐ Other

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Specify 'Other' family history of cerebrovascular disease

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Specify family history of dementia

- ☐ Alzheimer dementia
- ☐ Fronto-temporal dementia
- ☐ Vascular dementia
- ☐ Idiopathic basal ganglia calcification
- ☐ Creutzfeldt-Jakob disease (CJD)/Other prion disease
- ☐ Other

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Specify 'Other' family history of dementia

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Specify family history of neurocutaneous disorder

- ☐ Neurofibromatosis
- ☐ Tuberous sclerosis
- ☐ Schwannomatosis
- ☐ Other

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Specify 'Other' family history of neurocutaneous disorder

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Specify family history of epilepsy

- ☐ Congenital onset
- ☐ Infant onset
- ☐ Childhood onset
- ☐ Adult onset
- ☐ Focal epilepsy
- ☐ Generalized epilepsy
- ☐ Absence seizures
- ☐ Epileptic encephalopathy
- ☐ West syndrome
- ☐ Structural brain anomalies present
- ☐ Other

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Specify 'Other' family history of epilepsy

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Specify family history of headache

- ☐ Migraine
- ☐ Unspecified headache type

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Specify 'Other' family history of neurological/neuromuscular disorders

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Specify family history of ophthalmic disorders

- ☐ Retinal dystrophy
- ☐ Vitreoretinopathies
- ☐ Ocular dysgenesis
- ☐ Optic nerve abnormalities/Septo-optic dysplasia
- ☐ Glaucoma
- ☐ Corneal dystrophy
- ☐ Cataracts
- ☐ Oculocutaneous albinism
- ☐ Other

---

Specify family history of retinal dystrophy

- ☐ Cone-rod dystrophy
- ☐ Rod-cone dystrophy
- ☐ Retinitis Pigmentosa
- ☐ Macular dystrophy
- ☐ Leber congenital amaurosis
- ☐ Syndromic retinal dystrophy
- ☐ Other

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Specify 'Other' family history of retinal dystrophy

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Specify family history of ocular dysgenesis

- ☐ Microphthalmia
- ☐ Anophthalmia
- ☐ Coloboma
- ☐ Anterior segment dysgenesis
- ☐ Other

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Specify 'Other' family history ocular dysgenesis

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Specify 'Other' family history of ophthalmic disorders

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Specify family history pulmonary disorders

- ☐ Bronchiectasis
- ☐ Central hypoventilation
- ☐ Chronic obstructive pulmonary disease (COPD)
- ☐ Cystic fibrosis
- ☐ Pneumothorax
- ☐ Pulmonary alveolar proteinosis
- ☐ Pulmonary cysts
- ☐ Pulmonary Fibrosis/Interstitial lung disease
- ☐ Other

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Specify 'Other' family history of pulmonary disorders

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Specify family history of skeletal disorders

- ☐ VACTERL/VATER association
- ☐ Congenital diaphragmatic hernia
- ☐ Craniofacial abnormalities
- ☐ Short stature
- ☐ Bone disorders
- ☐ Limb deformities
- ☐ Pterygium
- ☐ Tooth disorders
- ☐ Other

Specify family history of craniofacial abnormalities

- ☐ Blepharophimosis
- ☐ Cleft lip/palate
- ☐ Cleidocranial dysplasia
- ☐ Craniofacial microsomia
- ☐ Craniosynostosis
- ☐ Treacher Collins syndrome
- ☐ Jaw cysts
- ☐ Lip pits
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Parry Romberg atrophy
- ☐ Robin sequence
- ☐ Nose abnormality
- ☐ Other

Specify 'Other' family history of craniofacial abnormalities

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Specify family history of craniosynostosis

- ☐ Crouzon syndrome
- ☐ Apert syndrome
- ☐ Muenke syndrome
- ☐ Pfeiffer syndrome
- ☐ Saethre-Chotzen syndrome
- ☐ Other

Specify 'Other' family history of craniosynostosis

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Specify family history of short stature?

- ☐ Achondroplasia
- ☐ Campomelic dysplasia
- ☐ Chondrodysplasia punctata
- ☐ Silver-Russell syndrome
- ☐ Hypochondroplasia
- ☐ Pseudoachondroplasia
- ☐ Short rib skeletal dysplasia
- ☐ Thanatophoric dysplasia
- ☐ Other disproportionate short stature
- ☐ Other proportionate short stature
- ☐ Other

Specify 'Other' family history of short stature

---

Specify family history of bone disorders

- ☐ Osteogenesis imperfecta (OI)
- ☐ Exostoses
- ☐ Frequent bone fractures
- ☐ Osteoarthritis
- ☐ Osteopetrosis
- ☐ Osteoporosis
- ☐ Paget disease
- ☐ Other

Specify 'Other' family history of bone disorders

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Specify family history of limb deformities

- ☐ Club foot
- ☐ Oligodactyly
- ☐ Polydactyly
- ☐ Reduction defect
- ☐ Split hand/foot
- ☐ Syndactyly
- ☐ Thumb abnormality
- ☐ Other

Does family history of thumb abnormality include radial abnormality?

- ☐ Yes
- ☐ No
- ☐ Unknown

Specify 'Other' family history of limb deformities

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Specify family history of tooth disorders

- ☐ Amelogenesis imperfecta
- ☐ Single front incisor
- ☐ Tooth agenesis
- ☐ Other

Specify 'Other' family history of tooth disorder

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Specify 'Other' family history of skeletal disorders

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Is family history of skeletal disorders syndromic?

- ☐ Yes
- ☐ No

Specify family history of syndromic disease

- ☐ Neurofibromatosis type 1
- ☐ Neurofibromatosis type 2
- ☐ Trisomy 21
- ☐ Noonan syndrome
- ☐ Turners syndrome
- ☐ Tuberous Sclerosis
- ☐ Birt-Hogg-Dube syndrome
- ☐ DiGeorge syndrome
- ☐ Lynch syndrome
- ☐ Li-Fraumeni syndrome
- ☐ Von Hippel-Lindau
- ☐ Hereditary breast ovarian cancer syndrome (HBOC)

Specify 'Other' family history

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Specify hearing loss

- ☐ Sensorineural hearing loss
- ☐ Conductive hearing loss
- ☐ Mixed hearing loss

Is there specific frequency hearing loss?

- ☐ Yes
- ☐ No

Specify specific frequency hearing loss

- ☐ High frequency hearing loss
- ☐ Low frequency hearing loss
- ☐ All frequency hearing loss

---

Are there any structural ear abnormalities?

- ☐ Yes  
☐ No

---

Specify structural ear abnormalities

- ☐ Enlarged vestibular aqueduct  
☐ External ear anomalies  
☐ Internal ear anomalies  
☐ Other

---

Specify laterality of hearing loss

- ☐ Unilateral  
☐ Bilateral

---

Specify age of onset of hearing loss

- ☐ Congenital  
☐ Childhood  
☐ Adult  
☐ Non-progressive  
☐ Progressive  
☐ Unknown

---

Is hearing loss syndromic?

- ☐ Yes  
☐ No

---

Specify cancers

- ☐ Personal history  
☐ Active cancer

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Specify personal history of cancers

- ☐ Adrenocortical
- ☐ Bladder
- ☐ Brain
- ☐ Bone
- ☐ Breast
- ☐ Cervical
- ☐ Colorectal
- ☐ Endometrial
- ☐ Esophageal
- ☐ Gastrointestinal
- ☐ Kidney
- ☐ Leukemia
- ☐ Liver/Intrahepatic Bile Duct
- ☐ Lung
- ☐ Lymphoma
- ☐ Melanoma
- ☐ Multiple Myeloma
- ☐ Non-melanoma skin cancer
- ☐ Oropharyngeal
- ☐ Ovarian
- ☐ Pancreatic
- ☐ Paraganglioma
- ☐ Parathyroid
- ☐ Peritoneal
- ☐ Pheochromocytoma
- ☐ Prostate
- ☐ Sarcoma
- ☐ Stomach
- ☐ Testicular
- ☐ Thymus
- ☐ Thyroid
- ☐ Upper urothelial
- ☐ Uterine
- ☐ Other

---

Specify 'Other' personal history of cancers

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Specify active cancers

- ☐ Adrenocortical
- ☐ Bladder
- ☐ Brain
- ☐ Bone
- ☐ Breast
- ☐ Cervical
- ☐ Colorectal
- ☐ Endometrial
- ☐ Esophageal
- ☐ Gastrointestinal
- ☐ Kidney
- ☐ Leukemia
- ☐ Liver/Intrahepatic Bile Duct
- ☐ Lung
- ☐ Lymphoma
- ☐ Melanoma
- ☐ Multiple Myeloma
- ☐ Non-melanoma skin cancer
- ☐ Oropharyngeal
- ☐ Ovarian
- ☐ Pancreatic
- ☐ Paraganglioma
- ☐ Parathyroid
- ☐ Peritoneal
- ☐ Pheochromocytoma
- ☐ Prostate
- ☐ Sarcoma
- ☐ Stomach
- ☐ Testicular
- ☐ Thymus
- ☐ Thyroid
- ☐ Upper urothelial
- ☐ Uterine
- ☐ Other

Specify 'Other' active cancer

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Specify pre-cancerous/benign non-dermatological tumors

- ☐ Breast lesions
- ☐ Colon polyps
- ☐ Chondrioma
- ☐ Desmoid tumor/Agressive fibromatosis
- ☐ Fibroids/Leiomyoma/Myoma
- ☐ Hemangioma
- ☐ Lipoma
- ☐ Lymphangioma
- ☐ Meningioma
- ☐ Osteoma
- ☐ Papiloma
- ☐ Pituitary adenoma
- ☐ Plexiform neurofibroma/Neurofibroma
- ☐ Schwannoma
- ☐ Other

Specify breast lesions

- ☐ Ductal carcinoma in situ (DCIS)
- ☐ Lobular carcinoma in situ (LCIS)
- ☐ Other/Unspecified

Specify colon polyps

- ☐ Juvenile/Hamartoma polyps
- ☐ Adenoma polyps
- ☐ Sessile polyps
- ☐ Unspecified/Other

Specify 'Other' family history of pre-cancerous/benign non-dermatological tumors

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Specify cardiovascular disorders

- ☐ Arrhythmia
- ☐ Cardiomyopathy
- ☐ Congenital heart anomaly
- ☐ Connective tissue disease/Aneurysm/Dissection
- ☐ Hyperlipidemia
- ☐ Vascular/Lymphatic malformation
- ☐ Other

Specify arrhythmia

- ☐ Sudden cardiac arrest/Ventricular fibrillation
- ☐ Atrial fibrillation/Supra-ventricular tachycardia
- ☐ Ventricular tachycardia/Premature ventricular contraction
- ☐ Cardiac conduction disease

Specify cardiac conduction disease

- ☐ Sick sinus disease/AV block/Bradycardia
- ☐ Long QT syndrome
- ☐ Short QT syndrome
- ☐ Brugada syndrome
- ☐ Wolff-Parkinson-White syndrome(WPW)
- ☐ Other

Specify 'Other' cardiac conduction disease

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Specify cardiomyopathy

- ☐ Hypertrophic cardiomyopathy
- ☐ Dilated cardiomyopathy
- ☐ Restrictive cardiomyopathy
- ☐ Arrhythmogenic cardiomyopathy
- ☐ Left ventricular non-compaction cardiomyopathy
- ☐ Stress induced cardiomyopathy/Reduced ejection fraction

Specify congenital heart anomaly

- ☐ Syndromic
- ☐ Non-syndromic

Specify connective tissue disease/aneurysm/dissection

- ☐ Aortic aneurysm
- ☐ Aortic dissection
- ☐ Aneurysm of other portions of the arterial tree
- ☐ Dissection of other portions of the arterial tree
- ☐ Clinical concern for Marfan syndrome
- ☐ Clinical concern for Loeys-Dietz syndrome
- ☐ Clinical concern for Ehlers-Danlos Syndrome
- ☐ Clinical concern for non-syndromic familial thoracic aortic aneurysm and dissection (TAAD)
- ☐ Other

Specify aortic aneurysm

- ☐ Root
- ☐ Ascending
- ☐ Descending
- ☐ Abdominal

Specify aortic dissection

- ☐ Root
- ☐ Ascending
- ☐ Descending
- ☐ Abdominal

---

Specify aneurysms of other portions of the arterial tree

- ☐ Cerebral artery
- ☐ Cervical artery
- ☐ Coronary artery
- ☐ Iliac/Mesenteric artery
- ☐ Renal artery
- ☐ Other

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Specify 'Other' aneurysms of other portions of the arterial tree

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Specify dissection of other portions of the arterial tree

- ☐ Cerebral artery
- ☐ Cervical artery
- ☐ Coronary artery
- ☐ Iliac/Mesenteric artery
- ☐ Renal artery
- ☐ Other

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Specify 'Other' dissection of other portions of the arterial tree

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Specify clinical concern for Ehlers-Danlos syndrome

- ☐ Classical
- ☐ Hypermobile/Type III
- ☐ Vascular EDS/Type IV
- ☐ Arthrochalasis/Type VII
- ☐ Other

---

Specify 'Other' clinical concern for Ehlers-Danlos syndrome

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Specify 'Other' connective tissue disease/aneurysm/dissection

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Specify hyperlipidemia

- ☐ Hypercholesterolemia
- ☐ Hypertriglyceridemia

---

Specify vascular/lymphatic malformations

- ☐ Arteriovenous malformations
- ☐ Venous malformations
- ☐ Capillary malformations
- ☐ Lymphatic malformations
- ☐ Telangiectasias

---

Specify 'Other' cardiovascular disorder

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Specify dermatological disorders

- ☐ Hyperkeratosis or scaling skin
- ☐ Epidermolysis bullosa
- ☐ Abnormal skin/epidermal components
- ☐ Abnormal skin pigment
- ☐ Cutis laxa
- ☐ Collagen disorders
- ☐ Dermal vascular malformations
- ☐ Lipomas
- ☐ Lipodystrophy
- ☐ Lymphedema
- ☐ Urticaria
- ☐ Abnormal skin growth
- ☐ Photosensitivity
- ☐ Premature aging/Progeroid disorder
- ☐ Eczema/Abnormal rash
- ☐ Other

Specify hyperkeratosis or scaling skin

- ☐ Ichthyosis
- ☐ Erythrokeratoderma
- ☐ Acrokeratoderma
- ☐ Palmoplantar keratoderma
- ☐ Other

Specify 'Other' hyperkeratosis or scaling skin

\_\_\_\_\_

Specify Epidermolysis bullosa

- ☐ Simplex
- ☐ Junctional
- ☐ Dystrophic
- ☐ Suspected Kindler syndrome
- ☐ Unspecified
- ☐ Other

Specify 'Other' epidermolysis bullosa

\_\_\_\_\_

Specify abnormal skin/epidermal components

- ☐ Alopecia
- ☐ Hair shaft abnormality
- ☐ Nail disorders
- ☐ Ectodermal dysplasia
- ☐ Hirsutism
- ☐ Sweat gland disorders
- ☐ Sebaceous gland disorders
- ☐ Other

Specify 'Other' abnormal skin/epidermal components

\_\_\_\_\_

Specify abnormal skin pigment

- ☐ Hypopigmentation
- ☐ Patterned hyperpigmentation
- ☐ Cafe-au-lait macules
- ☐ Suspected neurofibromatosis
- ☐ Suspected tuberous sclerosis complex
- ☐ Other

---

Specify hypopigmentation

- ☐ Albinism
- ☐ Oculocutaneous albinism
- ☐ Piebaldism
- ☐ Vitiligo
- ☐ Waardenburg syndrome
- ☐ Other

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Specify 'Other' hypopigmentation

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Specify 'Other' abnormal skin pigment

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Specify collagen disorders

- ☐ Skin hyperextensibility
- ☐ Skin fragility
- ☐ Poor wound healing

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Specify dermal vascular malformations

- ☐ Port-wine stain
- ☐ Telangiectasia
- ☐ Angiokeratoma
- ☐ Venous malformations
- ☐ Lymphovenous malformations
- ☐ Other

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Specify 'Other' dermal vascular malformations

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Specify abnormal skin growth

- ☐ Atypical nevus/nevi
- ☐ Fibrofolliculomas
- ☐ Steatocystoma
- ☐ Angiofibroma
- ☐ Other

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Specify 'Other' abnormal skin growth

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Specify photosensitivity

- ☐ Suspected porphyria
- ☐ Xeroderma pigmentosum
- ☐ Other

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Specify 'Other' photosensitivity

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Specify 'Other' dermatological disorder

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Specify endocrine disorders

- ☐ Adrenal
- ☐ Calcium/phosphate homeostasis
- ☐ Pituitary
- ☐ Thyroid
- ☐ Glucose/Insulin
- ☐ Sex hormones
- ☐ Multiple endocrine organ autoimmune disease
- ☐ Obesity
- ☐ Other

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Specify adrenal disorder

- ☐ Cushing syndrome
  - ☐ Adrenal insufficiency
  - ☐ Hyperaldosteronism
  - ☐ Other
- 

Specify 'other' adrenal disorder

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Specify calcium/phosphate homeostasis disorder

- ☐ Hypercalcemia
  - ☐ Hyperparathyroidism
  - ☐ Hypoparathyroidism/Pseudohypoparathyroidism
  - ☐ Hypophosphatemia
  - ☐ Low alkaline phosphatase
  - ☐ Osteoporosis/Osteopenia
  - ☐ Other
- 

Specify 'Other' calcium/phosphate homeostasis disorder

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Specify pituitary disorder

- ☐ Combined pituitary hormone deficiency
  - ☐ Isolated growth hormone deficiency
  - ☐ Pituitary adenoma
  - ☐ Other
- 

Specify 'Other' pituitary disorder

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Specify thyroid disorder

- ☐ Hypothyroidism
  - ☐ Hyperthyroidism
  - ☐ Other
- 

Specify 'Other' thyroid disorder

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Specify glucose/insulin disorder

- ☐ Diabetes
  - ☐ Hyperinsulinemia
  - ☐ Other
- 

Specify 'Other' glucose/insulin disorder

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Specify sex hormone disorder

- ☐ Androgen insensitivity
  - ☐ Hypogonadism
  - ☐ Precocious puberty
  - ☐ Premature ovarian insufficiency
  - ☐ Male infertility
  - ☐ Female infertility
  - ☐ Other
- 

Specify 'Other' sex hormone disorder

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Specify 'Other' endocrine disorders

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Specify hematologic disorders

- ☐ Bone marrow failure
- ☐ Clotting disorder
- ☐ Platelet disorder
- ☐ Red blood cell disorder
- ☐ White blood cell disorder

Specify bone marrow failure

- ☐ Fanconi anemia
- ☐ Short telomere syndrome
- ☐ Other

Specify 'Other' bone marrow failure

\_\_\_\_\_

Specify clotting disorder

- ☐ Frequent bleeding
- ☐ Unusual venous blood clotting
- ☐ Unusual arterial blood clotting
- ☐ Antithrombin III deficiency
- ☐ Factor V Leiden
- ☐ Protein C deficiency
- ☐ Protein S deficiency
- ☐ Thrombophilia
- ☐ Other

Specify 'Other' clotting disorder

\_\_\_\_\_

Specify platelet disorder

- ☐ Platelet dysfunction
- ☐ Thrombocythemia
- ☐ Thrombocytopenia
- ☐ Other

Specify 'Other' platelet disorder

\_\_\_\_\_

Specify red blood cell disorders

- ☐ Aplastic anemia
- ☐ Erythrocytosis
- ☐ Globin disorders
- ☐ Glycolysis-related hemolytic anemia
- ☐ Megaloblastic anemia
- ☐ Red blood cell membrane disorders
- ☐ Sideroblastic anemia
- ☐ Thrombotic microangiopathies/Atypical hemolytic uremic syndrome
- ☐ Other anemia
- ☐ Other red blood cell disorder

Specify globin disorder

- ☐ Beta-thalassemia
- ☐ Alpha-thalassemia
- ☐ Sickle-cell anemia
- ☐ Abnormal serum electrophoresis
- ☐ Other

Specify 'other' globin disorder

\_\_\_\_\_

Specify glycolysis-related hemolytic anemia

- ☐ G6PD deficiency
- ☐ Hexokinase deficiency
- ☐ Pyruvate kinase (PK) deficiency
- ☐ Other

Specify 'Other' glycolysis-related hemolytic anemia

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Specify red blood cell membrane disorder

- ☐ Spherocytosis  
☐ Elliptocytosis  
☐ Stomatocytosis  
☐ Other
- 

Specify 'Other' red blood cell membrane disorders

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Specify 'Other' anemia

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Specify 'Other' red blood cell disorder

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Specify white blood cell disorder

- ☐ Neutropenia  
☐ Severe combined immunodeficiency  
☐ Other
- 

Specify 'Other' white blood cell disorder

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Specify immune system disorders

- ☐ Immunodeficiencies affecting cellular and humoral immunity  
☐ Combined immune deficiencies with associated or syndromic features  
☐ Predominantly antibody deficiency  
☐ Disease of immune dysregulation  
☐ Congenital defects of phagocytic number of function  
☐ Defects of intrinsic and innate immunity  
☐ Autoinflammatory disorder  
☐ Complement deficiency  
☐ Recurrent fever  
☐ Phenocopies of inborn errors of immunity
- 

Specify Immunodeficiencies affecting cellular and humoral immunity

- ☐ T Cell (-) B Cell (+) severe combined immune deficiencies (SCID)  
☐ T Cell (-) B Cell (-) severe combined immune deficiencies (SCID)  
☐ Combined immune deficiencies (CID)
- 

Specify combined immune deficiencies with associated or syndromic features

- ☐ Immunodeficiency with congenital thrombocytopenia  
☐ DNA repair defects other than those classified by Immunodeficiencies affecting cellular and humoral immunity  
☐ Thymic defects with additional congenital anomalies  
☐ Immuno-osseous dysplasias  
☐ Hyper IgE syndrome (HIES)  
☐ Other



Specify predominantly antibody deficiencies	<input type="checkbox"/> Severe reduction in all serum immunoglobulin isotypes with profoundly decreased or absent B cells, agammaglobulinemia <input type="checkbox"/> Severe reduction in at least 2 serum immunoglobulin isotypes with normal or low number of B cells, CVID phenotype <input type="checkbox"/> Severe reduction in serum IgG and IgA with normal/elevated IgM and normal numbers of B cells, hyper IgM <input type="checkbox"/> Isotype, light chain, or functional deficiencies with generally normal numbers of B cells
Specify disease of immune dysregulation	<input type="checkbox"/> Familial hemophagocytic lymphohistiocytosis (FHL syndromes) <input type="checkbox"/> FHL syndromes with hypopigmentation <input type="checkbox"/> Regulatory T cell defects <input type="checkbox"/> Autoimmunity with or without lymphoproliferation <input type="checkbox"/> Immune dysregulation with colitis <input type="checkbox"/> Autoimmune lymphoproliferative syndrome (ALPS, Canale-Smith syndrome) <input type="checkbox"/> Susceptibility to EBV and lymphoproliferative conditions
Specify congenital defects of phagocytic number or function	<input type="checkbox"/> Congenital neutropenias <input type="checkbox"/> Defects of motility <input type="checkbox"/> Defects of respiratory burst <input type="checkbox"/> Other non-lymphoid defects
Specify defects in intrinsic and innate immunity	<input type="checkbox"/> Mendelian susceptibility to mycobacterial disease (MSMID) <input type="checkbox"/> Epidermodysplasia verruciformis (HPV) <input type="checkbox"/> Predisposition to severe viral infection <input type="checkbox"/> Herpes simplex encephalitis (HSE) <input type="checkbox"/> Predisposition to invasive fungal diseases <input type="checkbox"/> Predisposition to mucocutaneous candidiasis <input type="checkbox"/> TLR signaling pathway deficiency with bacterial susceptibility <input type="checkbox"/> Other inborn errors of immunity related to non-hematopoietic tissues <input type="checkbox"/> Other inborn errors of immunity related to leukocytes
Specify autoinflammatory disorders	<input type="checkbox"/> Type 1 interferonopathies affecting the inflammasome <input type="checkbox"/> Non-inflammasome-related conditions
Specify intellectual disability/developmental delay	<input type="checkbox"/> Intellectual disability <input type="checkbox"/> Developmental delay <input type="checkbox"/> Autism <input type="checkbox"/> Cerebral palsy <input type="checkbox"/> Other
Specify developmental delay	<input type="checkbox"/> Gross motor <input type="checkbox"/> Fine motor <input type="checkbox"/> Speech <input type="checkbox"/> Global developmental delay
Specify 'Other' intellectual disability/developmental delay	<hr/>

Specify metabolic disorders

- ☐ Abnormal amino acid Levels
- ☐ Abnormal carnitine levels
- ☐ Abnormal carbohydrate levels
- ☐ Abnormal copper metabolism
- ☐ Abnormal fatty acid oxidation
- ☐ Abnormal glycosylation
- ☐ Abnormal iron metabolism
- ☐ Abnormal liver enzymes
- ☐ Abnormal organic acid metabolism
- ☐ Abnormal urea cycle/Hyperammonemia
- ☐ Abnormal very long chain fatty acids
- ☐ Abnormal zinc metabolism
- ☐ Alpha-1-antitrypsin deficiency
- ☐ Glycogen storage disease
- ☐ Lysosomal storage disorder
- ☐ Macrocytic anemia/abnormal B12/Folate/Homocysteine
- ☐ Mitochondrial disorder
- ☐ Porphyria
- ☐ Rhabdomyolysis
- ☐ Other

Specify abnormal carbohydrate levels

- ☐ Hypoglycemia
- ☐ Glycosuria
- ☐ Other

Specify 'Other' abnormal carbohydrate levels

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Specify abnormal copper metabolism

- ☐ Concern for Menkes disease
- ☐ Concern for Wilson's disease
- ☐ Low ceruloplasmin/serum copper/urine copper
- ☐ Elevated ceruloplasmin/serum copper/urine copper
- ☐ Other

Specify 'Other' abnormal copper metabolism

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Specify abnormal iron metabolism

- ☐ Hemochromatosis/elevated ferritin
- ☐ Aceroplasminemia
- ☐ Other

Specify 'Other' abnormal iron metabolism

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Specify abnormal very long chain fatty acids

- ☐ X-linked adrenoleukodystrophy
- ☐ Refsum disease
- ☐ Zellweger spectrum disorder
- ☐ Rhizomelic chondrodysplasia punctata
- ☐ Other

Specify 'Other' abnormal very long chain fatty acids

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Specify lysosomal storage disorder

- ☐ Concern for Gaucher disease
- ☐ Concern for Fabry disease
- ☐ Concern for Pompe disease
- ☐ Concern for Morquio Syndrome
- ☐ Metachromatic leukodystrophy
- ☐ Other

Specify 'Other' lysosomal storage disorder

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Specify macrocytic anemia/abnormal  
B12/folate/homocysteine

- ☐ Methylmalonic aciduria  
☐ Homocystinuria  
☐ Other

Specify 'Other' macrocytic anemia/abnormal  
B12/folate/homocysteine

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Specify mitochondrial disorder

- ☐ Abnormal mitochondrial testing  
☐ Chronic progressive external ophthalmoplegia  
☐ Lactic acidemia  
☐ Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)  
☐ Severe gastrointestinal dysmotility  
☐ Unexplained fatigue and exercise intolerance  
☐ Other

Specify 'Other' mitochondrial disorder

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Specify 'Other' metabolic disorders

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Specify kidney disorders

- ☐ Congenital anomalies of the kidney and urinary tract (CAKUT)  
☐ Cystic disease  
☐ Glomerular and tubular disease  
☐ Tubulopathies  
☐ Nephrolithiasis

Specify congenital anomalies of kidney and urinary tract (CAKUT)

- ☐ Structural defects of kidney  
☐ Structural defects of urinary tract  
☐ Syndromic CAKUT

Specify cystic disease

- ☐ Renal cysts  
☐ Liver cysts  
☐ Nephronophthisis

Specify glomerular tubular disease

- ☐ Atypical hemolytic uremic syndrome (aHUS)  
☐ Biopsy-suggestive genetic condition  
☐ Chronic kidney disease (CKD)  
☐ Microscopic hematuria  
☐ Nephritic syndrome  
☐ Nephrotic syndrome  
☐ Other

Specify chronic kidney disease

- ☐ Childhood-onset chronic kidney disease (CKD)/end stage renal disease (ESRD)  
☐ Adult-onset chronic kidney disease (CKD)/end stage renal disease (ESRD)

Specify 'Other' glomerular tubular disease

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Specify tubulopathies

- ☐ Chronic hypokalemia with metabolic alkalosis (Bartter/Gittelman syndrome)
- ☐ Chronic hypokalemia with metabolic acidosis/Renal tubular acidosis
- ☐ Other

Specify 'Other' tubulopathies

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Specify nephrolithiasis

- ☐ Calcium oxalate stones/Hyperoxaluria
- ☐ Calcium phosphate stones/Renal tubular acidosis
- ☐ Uric acid renal stones/Hyperuricemia
- ☐ Cystine renal stones/Cystinuria

Specify neurology/neuromuscular disorders

- ☐ Neuromuscular/Muscular
- ☐ Brain iron accumulation
- ☐ Movement disorders
- ☐ Charcot Marie Tooth (CMT)/Neuropathy
- ☐ Cerebrovascular disease
- ☐ Dementia
- ☐ Neurocutaneous
- ☐ Epilepsy
- ☐ Leukodystrophies/Abnormal white matter
- ☐ Encephalopathy
- ☐ Headache
- ☐ Other

Specify neuromuscular/muscular disorder

- ☐ Myopathy/Muscle weakness
- ☐ Congenital myasthenia
- ☐ Myotonia
- ☐ Periodic paralysis
- ☐ Muscular dystrophy
- ☐ Motor neuron disease
- ☐ Hypotonia

Specify myopathy/muscle weakness

- ☐ Proximal muscle weakness
- ☐ Distal muscle weakness
- ☐ Bulbar muscle weakness
- ☐ Elevated creatinine kinase

Specify muscular dystrophy

- ☐ Becker
- ☐ Congenital
- ☐ Distal
- ☐ Duchene
- ☐ Emery-Dreifuss
- ☐ Facioscapulohumeral (FSHD)
- ☐ Limb-girdle
- ☐ Myotonic
- ☐ Oculopharyngeal
- ☐ Other

Specify 'Other' muscular dystrophy

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Specify motor neuron disease

- ☐ Amyotrophic lateral sclerosis (ALS)
- ☐ Progressive bulbar palsy (PBP)
- ☐ Pseudobulbar palsy
- ☐ Primary lateral sclerosis (PLS)
- ☐ Progressive muscular atrophy (PMA)
- ☐ Spinal muscular atrophy (SMA)
- ☐ Monomelic amyotrophy (MMA)
- ☐ Other

Specify 'Other' motor neuron disease

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Specify movement disorder

- ☐ Ataxia
- ☐ Generalized dystonia
- ☐ Hereditary spastic paraplegia (HSP)
- ☐ Huntington's disease/Chorea
- ☐ Parkinsonism
- ☐ Paroxysmal kinesigenic dyskinesia
- ☐ Other

Specify 'Other' movement disorder

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Specify Charcot Marie Tooth (CMT)/Neuropathy

- ☐ Autonomic neuropathy
- ☐ Motor neuropathy
- ☐ Small fiber neuropathy
- ☐ Sensory neuropathy
- ☐ Mixed
- ☐ Other

Specify 'Other' Charcot Marie Tooth (CMT)/Neuropathy

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Specify cerebrovascular disease

- ☐ Ischemic stroke
- ☐ Hemorrhagic stroke
- ☐ Cerebral aneurysm
- ☐ Cerebral artery amyloidosis
- ☐ Suspected CADASIL
- ☐ Moya moya
- ☐ Other

Specify 'Other' cerebrovascular disease

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Specify dementia

- ☐ Alzheimer dementia
- ☐ Fronto-temporal dementia
- ☐ Vascular dementia
- ☐ Idiopathic basal ganglia calcification
- ☐ Creutzfeldt-Jakob disease (CJD)/Other prion disease
- ☐ Other

Specify 'Other' dementia

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Specify Neurocutaneous disorder

- ☐ Neurofibromatosis
- ☐ Tuberous sclerosis
- ☐ Schwannomatosis
- ☐ Other

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Specify 'Other' neurocutaneous disorder

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Specify epilepsy

- ☐ Congenital onset
  - ☐ Infant onset
  - ☐ Childhood onset
  - ☐ Adult onset
  - ☐ Focal epilepsy
  - ☐ Generalized epilepsy
  - ☐ Absence seizures
  - ☐ Epileptic encephalopathy
  - ☐ West syndrome
  - ☐ Structural brain anomalies present
  - ☐ Other
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Specify 'Other' epilepsy

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Specify headache

- ☐ Migraine
  - ☐ Unspecified headache type
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Specify 'Other' neurological/neuromuscular disorders

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Specify ophthalmic disorders

- ☐ Retinal dystrophy
  - ☐ Vitreoretinopathies
  - ☐ Ocular dysgenesis
  - ☐ Optic nerve abnormalities/ septo-optic dysplasia
  - ☐ Glaucoma
  - ☐ Corneal dystrophy
  - ☐ Cataracts
  - ☐ Oculocutaneous albinism
  - ☐ Other
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Specify retinal dystrophy

- ☐ Cone-rod dystrophy
  - ☐ Rod-cone dystrophy
  - ☐ Retinitis pigmentosa
  - ☐ Macular dystrophy
  - ☐ Leber congenital amaurosis
  - ☐ Syndromic retinal dystrophy
  - ☐ Other
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Specify 'Other' retinal dystrophy

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Specify ocular dysgenesis

- ☐ Microphthalmia
  - ☐ Anophthalmia
  - ☐ Coloboma
  - ☐ Anterior segment dysgenesis
  - ☐ Other
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Specify 'Other' ocular dysgenesis

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Specify 'Other' ophthalmic disorders

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Specify pulmonary disorders

- ☐ Bronchiectasis
- ☐ Central hypoventilation
- ☐ Chronic obstructive pulmonary disease (COPD)
- ☐ Cystic fibrosis
- ☐ Pneumothorax
- ☐ Pulmonary Alveolar Proteinosis
- ☐ Pulmonary cysts
- ☐ Pulmonary Fibrosis/ interstitial lung disease
- ☐ Other

Specify 'Other' pulmonary disorders

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Specify skeletal disorders

- ☐ VACTERL/VATER association
- ☐ Congenital diaphragmatic hernia
- ☐ Craniofacial abnormalities
- ☐ Short stature
- ☐ Bone disorders
- ☐ Limb deformities
- ☐ Pterygium
- ☐ Tooth disorders
- ☐ Other

Specify craniofacial abnormalities

- ☐ Blepharophimosis
- ☐ Cleft lip/palate
- ☐ Cleidocranial dysplasia
- ☐ Craniofacial microsomia
- ☐ Craniosynostosis
- ☐ Concern for Treacher Collins syndrome
- ☐ Jaw cysts
- ☐ Lip pits
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Parry Romberg atrophy
- ☐ Robin sequence
- ☐ Nose abnormality
- ☐ Other

Specify craniosynostosis

- ☐ Concern for Crouzon syndrome
- ☐ Concern for Apert syndrome
- ☐ Concern for Muenke syndrome
- ☐ Concern for Pfeiffer syndrome
- ☐ Concern for Saethre-Chotzen syndrome
- ☐ Other

Specify 'Other' craniosynostosis

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Specify 'Other' craniofacial abnormalities

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Specify short stature

- ☐ Achondroplasia
- ☐ Campomelic dysplasia
- ☐ Chondrodysplasia punctata
- ☐ Concern for Silver-Russell syndrome
- ☐ Hypochondroplasia
- ☐ Pseudoachondroplasia
- ☐ Short rib skeletal dysplasia
- ☐ Thanatophoric dysplasia
- ☐ Other disproportionate short stature
- ☐ Other proportionate short stature
- ☐ Other

Specify 'Other' short stature

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Specify bone disorders

- ☐ Clinical concern for osteogenesis imperfecta (OI)
- ☐ Exostoses
- ☐ Frequent bone fractures
- ☐ Osteoarthritis
- ☐ Osteopetrosis
- ☐ Osteoporosis
- ☐ Paget disease
- ☐ Other

Specify 'Other' bone disorders

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Specify limb deformities

- ☐ Club foot
- ☐ Oligodactyly
- ☐ Polydactyly
- ☐ Reduction defect
- ☐ Split hand/foot
- ☐ Syndactyly
- ☐ Thumb abnormality
- ☐ Other

Does thumb abnormality include radial abnormality?

- ☐ Yes
- ☐ No
- ☐ Unknown

Specify 'Other' limb deformity

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Specify tooth disorders

- ☐ Amelogenesis Imperfecta
- ☐ Single front incisor
- ☐ Tooth agenesis
- ☐ Other

Specify 'Other' tooth disorder

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Specify 'Other' skeletal disorders

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Is skeletal disorder syndromic?

- ☐ Yes
- ☐ No



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Specify suspected syndrome(s)

- ☐ Neurofibromatosis type 1
- ☐ Neurofibromatosis type 2
- ☐ Trisomy 21
- ☐ Noonan syndrome
- ☐ Turners syndrome
- ☐ Tuberous Sclerosis
- ☐ Birt-Hogg-Dube syndrome
- ☐ DiGeorge syndrome
- ☐ Lynch syndrome
- ☐ Li-Fraumeni syndrome
- ☐ Von Hippel-Lindau
- ☐ Hereditary breast ovarian cancer syndrome (HBOC)

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Specify 'Other' test indication

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History of allogeneic stem cell transplant?

- ☐ Yes
- ☐ No

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History of solid organ transplant?

- ☐ Yes
- ☐ No