Washington University Physicians®

Clinical Genomics Laboratory: Clinical Exome Sequencing

Shipping address:

Washington University Department of Pathology & Immunology
Clinical Support Office
425 S. Euclid Ave. | MSC 8024-14-4711 | St. Louis MO 63110
Tel: (314) 747-7337 | Fax: (314) 747-7336

Specimen drop-off locations:

Children's Hospital
One Children's Place
Central Receiving 2N-25
St. Louis, MO 63110
Tel: (314) 454-4161

North Campus Lab
Institute of Health (IOH) Core Lab
425 S. Euclid Ave. | Room 4701
St. Louis, MO 63110
Tel: (314) 362-1470

This requisition has two pages, please complete both pages to ensure testing.

PATIENT IDENTIFICATION

Patient Status: ☐ Inpatient ☐ Outpatient ☐ Office Visit

Name Last: | First: | MI:

DOB (mm/dd/yyyy): | Sex: ☐ Male ☐ Female

Medical Record # (if applicable):

Address:

City: | State: | Zip: | Country:

Ethnicity (select all that apply):

☐ African American ☐ Asian ☐ Caucasian/NW European
☐ E Indian ☐ Hispanic ☐ Jewish-Ashkenazi ☐ Jewish-Sephardic
☐ Mediterranean ☐ Native Hawaiian/Pacific Islander ☐ Others

PHYSICIAN ORDERING TEST (NPI required)

Name:

Institution:

NPI: | Email:

Address:

City: | State: | Zip: | Country:

Phone: | Fax:

Alternative Contact Information:

Phone: | Email:

Notes:

SPECIMEN TYPE

Date Collected (mm/dd/yyyy): | Time Collected:

Collected By:

Sample Type (Select one):

☐ Peripheral Blood ☐ Other (please specify)

Directions
1. Draw 3-5 ml of peripheral blood in lavender top EDTA tube.
2. Label tube with patient first/last name, DOB, and collection date/time.
3. Place tube in a biohazard bag and form into document sleeve of the biohazard bag ensuring no patient information is visible.
4. Ship specimen overnight in appropriate packaging at room temperature or with cold pack (Monday-Thursday only).

REASON FOR TESTING (Required-failure to include complete information may delay testing)

Diagnosis:

ICD10 Code(s): | Age of Onset:

CLINICAL INFORMATION

(Orders MUST include the completed clinical features checklist OR clinical notes/records. Also include family medical history/pedigree, if available.)

TESTING REQUESTED

Individual(s) Tested:

☐ Patient Only

☐ Family—Trio, Duo, etc. *please complete the additional family member section below*

Please note: *a completed test requisition form is required for each individual providing a sample for sequencing, including family members.*

FOR ADDITIONAL FAMILY MEMBER(S)

Name (Last, First MI):	Date of Birth (mm/dd/yyyy):	Sample Type: ☐ Peripheral Blood ☐ Other	Relationship to patient	Affected Status ☐ Yes ☐ No
Name (Last, First MI):	Date of Birth (mm/dd/yyyy):	Sample Type: ☐ Peripheral Blood ☐ Other	Relationship to patient	Affected Status ☐ Yes ☐ No
Name (Last, First MI):	Date of Birth (mm/dd/yyyy):	Sample Type: ☐ Peripheral Blood ☐ Other	Relationship to patient	Affected Status ☐ Yes ☐ No

SECONDARY FINDINGS

See consent documentation for details. Please note: this selection must be consistent with the choice selected in the informed consent.

☐ Opt in secondary findings ☐ Opt out secondary findings ☐ Patient's informed consent is completed.

Healthcare Professional Signature to Authorize Testing, Statement of Medical Necessity and Transmission of Results Verification

I certify that the patient specified above and/or their legal guardian has been informed of the benefits, risks, and limitations of the laboratory test(s) requested and Informed Consent has been obtained, as well as any other consent from the patient required by my state in order to perform a genetic test on a specimen has been obtained. I further certify that the test(s) requested is/are medically necessary and the results of this test will be used in the medical management of the patient. The undersigned Client authorizes the Washington University School of Medicine to send Protected Healthcare Information (PHI) as identified in the Health Insurance Portability and Accountability Act (HIPAA) to the facsimile phone number above. Client acknowledges they are solely responsible for adopting and implementing appropriate policies and procedures, including physical safeguards, so that the location and use of the facsimile machine complies with all applicable HIPAA regulations.

Signature:	Date:

Below, office use only:

Date/Time Received:	Accession Number:	Technician Initial:

Insurance and Precertification

Patients are responsible for non-covered services, deductibles, co-insurance, contract exclusions, non-authorized services, and remaining balances after insurance reimbursement. Washington University School of Medicine can only accept authorized Missouri and Illinois MEDICAID covered charges for genetic testing. Other out-of-state welfare programs cannot be billed. Please contact our Patient Accounts Manager office at (314) 362-5641 or via email at pathbillingoffice@path.wustl.edu.

Prior Authorization Number:	ICD10 Code(s):	CPT Codes and Units Authorized :

ATTACH COPY OF INSURANCE CARD (if not available, complete the following)

Policy Holder's Name:	Last:	First:	MI:
Policy Holder's DOB (mm/dd/yyyy):			
Relationship to patient:			
Insurance Co. Name:			
Insurance Co. Phone:			
Plan Name:			
ID #:			
Group #:			

Self-Pay/Patient Financial Assistance

Patients who are self-pay should contact our office to arrange for payment. Financial assistance may be available. For more information, contact our Patient Accounts Manager office at (314) 362-5641 or via email at path-billing@email.wustl.edu.

INSTITUTIONAL BILLING: COMPLETE SECTION BELOW

Institutional Billing

Institution Name:		
Contact Name:		
Email:		
Billing Address:		
City:	State:	Zip:
Phone:	Fax:	

CLINICAL FEATURES CHECKLIST

PRE/PERINATAL

- ☐ Abnormality of septum pellucidum:
  - ☐ Absent septum pellucidum
  - ☐ Cavum septum pellucidum
- ☐ Choroid plexus cyst (CPC)
- ☐ Absent nasal bone
- ☐ Congenital heart defect
- ☐ Intracardiac echogenic focus (IEF)
- ☐ Cystic hygroma
- ☐ Increased nuchal translucency, Size (mm):_____
- ☐ Pleural effusion
- ☐ Pericardial effusion
- ☐ Generalized edema
- ☐ Fetal ascites
- ☐ Hydrops fetalis
- ☐ Diaphragmatic hernia
- ☐ Absent stomach bubble
- ☐ Omphalocele
- ☐ Gastroschisis
- ☐ Echogenic bowel
- ☐ Fetal pyelectasis/hydronephrosis
- ☐ Decreased fetal movement
- ☐ Encephalocele
- ☐ Myelomeningocele/Spina bifida
- ☐ Sacrococcygeal teratoma
- ☐ Intrauterine growth retardation (IUGR)
- ☐ Small for gestational age (SGA)
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Short long bones
- ☐ Small thorax
- ☐ Fetal demise
- ☐ Prematurity, Gestational Age:_____
- ☐ Other:_____

STRUCTURAL BRAIN ABNORMALITIES/IMAGING

- ☐ Abnormal/delayed myelination
- ☐ Abnormality of basal ganglia
- ☐ Abnormality of brainstem
- ☐ Abnormality of white matter:
  - ☐ Periventricular
  - ☐ Other:_____
- ☐ Abnormality of cerebral ventricles:
  - ☐ Colpocephaly
  - ☐ Hydrocephalus
  - ☐ Ventriculomegaly
- ☐ Abnormality of corpus callosum: morphology
  - ☐ Agenesis
    - ☐ Complete
    - ☐ Partial
- ☐ Aplasia/hypoplasia
- ☐ Aplasia/hypoplasia of cerebellar vermis
- ☐ Aplasia/hypoplasia of cerebellum
- ☐ Arnold-Chiari malformation:
  - ☐ Type I
  - ☐ Type II
  - ☐ Unspecified
- ☐ Cerebral atrophy/hypoplasia
- ☐ Cerebral calcification
- ☐ Holoprosencephaly
- ☐ Iron deposition
- ☐ Leukodystrophy
- ☐ Neuronal migration abnormality:
  - ☐ Cortical gyration
    - ☐ Lisssencephaly
    - ☐ Pachygyria
      - ☐ Polymicrogyria
      - ☐ Macrogyria
      - ☐ Simplified gyria
- ☐ Gray matter heterotopia:
  - ☐ Subcortical
  - ☐ Periventricular
- ☐ Other:_____

DEVELOPMENTAL/BEHAVIORAL

- ☐ Aggressive/violent behavior
- ☐ Anxiety
- ☐ Attention-deficit hyperactivity disorder
- ☐ Autistic behavior
- ☐ Autism/autism spectrum disorder
- ☐ Cognitive impairment
- ☐ Delayed fine motor development
- ☐ Delayed gross motor development
- ☐ Developmental regression
- ☐ Gait disturbance Specify:_____
- ☐ Global developmental delay
- ☐ Hyperactivity
- ☐ Incoordination
- ☐ Intellectual disability:
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe/profound
- ☐ Learning disability
- ☐ Language impairment:
  - ☐ Absent speech
  - ☐ Apraxia
  - ☐ Articulation difficulties
  - ☐ Delayed speech and language development
    - ☐ Expressive
    - ☐ Receptive
    - ☐ Dysarthria
    - ☐ Echolalia
    - ☐ Loss of speech
- ☐ Memory impairment
- ☐ Obsessive-compulsive behavior
- ☐ Self-injurious behavior:
  - ☐ Biting
  - ☐ Head-banging
  - ☐ Skin picking
- ☐ Sensory processing disorder/ neurodevelopmental abnormality
- ☐ Sleep disturbance
- ☐ Stereotypy:
  - ☐ Recurrent hand flapping
  - ☐ Stereotypical hand wringing
- ☐ Other:_____

NEUROLOGICAL

- ☐ Abnormality of nervous system
- ☐ Ataxia
- ☐ Athetosis
- ☐ Bradykinesia
- ☐ Cerebral palsy
- ☐ Chorea
- ☐ Cortical visual impairment
- ☐ Dementia
- ☐ Dysarthria
- ☐ Dyskinesia
- ☐ Dysphasia
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Gait disturbance, Specify:_____
- ☐ Headache
- ☐ Hemiplegia
- ☐ Hypotonia
- ☐ Hypertonia
- ☐ Infantile spasms
- ☐ Migraine
- ☐ Myoclonus
- ☐ Neuropathy:
  - ☐ Peripheral
  - ☐ Sensory
- ☐ Parkinsonism/Parkinson Disease
- ☐ Seizures, Type:_____
- ☐ Spasticity
- ☐ Syncope
- ☐ Tremors
- ☐ Vertigo
- ☐ Other:_____

CRANIOFACIAL/DYSMORPHISM

- ☐ Abnormal facial shape, Specify:_____
- ☐ Abnormality of incisors, Specify:_____
- ☐ Ala nasi:
  - ☐ Cleft
  - ☐ Thick
  - ☐ Underdeveloped
- ☐ Anteverted nares
- ☐ Brachycephaly
- ☐ Chin abnormality, Specify:_____
- ☐ Cleft lip:
  - ☐ Unilateral
  - ☐ Bilateral
  - ☐ Midline
- ☐ Cleft palate:
  - ☐ Unilateral
  - ☐ Bilateral
  - ☐ Midline
  - ☐ Submucous cleft
- ☐ Cloverleaf skull
- ☐ Columella abnormality:
  - ☐ Broad
  - ☐ High insertion
  - ☐ Low hanging
  - ☐ Low insertion
  - ☐ Short
- ☐ Craniosynostosis:
  - ☐ Coronal
  - ☐ Lambdoidal
  - ☐ Metopic
  - ☐ Orbital
  - ☐ Sagittal
- ☐ Dolichocephaly
- ☐ Face abnormality:
  - ☐ Broad
  - ☐ Coarse facial features
  - ☐ Flat
  - ☐ Long
  - ☐ Narrow
  - ☐ Round
  - ☐ Short
  - ☐ Square
  - ☐ Triangular
- ☐ Forehead abnormality:
  - ☐ Broad
  - ☐ Narrow
  - ☐ Prominent
  - ☐ Sloping
  - ☐ Creases
- ☐ Frontal bossing
- ☐ Jaw Abnormality:
  - ☐ Broad
  - ☐ Narrow
- ☐ Lip vermilion abnormality
- ☐ Lip abnormality:
  - ☐ Pit
  - ☐ Thin
  - ☐ Thick
  - ☐ Tented
  - ☐ Exaggerated cupid's bow
  - ☐ Absent cupid's bow
- ☐ Malar abnormality:
  - ☐ Flattening
  - ☐ Prominence
- ☐ Midface abnormality:
  - ☐ Flat
  - ☐ Prominence
  - ☐ Retrusion
- ☐ Macrocephaly:
  - ☐ Relative
  - ☐ True
- ☐ Metopic suture abnormality:
  - ☐ Depression
  - ☐ Ridge
- ☐ Microcephaly
- ☐ Micrognathia
- ☐ Nasal base abnormality:
  - ☐ Narrow
  - ☐ Wide
- ☐ Nasal bridge abnormality:
  - ☐ Depressed
  - ☐ Narrow
  - ☐ Prominent
  - ☐ Short

- ☐ Wide
- ☐ Prominent

☐ Nasal cartilage, absent
☐ Nasal ridge abnormality:
- ☐ Depressed
  - ☐ Narrow

☐ Wide
☐ Nasal tip abnormality:
- ☐ Bifid
- ☐ Broad
- ☐ Depressed
- ☐ Deviated
- ☐ Narrow
- ☐ Overhanging

☐ Nasolabial fold abnormality:
- ☐ Prominent
- ☐ Underdeveloped

☐ Neck abnormality:
- ☐ Broad
- ☐ Long
- ☐ Webbed
- ☐ Short
- ☐ Redundant nuchal skin

☐ Nose abnormality:
- ☐ Absent
- ☐ Bifid
- ☐ Long
- ☐ Narrow
- ☐ Prominent
- ☐ Short
- ☐ Wide

☐ Occiput abnormality:
- ☐ Flat
- ☐ Prominent

☐ Plagiocephaly
☐ Philtrum abnormality:
- ☐ Broad
- ☐ Deep
- ☐ Hypoplastic
- ☐ Long
- ☐ Narrow
- ☐ Smooth
- ☐ Short
- ☐ Tented

☐ Proboscis
☐ Prognathism
☐ Retrognathia
☐ Scaphocephaly
☐ Supraorbital ridge abnormality:
- ☐ Prominent
- ☐ Underdeveloped
- ☐ Trigonocephaly
- ☐ Turricephaly
- ☐ Other:_____

EYE/VISION

☐ Age of onset of vision issues:_____
- ☐ Esotropia
- ☐ Exotropia
- ☐ Nystagmus
- ☐ Smooth pursuit
- ☐ Strabismus
- ☐ Other:_____

☐ Abnormality of vision,
- ☐ Specify:_____

☐ Abnormal anterior eye segment morphology
☐ Ablepharon
☐ Achromatopsia
☐ Aniridia
☐ Ankyloblepharon
☐ Anophthalmia
☐ Blepharochalasis
☐ Blepharophimosis
☐ Cataracts
☐ Cataracts, congenital
☐ Coloboma
☐ Corneal opacity
☐ Corneal dystrophy
☐ Cone/cone-rod dystrophy
☐ Congenital stationary night blindness
☐ Cryptophthalmos
☐ Deeply set eyes
☐ Distichiasis
☐ Dyschromatopsia (color blindness)
☐ Ectopia lentis
☐ Ectropion
☐ Entropion
☐ Epiblepharon
☐ Epicanthus/epicanthal folds
☐ Epicanthus inversus
☐ Eyebrow abnormality:
- ☐ Broad
- ☐ Highly arched
- ☐ Horizontal
- ☐ Sparse
- ☐ Thick

☐ Eyelash abnormality:
- ☐ Absent
- ☐ Long
- ☐ Prominent
- ☐ Sparse

☐ Eyelid cleft
☐ External ophthalmoplegia:
- ☐ Progressive

☐ Glaucoma
☐ Infraorbital abnormality:
- ☐ Crease
- ☐ Fold

☐ Iris abnormality,
Specify:_____
☐ Lagophthalmos
☐ Leber optic atrophy
☐ Lens subluxation
☐ Macular abnormality,
Specify:_____
☐ Macular dystrophy
☐ Microphthalmia
☐ Myopia
☐ Ocular albinism
☐ Optic atrophy
☐ Optic neuropathy
☐ Palpebral fissure abnormality:
- ☐ Downslanted
- ☐ Upslanted
- ☐ Long
- ☐ Short
- ☐ Almond-shaped

☐ Proptosis
☐ Ptosis
☐ Retinal flecks
☐ Retinal detachment
☐ Retinitis pigmentosa:
- ☐ Synophrys
- ☐ Telecanthus
- ☐ Other:_____

EARS/HEARING

☐ Age of onset of hearing loss:_____
- ☐ Hearing impairment
  - ☐ Congenital
  - ☐ Progressive
  - ☐ Conductive
  - ☐ Sensorineural
  - ☐ Mixed
  - ☐ Unilateral
  - ☐ Bilateral

☐ Anotia
☐ Abnormal newborn screen,
- ☐ Specify:_____

☐ Antihelix abnormality:
- ☐ Absent
- ☐ Additional crus
- ☐ Angulated
- ☐ Inferior crus broad
- ☐ Inferior crus prominent
- ☐ Inferior crus underdeveloped
- ☐ Superior crus prominent
- ☐ Superior crus underdeveloped

☐ Antitragus abnormality:
- ☐ Absent
- ☐ Bifid
- ☐ Everted
- ☐ Prominent
- ☐ Underdeveloped

☐ Ear abnormality:
- ☐ Abnormality of the tragus
- ☐ Auricular pit
- ☐ Crumpled
- ☐ Cupped
- ☐ Long
- ☐ Low-set
- ☐ Posteriorly rotated
- ☐ Preauricular pit
- ☐ Protruding
- ☐ Short
- ☐ Satyr
- ☐ Tag

☐ Helix abnormality:
- ☐ Cleft
- ☐ Crimped
- ☐ Darwin notch
- ☐ Darwin tubercle
- ☐ Notching
- ☐ Overfolded
- ☐ Prominent
- ☐ Thin

☐ Lobe abnormality:
- ☐ Cleft
- ☐ Forward-facing
- ☐ Large
- ☐ Small
- ☐ Uplifted

☐ Macrotia
Other:_____

ENDOCRINE

☐ Adrenal insufficiency (Addison)
☐ Androgen insensitivity
☐ Androgen excess
☐ Congenital adrenal hypoplasia
☐ Congenital adrenal hyperplasia
☐ Delayed bone age
☐ Delayed puberty
☐ Diabetes insipidus
☐ Diabetes Mellitus
☐ Hyperandrogenism
☐ Hyperglycemia
☐ Hyperphosphatemia
☐ Hyperthyroidism
☐ Hypoglycemia
☐ Hypophosphatemia
☐ Hypothyroidism
☐ Increased cortisol level (Cushing)
☐ Maturity-onset diabetes of the young
☐ Precocious puberty
☐ Rickets
☐ Other:_____

RESPIRATORY

☐ Asthma
☐ Bronchiectasis
☐ Bronchomalacia
☐ Hyperventilation
☐ Hypoventilation
☐ Laryngomalacia
☐ Laryngeal cleft
☐ Pneumothorax
☐ Pulmonary fibrosis
☐ Respiratory insufficiency
☐ Tracheomalacia
☐ Tracheoesophageal fistula
☐ Other:_____

HEMATOLOGIC/IMMUNOLOGIC

☐ Agammaglobulinemia
☐ Allergic rhinitis
☐ Anemia
☐ Hemolytic anemia
☐ Immunodeficiency,
Specify:_____
☐ Lymphopenia
☐ Neutropenia
☐ Pancytopenia
☐ Recurrent infections
☐ Severe combined immunodeficiency
☐ Thrombocytopenia
☐ Other:_____

SKIN/HAIR

☐ Abnormal blistering of the skin,
Specify:_____

- ☐ Abnormality of nail:
  - ☐ Broad
  - ☐ Deep-set
  - ☐ Pits
- ☐ Albinism
- ☐ Alopecia
- ☐ Anhidrosis
- ☐ Cafe-au-lait spot:
  - ☐ Single
  - ☐ Multiple
- ☐ Coarse hair
- ☐ Collodion baby
- ☐ Cutaneous photosensitivity
- ☐ Cutis laxa
- ☐ Dry skin
- ☐ Eczema
- ☐ Erythematous skin
- ☐ Hemangioma
- ☐ Hairline:
  - ☐ Low
  - ☐ High
  - ☐ Anterior
  - ☐ Posterior
- ☐ Hyperextensible skin
- ☐ Hyperpigmentation of the skin
- ☐ Hypopigmentation of the skin
- ☐ Hypohidrosis
- ☐ Ichthyosis
- ☐ Jaundice
- ☐ Lipoma
- ☐ Lymphedema
- ☐ Palmoplantar keratoderma
- ☐ Scarring of skin
- ☐ Skin rash
- ☐ Sparse hair
- ☐ Telangiectasia
- ☐ Vascular skin abnormality
- ☐ Velvety skin
- ☐ Other:_____

CARDIAC

- ☐ Amyloidosis
- ☐ Aortic root dilatation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Atrioventricular canal defect
- ☐ Arrhythmogenic right ventricular dysplasia
- ☐ Bicuspid aortic valve
- ☐ Bradycardia
- ☐ Coarctation of the aorta
- ☐ Congenital heart defect
- ☐ Dilated cardiomyopathy
- ☐ Double outlet right ventricle
- ☐ Ebstein anomaly
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Hypertrophic cardiomyopathy
- ☐ Mitral valve prolapse
- ☐ Noncompaction cardiomyopathy
- ☐ Patent ductus arteriosus
- ☐ Patent foramen ovale
- ☐ Prolonged QTc interval
- ☐ Pulmonary hypertension:
  - ☐ Arterial
  - ☐ Vascular
- ☐ Sudden death
- ☐ Tetralogy of Fallot
- ☐ Transposition of the great vessels
- ☐ Truncus arteriosus
- ☐ Ventricular septal defect
- ☐ Ventricular tachycardia
- ☐ Other:_____

GASTROINTESTINAL

- ☐ Biliary atresia
- ☐ Cholestasis
- ☐ Constipation:
  - ☐ Acute
  - ☐ Chronic
  - ☐ Diarrhea
- ☐ Diaphragmatic hernia
- ☐ Duodenal stenosis/atresia
- ☐ Esophageal stenosis/atresia
- ☐ Exocrine pancreatic insufficiency
- ☐ Failure to thrive
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Gastroschisis
- ☐ Hepatomegaly
- ☐ Hepatosplenomegaly
- ☐ Inflammatory bowel disease
- ☐ Jaundice
- ☐ Liver disease
- ☐ Liver failure
- ☐ Nausea
- ☐ Omphalecele
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Splenomegaly
- ☐ Tracheoesophageal fistula
- ☐ Tube feeding:
  - ☐ Nasogastric
  - ☐ Gastrostomy
  - ☐ Gastrojejunal
- ☐ Umbilical hernia
- ☐ Vomiting
- ☐ Other:_____

GENITOURINARY

- ☐ Abnormality of the uterus, Specify:_____
- ☐ Ambiguous genitalia
- ☐ Chordee
- ☐ Cryptorchidism
- ☐ Duplicated collecting system
- ☐ Horseshoe kidney
- ☐ Hydronephrosis
- ☐ Hypospadias/epispadias
- ☐ Inguinal hernia
- ☐ Micropenis
- ☐ Multicystic kidney dysplasia
- ☐ Nephrolithiasis
- ☐ Polycystic kidney disease
- ☐ Renal agenesis/hypoplasia:
  - ☐ Unilateral
  - ☐ Bilateral
- ☐ Sex reversal
- ☐ Vesicoureteral reflux
- ☐ Other:_____

MUSCULOSKELETAL

- ☐ Abnormal connective tissue
- ☐ Abnormal digit morphology:
  - ☐ Broad
  - ☐ Short
  - ☐ Clinodactyly
  - ☐ Ectrodatyly
  - ☐ Oligodactyly
  - ☐ Polydactyly
    - ☐ Postaxial
    - ☐ Preaxial
  - ☐ Syndactyly
- ☐ Arachnodactyly
- ☐ Arthralgia
- ☐ Arthrogryposis
- ☐ Bruising susceptibility
- ☐ Chest abnormality:
  - ☐ Small chest
  - ☐ Barrel-shaped
  - ☐ Bell-shaped thorax
  - ☐ Pectus carinatum
  - ☐ Pectus excavatum
- ☐ Contractures of joint(s)
- ☐ Decreased muscle mass
- ☐ Delayed bone age
- ☐ Dolichostenomelia
- ☐ Exercise intolerance
- ☐ Fatigue
- ☐ Fracture(s)
- ☐ Hemihypertrophy
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Joint hypermobility
- ☐ Kyphosis
- ☐ Limb shortening:
  - ☐ Mesomelic
  - ☐ Micromelic
  - ☐ Rhizomelic
- ☐ Metaphyseal abnormalities:
  - ☐ Dumbbell
  - ☐ Flared
- ☐ Muscle weakness
- ☐ Myalgia
- ☐ Myopathic facies
- ☐ Myopathy
- ☐ Myelomeningocele/Spina Bifida/ Neural
- ☐ Tube Defect
- ☐ Osteoarthritis
- ☐ Osteoporosis
- ☐ Osteopenia
- ☐ Pain:
  - ☐ Absent/decreased
  - ☐ Abnormal sensation
  - ☐ Episodic
  - ☐ Limb
  - ☐ Muscle
- ☐ Platyspondyly
- ☐ Recurrent fractures
- ☐ Rhabdomyolysis
- ☐ Rib abnormalities:
  - ☐ Cupped
  - ☐ Fused
  - ☐ Supernumerary
  - ☐ Missing
  - ☐ Short
  - ☐ Spatulate
  - ☐ Other:_____
- ☐ Rickets
- ☐ Scoliosis
- ☐ Short stature
- ☐ Skeletal dysplasia
- ☐ Talipes:
  - ☐ Equinovarus
  - ☐ Other:_____
- ☐ Tall stature
- ☐ Thoracic dysplasia
- ☐ Thumb abnormality:
  - ☐ Adducted
  - ☐ Broad
  - ☐ Triphalangeal
- ☐ Vertebral bodies, abnormal form:
  - ☐ Aplasia/hypoplasia
  - ☐ Butterfly
  - ☐ Fusion
  - ☐ Hemivertebrae
- ☐ Other:_____

VASCULAR SYSTEM:

- ☐ Aneurysm:
  - ☐ Aortic
    - ☐ Abdominal
    - ☐ Dissecting
    - ☐ Thoracic
  - ☐ Cerebral
  - ☐ Other:_____
- ☐ Arterial calcification
- ☐ Arterial dissection
- ☐ Arterial tortuosity
- ☐ Arteriovenous malformation
- ☐ Epistaxis
- ☐ Lymphedema
- ☐ Pulmonary hypertension:
  - ☐ Arterial
  - ☐ Vascular
- ☐ Stroke
- ☐ Other:_____

CANCER

Primary Diagnosis:

Type:_____
Histologic Type:_____
Location:_____
- ☐ Unilateral
- ☐ Bilateral
- ☐ Metastatic

Age of onset:_____

Secondary Diagnosis:

Type:_____
Histologic Type:_____
Location:_____
- ☐ Unilateral
- ☐ Bilateral

☐ Metastatic
☐ Family history of cancer,
Specify: _____
Include pathology reports

OTHER TESTING
Provide copy of report(s)

Echocardiogram: _____
EEG: _____
EMG/NCV: _____
Biopsy: _____
Gene testing: _____
Results: _____

If you would like us to comment on the presence/absence of previously identified variants, provide a copy of the original report.
Chromosomal Microarray:
(CMA): _____
MRI Brain: _____

MRI (other): _____
CT Brain: _____
CT (other): _____
Muscle biopsy: _____
Ultrasound: _____
X-Ray: _____

METABOLIC FINDINGS
Attach relevant lab reports and values
Mark where appropriate increased (↑), decreased (↓), or within normal limits (WNL).

	↑	↓	WNL
☐ Abnormal newborn screen	☐	☐	☐
Specify: _____			
☐ Acylcarnitine profile	☐	☐	☐
Specify: _____			
☐ Acylglycines	☐	☐	☐
Specify: _____			
☐ Amino Acids			
☐ Plasma	☐	☐	☐
Specify: _____			
☐ Urine	☐	☐	☐
Specify: _____			
☐ Muscle	☐	☐	☐
Specify: _____			
☐ Creatine phosphokinase (CPK)	☐	☐	☐
☐ Essential fatty acids			
☐ Plasma	☐	☐	☐
Specify: _____			
☐ Folate	☐	☐	☐
☐ Glycosylation studies	☐	☐	☐
Specify: _____			

	↑	↓	WNL
☐ Hepatic transaminases	☐	☐	☐
☐ Homocystine	☐	☐	☐
☐ Hormones	☐	☐	☐
Specify: _____			
☐ Hyperammonemia	☐	☐	☐
☐ Hyperbillirubinemia	☐	☐	☐
☐ Hyperglycemia	☐	☐	☐
☐ Hyperlipidemia	☐	☐	☐
☐ Ketones			
☐ Plasma	☐	☐	☐
Specify: _____			
☐ Urine	☐	☐	☐
Specify: _____			
☐ Other enzymes	☐	☐	☐
Specify: _____			
☐ Porphyrins			
☐ Plasma	☐	☐	☐
Specify: _____			
☐ Urine	☐	☐	☐
Specify: _____			
☐ Stool	☐	☐	☐
Specify: _____			
☐ Pterins	☐	☐	☐
Specify: _____			

	↑	↓	WNL
☐ Purine/pyrimidines	☐	☐	☐
Specify: _____			
☐ Serum alpha fetoprotein (AFP)	☐	☐	☐
☐ Serum pyruvate	☐	☐	☐
☐ Sterols/Oxysterols	☐	☐	☐
Specify: _____			
☐ Transferrin IEF	☐	☐	☐
Specify: _____			
☐ Uric Acid	☐	☐	☐
☐ Very long chain fatty acids (VLCFA)			
☐ Plasma	☐	☐	☐
Specify: _____			
☐ Vitamins/minerals			
☐ Copper	☐	☐	☐
☐ Magnesium	☐	☐	☐
☐ Manganese	☐	☐	☐
☐ Vitamin B6	☐	☐	☐
☐ Vitamin B12	☐	☐	☐
☐ Vitamin D	☐	☐	☐
☐ Zinc	☐	☐	☐
☐ Other: _____	☐	☐	☐