

Referrer Information

Physician: UPIN/NPI:

Genetic counselor: Email:

Institution:

Address:

Phone: Fax:

Additional reports to:

Address:

Phone: Fax:

MANDATORY SIGNATURE

I have confirmed that the patient has consented for the testing ordered and that two matching identifiers are present on each page of this requisition.

Signature*:

Date:

Patient Information (*two of these identifiers MUST also appear on the sample)

Legal Name* (Last): (First):

Preferred Name (Last): (First):

Date of birth* (mm/dd/yy): Sex assigned at birth: Gender:

Patient ID/MRN*:

Billing Information (Contact Billing Coordinator at 443-287-2486 prior to submitting)

- Referring center
- Patient insurance
- Maryland Medicaid
- Self-pay
- Medicare

Billing contact:

Phone:

Fax:

Email:

Shipping Address

1812 Ashland Ave
Sample Intake Rm 245
Baltimore, MD 21205

For sample specific requirements please reference our website.

Clinical Information

Patient Name*: DOB*:

Indication:

ICD Codes:

Please attach detailed medical records.

Ancestry:

- | | | |
|--|---|---|
| <input type="checkbox"/> Caucasian | <input type="checkbox"/> African | <input type="checkbox"/> Pacific Islander |
| <input type="checkbox"/> Northern European | <input type="checkbox"/> Hispanic | <input type="checkbox"/> Native American |
| <input type="checkbox"/> Western European | <input type="checkbox"/> Central/South American | <input type="checkbox"/> Other |
| <input type="checkbox"/> Eastern European | <input type="checkbox"/> Caribbean | |
| <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Asian | |
| <input type="checkbox"/> African American | | |

Test ordered:

Targeted variant Gene _____ c. _____
Relationship to proband _____

Was this variant previously identified by the DNA Diagnostic Laboratory? Yes

*Please attach family member's report. No

Pregnancy-related

Sample Information

Sample type:

- | | |
|---|---|
| <input type="checkbox"/> Whole blood | <input type="checkbox"/> Cleaned chorionic villi |
| <input type="checkbox"/> Extracted DNA | <input type="checkbox"/> Cultured chorionic villi |
| <input type="checkbox"/> Cultured amniocytes | <input type="checkbox"/> Cord blood |
| <input type="checkbox"/> Saliva (Zoom panels only) | |
| <input type="checkbox"/> Other <input type="text"/> | |

The DNA Diagnostic Laboratory only accepts isolated or extracted nucleic acids for which the extraction or isolation is performed in a CLIA-certified laboratory or a laboratory meeting equivalent (or more stringent) requirements as determined by the College of American Pathologists (CAP) and/or the Centers for Medicare and Medicaid Services (CMS)

Collection date:

Patient has had a transfusion or bone marrow transplant?

Yes No

*If yes, please contact the laboratory prior to sending.

Patient Informed Consent (Either consent MUST be completed before testing is initiated)

I grant permission for Johns Hopkins Genomics to perform the genetic test listed on this form for me/my child. The results of genetic testing may be dependent upon the clinical information provided to the laboratory by my physician. The laboratory can not guarantee turn-around-time. Risks and limitations of this test may include, but are not limited to, disclosure of unexpected family information (non-paternity, consanguinity), uninformative negative results, unexpected findings, and lab error. De-identified clinical or genetic information may be used for quality control purposes, research, and shared in public healthcare databases. Results will be released only to the providers authorized on the test requisition. I understand the benefits, risks, and limitations of this genetic testing.

Signature:

Date:

Provider Alternate Consent

I, the health care provider requesting the above testing, have explained the benefits and drawbacks of genetic testing to the patient and have obtained verbal consent or an alternate written consent (please attach) to order the test indicated.

Signature:

Date:



Patient Name*:

DOB*:

Phenotypic Information

Perinatal

- IUGR
- Oligohydramnios
- Polyhydramnios
- Prematurity

Developmental

- Fine motor delay
- Gross motor delay
- Speech delay
- Intellectual disability
- Learning disability
- Regression

Behavioral

- Anxiety
- Depression
- Autism spectrum disorder
- Autistic features
- Obsessive-compulsive disorder

Craniofacial

- Macrocephaly
- Microcephaly
- Craniosynostosis
- Cleft lip/palate
- Dental abnormalities
- Facial dysmorphism:

Eyes and Vision

- Myopia
- Hyperopia
- Visual Impairment
- Blindness
- Coloboma
- Strabismus

Ears and Hearing

- Sensorineural hearing loss
- Conductive hearing loss
- Mixed hearing loss
- Preauricular pit
- External ear malformation
- Low-set ears

Brain Abnormalities

- Agenesis of the corpus callosum
- Holoprosencephaly
- Lissencephaly
- Encephalocele
- Hydrocephalus
- Cortical dysplasia
- Brain atrophy

Neuromuscular

- Ataxia
- Chorea
- Dystonia
- Hypertonia
- Hypotonia
- Seizures

Skeletal

- Tall stature
- Short stature
- Overgrowth
- Joint hypermobility
- Contractures
- Polydactyly
- Syndactyly
- Scoliosis

Skin and Hair

- Café-au-lait spots
- Blistering
- Ichthyosis
- Abnormal pigmentation
- Alopecia
- Coarse hair
- Brittle hair
- Nail abnormalities

Cardiac

- ASD
- VSD
- Tetralogy of Fallot
- Cardiomyopathy
- Arrhythmia

Gastrointestinal

- Gastroschisis/omphalocele
- Pyloric stenosis
- Tracheoesophageal fistula
- Gastroesophageal reflux
- Hirschsprung disease
- Constipation

Additional Clinical Information

Patient Information

Patient Name*:

DOB*:

Test Directory

Clinical Exome Sequencing

Please see separate requisition form

Zoom Panels (see website for full gene lists)

* **CardioZoom** (coming soon!)

ImmunoZoom

- Auto-inflammatory disorders
- Complement deficiencies
- Intrinsic/innate immune defects
- Defects of phagocyte number and function
- Immune dysregulation
- Antibody deficiencies
- Syndromic immunodeficiencies
- Combined immunodeficiencies

* **NeuroZoom** (coming soon!)

SkeletalZoom

- Dysplasias and craniosynotoses
- Osteogenesis imperfecta
- * Connective tissue disorders (coming soon!)

PulmZoom

- Mucociliary disorders
- Interstitial lung disease
- Pulmonary vascular disease

RenalZoom

- CAKUT, ciliopathies, and tubulointerstitial diseases
- Disorders of ion transport, nephrolithiasis and nephrocalcinosis
- Glomerular disease and complement genes

TeloZoom

Lung disease

- CFTR* intron 8 T/TG tract only
- Cystic fibrosis (*CFTR*) NGS
- CFTR* and CF-related disorders NGS panel (with *CFTR*)
- CF-related disorders NGS panel (without *CFTR*)
- Neonatal respiratory distress NGS panel

Linkage assays

- Cystic fibrosis (*CFTR*) linkage analysis
- Duchenne/Becker muscular dystrophy (*DMD*)
- Maternal cell contamination study

Peroxisomal disorders

- Acatalasemia (*CAT*) NGS
- Alpha-methylacyl-CoA racemase deficiency (*AMACR*) NGS
- Comprehensive peroxisomal genes NGS panel
- Mulibrey nanism (*TRIM37*) NGS
- Peroxisomal β -oxidation defects NGS panel
- Primary hyperoxaluria type I (*AGXT*) NGS
- Refsum disease NGS panel
- Rhizomelic chondrodysplasia punctata NGS panel
- Zellweger spectrum disorders (ZSD) NGS panel
- ZSD + peroxisomal β -oxidation defects NGS panel

Movement disorders

- Benign hereditary chorea (*NKX2-1* sequence; reflex to MLPA)
- Huntington disease (*HTT* repeat sizing)
- Huntington disease like 2 (*JPH3* repeat sizing)

GNAS spectrum disorders

- Pseudohypoparathyroidism type 1A (*PHP1A*) / Albright hereditary osteodystrophy (*GNAS* sequencing)

Other conditions

- Choreoathetosis, hypothyroidism, respiratory distress syndrome (*NKX2-1* sequence; reflex to MLPA)
- Hereditary non-syndromic sensorineural hearing loss (*GJB2* sequence; *GJB6* deletion assay)
- Pseudohypoaldosteronism (*PHA*) type 1 NGS panel
- X-Adrenoleukodystrophy (*ABCD1* sequence; reflex to MLPA)

Targeted sequencing is available upon request. Please contact the lab prior to ordering.