

EXOME SEQUENCING TEST REQUISITION

All Information Must Be Completed Before Sample Can Be Processed

PATIENT INFORMATION

Patient Name: _____, _____, _____
Last First MI

Address: _____

Home Phone: _____

MR# _____ Date of Birth ____/____/____

Gender: Male Female

ETHNIC/RACIAL BACKGROUND (Choose All)

- European American (White) African-American (Black)
- Native American or Alaskan Asian-American
- Pacific Islander Ashkenazi Jewish ancestry
- Latino-Hispanic _____
(specify country/region of origin)
- Other _____
(specify country/region of origin)

BILLING INFORMATION (Choose ONE method of payment)

REFERRING INSTITUTION

Institution: _____

Address: _____

City/State/Zip: _____

Accounts Payable Contact Name: _____

Phone: _____

Fax: _____

Email: _____

COMMERCIAL INSURANCE*

Insurance can only be billed if requested at the time of service.

Policy Holder Name: _____

Gender: _____ Date of Birth ____/____/____

Authorization Number: _____

Insurance ID Number: _____

Insurance Name: _____

Insurance Address: _____

City/State/Zip: _____

Insurance Phone Number: _____

* PLEASE NOTE:

- We will not bill Medicaid, Medicaid HMO, or Medicare except for the following: Cincinnati Children's Patients, Cincinnati Children's Providers, or Designated Regional Counties.
- If you have questions, please call 1-866-450-4198 for complete details.

REFERRING PHYSICIAN

Physician Name (print): _____

Address: _____

Phone: (____) _____ Fax: (____) _____ Email: _____

Genetic Counselor/Lab Contact Name: _____

Phone: (____) _____ Fax: (____) _____ Email: _____

Date: ____/____/____

Referring Physician Signature (REQUIRED)

Patient signed completed ABN

Medical Necessity Regulations: At the government's request, the Molecular Genetics Laboratories would like to remind all physicians that when ordering tests that will be paid under federal health care programs, including Medicare and Medicaid programs, that these programs will pay only for those tests the relevant program deems to be (1) included as covered services, (2) reasonable, (3) medically necessary for the treatment and diagnosis of the patient, and (4) not for screening purposes.

SAMPLE/SPECIMEN INFORMATION

Has patient received a bone marrow transplant? Yes No

If yes, date of bone marrow transplant _____

Percent engraftment _____

Please send saliva kit. Note: STR analysis at an additional charge is required on saliva samples obtained on all patients post BMT.

Specimen Type:

Blood (> 3ml) DNA (>15 ug)

Other _____

Proband: Specimen Date: _____ / _____ / _____ Time: _____

Specimen Amount: _____

DRAWN BY: _____

*Phlebotomist must initial tube of specimen to confirm sample identity

Each test requires 3 mL of whole blood in EDTA tube. Please call before sending other sample types.

Mother: Name: _____

Date of Birth: _____ / _____ / _____

Specimen Date: _____ / _____ / _____ Time: _____

Specimen Amount: _____ DRAWN By: _____

Father: Name: _____

Date of Birth: _____ / _____ / _____

Specimen Date: _____ / _____ / _____ Time: _____

Specimen Amount: _____ DRAWN By: _____

Other: Name: _____

Date of Birth: _____ / _____ / _____

Relationship to proband: _____

Specimen Date: _____ / _____ / _____ Time: _____

Specimen Amount: _____ DRAWN By: _____

Please use the space on the last page if needed.

CLINICAL EXOME SEQUENCING TEST REQUESTED

- Family trio (proband and two family members)
- Proband only
- Additional family member; please provide the proband's full name and birthdate:
Proband's full name: _____
Birthdate: _____ / _____ / _____

Has the proband had any of the following sequencing tests performed in our lab at CCHMC in the past 5 years? Select all that apply:

- Autism, Intellectual Disability, and Developmental Delay Panel
- Cardiovascular Diseases Sequencing Panel (*See list of panels below)
- Circadian and Complex Sleep Disorders Gene Sequencing Panel
- Immunology Exome
- Neurovascular Diseases and Stroke Gene Panel
- Platelet Disorders Gene Sequencing Panel
- Rubinstein-Taybi and Related Syndromes Gene Panel

Note: The lab will review each case with prior testing from the list above to determine if data re-analysis is applicable or if full processing is required.

***Cardiovascular Diseases Sequencing Panels include:** Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel, Atrial Fibrillation (A Fib) Panel, Atrioventricular Block (AV Block) Panel, Brugada Syndrome Panel, Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel, Comprehensive Arrhythmia Panel, Comprehensive Cardiomyopathy Panel, Congenital Heart Disease Panel, Dilated Cardiomyopathy (DCM) Panel, Heterotaxy Panel, Hypertrophic Cardiomyopathy (HCM) Panel, Left Ventricular Noncompaction (LVNC) Panel, Long QT Syndrome Panel, Marfan Syndrome and MFS Related Disorders Panel, Pulmonary Arterial Hypertension (PAH) Panel, RASopathy/Noonan Spectrum Disorders Panel, Restrictive Cardiomyopathy (RCM) Panel, Short QT Syndrome Panel, or Thoracic Aortic Aneurysm Panel

Checklist of items to include

- Proband's sample
- Maternal sample (for trio testing)
- Paternal sample (for trio testing)
- Other family member's sample (following discussion with laboratory)
- Test requisition (all billing and clinical information must be completed)
- Signed informed consent form
- Family history and pedigree
- Detailed patient clinical history/clinical summary or notes
- Summary of previous genetic test results
- Letter of medical necessity stating impact of whole exome sequencing results on medical management

Please include all information with sample(s) or fax to 513-636-4373.

CLINICAL HISTORY

Complete clinical information is critical to interpretation of exome sequencing results

Please indicate the presence of and describe the symptoms found in the patient and other family members. In addition, please submit the proband's clinical summary, relevant medical records and summary of previous test results.

Proband's working diagnosis: _____

Congenital anomalies (specify): _____

Dysmorphic features (specify): _____

Previous diagnostic testing (please include reports, when available)

Chromosome/FISH analysis _____

Microarray _____

Single gene testing _____

Radiologic studies (MRI, CT, Xrays) _____

Muscle/skin biopsy _____

Metabolic testing _____

Other relevant results: _____

CLINICAL HISTORY, CONTINUED

Please check all that apply and specify in the space provided. If family member is similarly affected, please indicate relation to proband.

Prenatal/Perinatal History

Proband Family

- Intrauterine growth retardation _____
- Prematurity/postmaturity _____
- Poly/oligohydramnios _____
- Maternal complications of pregnancy _____
- Other _____

General History

Proband Family

- Failure to thrive _____
- Obesity/overgrowth/tall stature _____
- Postnatal growth retardation/short stature _____
- Developmental delay _____
 Gross motor Fine motor Speech
- Intellectual disability _____
- Developmental regression _____
- Autism/autism spectrum disorder _____
- Psychiatric disorder _____
- Behavioral disorder _____
- Cancer Age of onset _____ Type _____
- Sudden infant death / Sudden unexplained death
- Other _____

Review of Systems

Proband Family

Cardiovascular findings

- Arrhythmia/conduction defect
- Cardiomyopathy
- Congenital heart defect (specify _____)
- Vascular abnormalities
- Others: _____

Craniofacial/Ophthalmologic/Auditory findings

- Blindness
- Cataract
- Cleft palate / cleft lip
- Coloboma
- Craniosynostosis
- Dysmorphic facial features
- External ear malformation
- Glaucoma
- Hearing loss
- Hemangioma
- Hemifacial microsomia
- Macrocephaly
- Microcephaly
- Retinal disorder
- Vascular malformation
- Others: _____

Dermatologic findings

- Alopecia
- Angiokeratoma
- Blistering of skin or mucosa
- Cafe au lait spots
- Hypopigmentation/ hyperpigmentation
- Nail dysplasia
- Rash/dermatitis/eczema
- Others: _____

Proband Family

Endocrine disorders

- Adrenal abnormality
- Diabetes type 1
- Growth hormone abnormality
- Sex hormone abnormality
- Thyroid disease
- Others: _____

Gastrointestinal findings

- Abnormal liver function
- Achalasia
- Acute liver failure
- Bile duct proliferation
- Biliary atresia
- Cirrhosis
- Chronic constipation
- Chronic diarrhea
- Congenital anomaly (specify _____)
- Hepatomegaly
- Jaundice
- Liver cysts
- Others: _____

Genitourinary findings

- Abnormal renal function (specify _____)
- Ambiguous genitalia
- Congenital genital anomaly (specify _____)
- Obstructive renal disease
- Renal hypoplasia/agenesis/dysgenesis
- Renal cysts
- Renal morphological anomalies (such as horseshoe kidney)
- Sex reversal
- Undescended testicles
- Others: _____

Hematologic findings

- Anemia
- Bone marrow failure
- Excessive bruising
- Hematomas
- Leukemia/lymphoma
- Leukopenia/neutropenia
- Splenomegaly
- Thrombosis
- Thrombocytopenia/small platelets
- Others: _____

Immunologic/allergic findings

- Allergies (specify _____)
- Autoimmune disorders
- Diffuse inflammation
- Enlarged lymph nodes
- Fevers
- Recurrent, unusual or difficult to treat infections
 viral bacterial fungal
- Small lymph nodes and/or tonsils
- Thymic hypoplasia

Neuromuscular findings

- Abnormal movements
- Ataxia
- Exercise intolerance/fatigue
- Headaches/migraines
- Hypertonia
- Hypotonia
- Neuropathy
- Seizure / epilepsy
- Stroke/stroke like episodes
- Torticollis
- Others: _____

CLINICAL HISTORY, CONTINUED

Proband Family

Skeletal findings

- | | | |
|--------------------------|--------------------------|---------------------------------|
| <input type="checkbox"/> | <input type="checkbox"/> | Arachnodactyly |
| <input type="checkbox"/> | <input type="checkbox"/> | Bone malformation |
| <input type="checkbox"/> | <input type="checkbox"/> | Clinodactyly |
| <input type="checkbox"/> | <input type="checkbox"/> | Disproportionate short limbs |
| <input type="checkbox"/> | <input type="checkbox"/> | Joint contractures |
| <input type="checkbox"/> | <input type="checkbox"/> | Kyphosis |
| <input type="checkbox"/> | <input type="checkbox"/> | Polydactyly |
| <input type="checkbox"/> | <input type="checkbox"/> | Scoliosis |
| <input type="checkbox"/> | <input type="checkbox"/> | Short stature, proportionate |
| <input type="checkbox"/> | <input type="checkbox"/> | Short stature, disproportionate |
| <input type="checkbox"/> | <input type="checkbox"/> | Syndactyly |
| <input type="checkbox"/> | <input type="checkbox"/> | Vertebral anomaly |
| <input type="checkbox"/> | <input type="checkbox"/> | Others: _____ |

ICD-9 Code: _____

Differential Diagnosis: _____

Additional Suspected Genes or Pathways: _____

Additional clinical info:

FAMILY HISTORY

Parental consanguinity Yes No

Please draw or attach patient's three generation pedigree: