Shodair Children's Hospital Genetics Laboratory
2755 Colonial Dr, Helena. MT, 59601
Phone (406) 444-7532 Toll Free (800) 447-6614
Fax (406) 444-1022
email: mtgene@shodair.org

GENETICS LABORATORY TEST REQUEST FORM

Shodair Lab Number

PATIENT INFORMATION

_____
Last Name

_____ **MI** _____
First Name

_____ / _____ / _____
Date of Birth

Sex:
☐ Female ☐ Male

Ethnicity *select all that apply*
- ❑ Caucasian
- ❑ Asian
- ❑ Hispanic
- ❑ African American
- ❑ Ashkenazi Jewish
- ❑ Hutterite
- ❑ American Indian

SAMPLE INFORMATION

- ❑ Whole Blood (≥3mL)
- ❑ Direct Amniotic Fluid
- ❑ Cultured Amniocytes (2-T25)
- ❑ Direct CVS
- ❑ Cultured CVS (2-T25)
- ❑ Saliva/Buccal Cells
- ❑ Extracted DNA (≥10ug) Source:_____
- ❑ Fresh/Frozen Tissue Source:_____

Date of Collection_____

Reference #_____

ADDITIONAL FAMILY SAMPLES (EDTA blood/saliva)

Relationship:_____
Name:_____
DOB:_____ Ref #:_____
Date Collected: _____
Affected: ☐ Yes ☐ No

Relationship:_____
Name:_____
DOB:_____ Ref #:_____
Date Collected: _____
Affected: ☐ Yes ☐ No

ORDERING HEALTH CARE PROFESSIONAL

Name: _____
NPI #: _____
Address: _____
City, State, Zip: _____
Telephone:(_____)_____ FAX:(_____)_____
Referring Facility:_____
Additional Reports To: _____

AUTHORIZATION

By submitting this requisition, I confirm that I have obtained the patient's informed consent for the requested test. I confirm that this test is clinically valuable for the patient.

_____ _____
Signature of ordering provider **Date**

INSTITUTIONAL BILLING

Institution: _____
Address: _____
City, State, Zip: _____

Billing Contact: _____
Phone #: _____
Fax #: _____

MEDICAID / MEDICARE

Name of policy holder: _____
Policy holder DOB: _____
Address: _____
City, State, Zip: _____

Passport ID: _____
Phone #: _____
MEDICAID / MEDICARE #: _____

INSURANCE BILLING

Name of policy holder: _____
Policy holder DOB: _____
Patient Relation to Policy Holder: _____
SS # (Guarantor): _____
Address: _____
City, State, Zip: _____
Phone #: _____
Ins. Co. Policy #: _____
Name of Ins. Co _____
Ins. Co Phone: _____

SELF PAY

Name of responsible party: _____
Relationship to patient: _____Phone: _____
Please call the Financial Assistance Coordinator at (406)444-7507 to arrange payment options

PREAUTHORIZATION ASSISTANCE*

- ❑ **YES** ***CLINICAL INFORMATION REQUIRED**
- ❑ **NO** Successful preauthorization requires complete clinical information.

Please designate a contact for preauthorization updates:
Name:_____
Phone or email: _____

SHODAIR INTERNAL USE ONLY

Med Rec #	Date Received	Tracking #	Sender	Initials

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REASON FOR TESTING, CLINICAL DIAGNOSIS AND ICD-10 CODES

PLEASE INCLUDE SECOND PAGE OF CLINICAL INDICATIONS AND/OR ADDITIONAL CLINICAL INFORMATION, MEDICAL RECORDS, PICTURES, FAMILY HISTORY TO AID IN RESULT INTERPRETATION.

❑ Diagnostic ❑ Prenatal ❑ Carrier Screening ❑ Family History ❑ No Family History	**Clinical Description:**	**Phenotypic Description:**	**ICD-10 Codes *(required)*:**	**Prenatal Information:** **LMP:_____ Gestational Age:_____** **G_____P_____Ab_____** **Fetal Sex (if known):_____**

CYTOGENETIC TESTS

Acceptable sample types: Sodium heparin blood (green top, NOT LITHIUM HEPARIN), direct amniotic fluid, CVS, cultured cells, fresh (unfixed) tissue—call 406-444-7532 with questions.

- ❑ **Chromosomes (Karyotype)**
- ❑ **FISH (select at least one of options below)**
  - ❑ **Direct Interphase Aneuploidy (AneuVysion)**
  - ❑ **Other _____**

For POC/CVS samples, cytogenetic studies cannot determine with certainty that a normal female result is not due to maternal cell contamination. We strongly recommend sending a maternal blood sample (3-5mL EDTA) to rule out maternal cell contamination. This maternal sample will be discarded in the event of an abnormal or male result.

- ❑ **Decline Maternal Cell Contamination Studies**

MOLECULAR GENETIC TESTS

Acceptable sample types: EDTA blood (purple top), cultured cells, fresh/frozen tissue—call 406-444-7532 with questions.

- ❑ **Angelman/Prader-Willi (AS/PWS) Methylation**
- ❑ **Beckwith-Wiedemann Syndrome (BWS)**
  - ❑ **KCNQ1OT1 & H19 Methylation**
  - ❑ **Reflex to UPD11 if positive (parent samples required)**
- ❑ **Fragile X Syndrome (FMR-1)**
- ❑ **Hemochromatosis (HFE) Mutations (C282Y / H63D)**
- ❑ **Huntington Disease (HTT) Mutation**
- ❑ **Maternal Cell Contamination (recommended for CVS)**
- ❑ **Myotonic Dystrophy**
- ❑ **Russell-Silver Syndrome (RSS)**
  - ❑ **H19 Methylation**
  - ❑ **UPD7 (parent samples required)**
- ❑ **Spinal Muscular Atrophy (SMA)**
- ❑ **Thrombophilia Gene Polymorphism Panel**
  - ❑ **Factor V Leiden** ❑ **Prothrombin** ❑ **MTHFR**
- ❑ **X-Chromosome Inactivation**
- ❑ **Uniparental Disomy Screen (parent samples required)**
  Select Chromosome(s) 2, 6, 7, 8, 9, 11, 13, 14, 15, 16, 20, 21

CHROMOSOMAL MICROARRAY (CMA)

Acceptable sample types: EDTA blood (purple top), cultured cells, fresh/frozen tissue—call 406-444-7532 with questions.

- ❑ **Chromosomal Microarray**

NEXT-GENERATION SEQUENCING (NGS)

Acceptable sample types: EDTA blood (purple top), cultured cells, fresh/frozen tissue —call 406-444-7532 with questions.

Gene Panels:

- ❑ **Developmental delay, Intellectual disability, Autism**
- ❑ **Epilepsy**
- ❑ **Neuromuscular**
- ❑ **Charcot-Marie-Tooth**
- ❑ **Aortopathy (Marfan syndrome, Loyes-Dietz syndrome, etc.)**
- ❑ **Cardiac arrhythmia including Long QT syndrome**
- ❑ **Noonan Syndrome**
- ❑ **Hearing Loss**

- ❑ **Order by Clinical Indication:**
- ❑ **Specify Gene(s):**

KNOWN FAMILIAL VARIANT STUDIES

Acceptable sample types: EDTA blood (purple top), cultured cells, fresh/frozen tissue—call 406-444-7532 with questions.

- ❑ **Copy Number Analysis**
- ❑ **Sequence Variant**

Proband Name_____

Relationship to Proband_____

Variant Description_____

If proband studies were not performed at Shodair, please include a copy of the proband report

Specific Test Instructions (Reflex testing, STAT, etc.)

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CLINICAL INDICATIONS

Perinatal history
(Please check all the apply)
- ❑ Prematurity
- ❑ Intrauterine growth retardation
- ❑ Oligohydramnios
- ❑ Polyhydramnios
- ❑ Cystic hygroma / increased NT

Growth
- ❑ Failure to thrive
- ❑ Growth retardation / short stature
- ❑ Overgrowth
- ❑ Macrocephaly
- ❑ Microcephaly

Physical/Cognitive Development
- ❑ Fine motor delay
- ❑ Gross motor delay
- ❑ Speech delay
- ❑ Intellectual disability
- ❑ Learning disability
- ❑ Developmental regression

Behavioral
- ❑ Autism spectrum disorder
- ❑ Autistic features
- ❑ Obsessive-compulsive disorder
- ❑ Stereotypic behaviors
- ❑ Other psychiatric symptoms

Craniofacial/Ophthalmalogic/Auditory
- ❑ Cataracts
- ❑ Cleft lip/palate
- ❑ Coloboma of eye
- ❑ CPEO (opthalmoplegia)
- ❑ Ptosis
- ❑ Blindness
- ❑ Optic atrophy
- ❑ Retinitis pigmentosis
- ❑ Hearing loss
- ❑ Ototoxicity (aminoglycoside-induced)
- ❑ External ear malformation
- ❑ Facial dysmorphism

  Describe: _____

Cardiac/congenital heart malformations
- ❑ ASD
- ❑ VSD
- ❑ Coactation of aorta
- ❑ Hypoplastic left heart
- ❑ Tetralogy of Fallot
- ❑ Cardiomyopathy
- ❑ Arrhythmia/conduction defect
- ❑ Other: _____

Skin, Hair, & Nail Abnormalities
- ❑ Abnormal nails _____
- ❑ Abnormal pigmentation _____
- ❑ Abnormal connective tissue _____
- ❑ Blistering
- ❑ Ichthyosis
- ❑ Skin tumors/Malignancies
- ❑ Other: _____

Brain Malformations/abnormal imaging
- ❑ Agenesis of the corpus callosum
- ❑ Holoprosencephaly
- ❑ Lissencephaly
- ❑ Cortical dysplasia
- ❑ Heterotopia
- ❑ Hydrocephalus
- ❑ Brain atrophy
- ❑ Periventricular leukomalacia
- ❑ Hemimegalencephaly
- ❑ Abnormalities of basal ganglia
- ❑ Other: _____

Neurological/Muscular
- ❑ Ataxia
- ❑ Chorea
- ❑ Dystonia
- ❑ Hypotonia
- ❑ Hypertonia
- ❑ Seizures (type: _____)
- ❑ Spasticity
- ❑ Exercise intolerance/easy fatigue
- ❑ Muscle weakness
- ❑ Stroke/stroke-like episodes
- ❑ Recurrent headache/migraine

Gastrointestinal
- ❑ Gastroschisis/omphalocele
- ❑ Pyloric stenosis
- ❑ Tracheoesophageal fistula
- ❑ Delayed gastric emptying
- ❑ Eosinophilic esophagitis
- ❑ Gastrointestinal reflux
- ❑ Recurrent vomiting
- ❑ Chronic diarrhea
- ❑ Constipation
- ❑ Chronic intestinal pseudo-obstruction
- ❑ Hirschsprung disease
- ❑ Hepatic failure
- ❑ Elevated transaminases

Skeletal/Limb abnormalities
- ❑ Contractures
- ❑ Club foot
- ❑ Polydactyly
- ❑ Syndactyly
- ❑ Scoliosis
- ❑ Vertebral anomaly
- ❑ Other: _____

Genitourinary abnormalities
- ❑ Ambiguous genitalia
- ❑ Hypospadias
- ❑ Hydronephrosis
- ❑ Undescended testis
- ❑ Kidney malformation
- ❑ Renal agenesis
- ❑ Renal tubulopathy
- ❑ Other: _____

Endrocrine
- ❑ Diabetes mellitus
  - ❑ Type I
  - ❑ Type II
- ❑ Hypothyroidism
- ❑ Hypoparathyroidism
- ❑ Pheochromocytoma/paraganglioma

Metabolic
- ❑ Ketosis
- ❑ Lactic academia/high CSF lactate
- ❑ Elevated pyruvate
- ❑ Elevated alanine
- ❑ Organic aciduria
- ❑ Low plasma carnitine
- ❑ CPK abnormalities

Hemotologic/Immunologic
- ❑ Recurrent fever
- ❑ Anemia/neutropenia/pancytopenia
- ❑ Immunodeficiency Type: _____
- ❑ Other: _____