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

Shodair Lab Number

GENETICS LABORATORY TEST REQUEST FORM

PATIENT INFORMATION

Last Name: _____ **First Name:** _____ **MI:** _____ **DOB:** _____

Sex Assigned at Birth: ☐ Male ☐ Female ☐ _____ **Gender Identity:** ☐ Male ☐ Female ☐ Non-binary ☐ _____

Address: _____

City, State, Zip: _____

Phone: _____ **Email**: _____

Ethnicity (select all that apply):

- ☐ Caucasian
- ☐ Asian
- ☐ Hispanic
- ☐ African American
- ☐ Ashkenazi Jewish
- ☐ Hutterite
- ☐ American Indian
- ☐ Other _____

ORDERING HEALTH CARE PROFESSIONAL & AUTHORIZATION

Name: _____ **NPI #:** _____

Address: _____ **City, State, Zip:** _____

Telephone:(_____)_____ **Fax**:(_____)_____ **Referring Facility:**_____

Additional Reports To: _____

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:** _____

ADDITIONAL FAMILY SAMPLES (EDTA BLOOD/saliva)

Last Name: _____ First Name: _____

DOB: _____ Relationship: _____ Affected: ☐ Yes ☐ No

Date of Collection: _____ Source: _____

Last Name: _____ First Name: _____

DOB: _____ Relationship: _____ Affected: ☐ Yes ☐ No

Date of Collection: _____ Source: _____

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 # :_____

PREAUTHORIZATION IS REQUIRED FOR ALL MOLECULAR TESTING

Has preauthorization been completed?

❑ **YES** Preauthorization completed & approved, or is not required.
Please include a copy of the approval letter with this form.

❑ **NO** Preauthorization has **not** been completed.
Please assist with preauthorization. Please provide visit note for medical necessity, ICD10, copy of front & back of insurance card, and demographics.

Please designate a contact for preauthorization updates:

Name:_____

Phone/email: _____

INSURANCE BILLING

Please provide a copy of front & back of card

Name of policy holder: _____

Policy holder DOB: _____ **Relationship:** _____

Name of Ins. Co: _____

Ins. Co. Policy #: _____

Ins. Co Phone: _____

MEDICAID / MEDICARE

Name of policy holder: _____

Policy holder DOB: _____ **Medicaid/Medicare # :** _____

Passport ID: _____ **Phone**: _____

Address: _____

City, State, Zip: _____

INSTITUTIONAL BILLING

Institution: _____

Address: _____

City, State, Zip: _____

Phone: _____ **Fax**: _____

Billing Contact: _____

SELF PAY

Please visit our website for the Self-Pay Patient Agreement & contact us at 406-444-7532 for service information and cost.

Name of responsible party: _____

Relation to patient: _____ **Phone:** _____

Received	Date Received	Tracking #	Sender	Initials

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Call 406-444-7532 with questions.

Patient Name: _____ **DOB:** _____

REASON FOR TESTING, CLINICAL DIAGNOSIS AND ICD-10 CODES

Please include page 3 and/or any additional clinical information (medical records, pictures, family history) to aid in result interpretation.

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

PRENATAL TESTING

Acceptable sample: Direct amniotic fluid, CVS, cultured cells, fresh (unfixed) tissue

☐ **Direct Interphase Aneuploidy FISH (AneuVysion, Chr 13,18,21,X,Y). Reflex to Microarray if Negative. Reflex to Karyotype if Positive.**

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**

CYTOGENETIC TESTS

Acceptable sample: Sodium heparin blood (NOT LITHIUM HEPARIN), direct amniotic fluid, CVS, cultured cells, fresh (unfixed) tissue

☐ **Chromosomes (Karyotype)**
☐ **Direct Interphase Aneuploidy FISH (AneuVysion, Chr 13,18,21,X,Y)**
☐ **Custom FISH:** _____

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**

CHROMOSOMAL MICROARRAY (CMA)

Acceptable sample: EDTA blood, cultured cells, fresh/frozen tissue

☐ **Chromosomal Microarray**

KNOWN FAMILIAL VARIANT STUDIES

Acceptable sample: EDTA blood, cultured cells, fresh/frozen tissue

☐ **Copy Number Analysis**
☐ **Sequence Variant**

Proband Name: _____

Relation to Proband: _____

Variant Description: _____

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

MOLECULAR GENETIC TESTS

Acceptable sample: EDTA blood, cultured cells, buccal/saliva, fresh/frozen tissue

☐ **Angelman/Prader-Willi (AS/PWS) Methylation**
☐ **Beckwith-Wiedemann Syndrome (BWS)**
  ☐ **KCNQ1OT1 & H19 Methylation**
  ☐ **Reflex to UPD11 if positive (parent samples required)**
☐ **Chromosome 14 Methylation (Temple/Kagami-Ogata S.)**
☐ **Duchenne Muscular Dystrophy Deletion/Duplication**
☐ **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)**
☐ **X-Chromosome Inactivation**
☐ **Uniparental Disomy Screen (parent samples required)**
Select Chromosome(s) 2, 6, 7, 8, 9, 11, 13, 14, 15, 16, 20, 21

NEXT-GENERATION SEQUENCING (NGS) : Gene Panels

Acceptable sample: EDTA blood, cultured cells, buccal/saliva, fresh/frozen tissue

☐ **Aortopathy (Marfan syndrome, Loeys-Dietz syndrome, etc.)**
☐ **Cardiac arrhythmia including Long QT syndrome**
☐ **Charcot-Marie-Tooth**
☐ **Epilepsy**
☐ **Hearing Loss**
☐ **Neurodevelopmental Disorder, first tier**
☐ **Neuromuscular**
☐ **Noonan Syndrome**

☐ **Order by Clinical Indication/Specify Gene(s):**

WHOLE EXOME SEQUENCING (WES)

Visit our website for the WES consent & test request forms, or call us at 406-444-7532.

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
- ☐ Shortened fetal long bones
- ☐ Choroid plexus cyst
- ☐ Ventriculomegaly
- ☐ Echogenic bowel
- ☐ Fetal pyelectasis/fetal renal pelvic dilatation
- ☐ Single umbilical artery (SUA)
- ☐ Maternal diabetes mellitus

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
- ☐ Polycystic kidneys
- ☐ Multicystic kidneys
- ☐ Other: _____

Endocrine
- ☐ 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
- ☐ Elevated CPK
- ☐ Hypoglycemia

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