Shodair Children's Hospital
Genetics Laboratory
2620 Shodair Dr. Helena, MT 59601
Phone (406) 444-7532 Fax (406) 444-1022

Shodair Lab Number

GENETICS LABORATORY WHOLE-EXOME SEQUENCING 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 ☐ Ashkenazi Jewish
☐ Asian ☐ Hutterite
☐ Hispanic ☐ American Indian
☐ African American ☐ 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:** _____

INFORMED CONSENT *Informed consent is required to be completed prior to testing*

Informed consent is required for each family member participating in whole-exome sequencing. Please complete the informed consent section on page 2 of this form to indicate whether you would like Shodair to provide whole-exome sequencing consenting services or if informed consent has already been obtained.

SAMPLE INFORMATION

☐ Whole Blood (≥3mL)
☐ Saliva/Buccal Cells
☐ Extracted DNA (≥10ug)
Source: _____

Date of Collection: _____
Reference #: _____

ADDITIONAL FAMILY SAMPLES (EDTA Whole Blood/Buccal Cells) *Family samples are required for whole-exome sequencing*

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: _____

PRIOR AUTHORIZATION IS REQUIRED FOR WHOLE-EXOME SEQUENCING

☐ Prior authorization has been completed & approved, or is not required. Please include a copy of the approval letter with this form.

☐ Prior authorization has **not** been completed. **Please assist with prior authorization.** You must provide a letter of medical necessity and/or clinical records, ICD-10 codes, copy of front & back of insurance card, and demographic information for our office to assist with prior authorization.

Designated contact for prior authorization 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 BILLING

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 contact us at 406-444-7532 for service information, cost and a copy of our Self-Pay Patient Agreement.

Name of responsible party: _____
Relation to patient: _____ **Phone:** _____
Email: _____

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Call 406-444-7532 with questions.

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Patient Name: _____ **DOB:** _____

REASON FOR TESTING, ICD-10 CODES, AND CLINICAL DIAGNOSIS

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

Reason for Testing	ICD-10 Codes (required)	Clinical Description	Phenotypic Description
☐ Diagnostic ☐ Prenatal ☐ Carrier Screening ☐ Family History ☐ No Family History			

Additional Information

WHOLE-EXOME SEQUENCING *(WES)*

Acceptable sample: EDTA blood, saliva/buccal cells, extracted DNA

☐ **Rapid Infant Whole-Exome Sequencing** (does **not** include analysis for secondary findings)

☐ **Reflex to Full Whole-Exome Sequencing** (can opt-in to analysis for secondary findings)

☐ **Whole-Exome Sequencing** (can opt-in to analysis for secondary findings)

Specific Test Instructions

WHOLE-EXOME SEQUENCING INFORMED CONSENT

Informed consent is required for each family member participating in whole-exome sequencing prior to initiating testing. Shodair can provide consenting services to your patient and each participating member at no cost.

☐ Informed consent has **not** been completed. **Please contact the patient/the patient's family to provide whole-exome sequencing consenting services.** You must complete the below section to provide demographic and contact information for each family member participating in whole-exome sequencing.

☐ Informed consent has been completed. The patient has completed the Shodair Patient Whole-Exome Sequencing Informed Consent form, and each family member participating in whole-exome sequencing has completed the Shodair Family Member Whole-Exome Sequencing Consent form. **You must provide a copy of these signed consent forms with this order.**

Family Member Name	DOB	Relationship to Patient	Phone Number	Best Time to Contact

What is whole-exome sequencing?

Whole-exome sequencing (*WES)* can be used to help diagnose one or more hereditary conditions. The risks and benefits of this test are explained in the *WES* Informed Consent form so that the patient can make an informed decision about whether to proceed with this test. Participation in *WES* is voluntary. Genetic counseling is required prior to, as well as following, this complex test.

Whole-exome sequencing is a complex test that looks at a large number of genes simultaneously (approximately 20,000) and is designed to identify genetic changes in the DNA that may cause disease or are important to the patient's health in other ways. Genetic disorders are caused by changes in the DNA sequence that affect the ability of a gene to function. Through *WES*, thousands of DNA variants are detected. Some variants are disease-causing while others are harmless or have an uncertain effect. This process involves analyzing genes that have been previously associated with human disease, known as characterized genes. Additionally, many human genes have *not* been associated with an underlying genetic condition; these are known as novel genes.

Most changes that cause disease affect the portions of our genes called exons. The exons of a gene contain the genetic information the body uses to make proteins, which are molecules that carry out all the essential functions in the body. The DNA within the exons of all the genes is collectively called the exome. *WES* is a test that looks for disease-causing changes in any gene that may be related to the patient's underlying clinical presentation.

Samples from other family members may be studied to help interpret the results. In the case of duos and trios, results are only reported on the patient's sample. Family members' results are *not* analyzed separately from the patient's, with the exception of secondary findings described in the *WES* Informed Consent form. Shodair performs this analysis with the expectation that family members are not affected; if a family member is suspected of also sharing the condition, then Shodair Genetics Laboratory should be notified.

WES interpretation and analysis is significantly enhanced by a complete clinical history. For informative results and the highest likelihood of a conclusive diagnosis, it is critical that the ordering provider includes all relevant clinical and family history.

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Patient Name: _____ **DOB:** _____

CLINICAL INDICATIONS *(Please check all the apply)*
Please include page 3 and/or any additional clinical information (medical records, pictures, family history) to aid in result interpretation.

Perinatal history
- ☐ 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/Ophthalmologic/Auditory
- ☐ Cataracts
- ☐ Cleft lip/palate
- ☐ Coloboma of eye
- ☐ CPEO (ophthalmoplegia)
- ☐ Ptosis
- ☐ Blindness
- ☐ Optic atrophy
- ☐ Retinitis pigmentosis
- ☐ Hearing loss
- ☐ Ototoxicity (aminoglycoside-induced)
- ☐ External ear malformation
- ☐ Facial dysmorphism
  Describe: _____

Cardiac/Congenital Heart Malformations
- ☐ ASD
- ☐ VSD
- ☐ Coarctation 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 acidemia/high CSF lactate
- ☐ Elevated pyruvate
- ☐ Elevated alanine
- ☐ Organic aciduria
- ☐ Low plasma carnitine
- ☐ Elevated CPK
- ☐ Hypoglycemia

Hematologic/Immunologic
- ☐ Recurrent fever
- ☐ Anemia/neutropenia/pancytopenia
- ☐ Immunodeficiency type: _____
- ☐ Other: _____

Additional Clinical Indications