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

Shodair Lab Number

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. For patients receiving consenting services through Shodair Genetics, I confirm that I am responsible for results disclosure.

Signature of Ordering Provider: _____ **Date:** _____

ADDITIONAL FAMILY SAMPLES (EDTA Whole Blood or Saliva/Buccal Cells)

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)
☐ Saliva/Buccal Cells
☐ Extracted DNA (≥10ug)
Source:_____

Date of Collection: _____

Reference #: _____

☐ Inpatient ☐ Outpatient

INFORMED CONSENT (REQUIRED)

Informed consent is required for the patient and each family member participating in whole exome sequencing prior to initiating testing. Please complete the Informed Consent section on page 2 of this form.

PRIOR AUTHORIZATION IS REQUIRED FOR WHOLE EXOME SEQUENCING

☐ Prior authorization has been completed & approved, or is not required. **I agree to include a copy of the prior authorization approval letter from the insurance company with this form.**

☐ Prior authorization has **not** been completed. **I would like assistance with prior authorization. I agree to provide clinical records and/or a letter of medical necessity, ICD-10 codes, a copy of the front & back of the insurance card, and demographic information with this form.**

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: _____ **Relation to patient:** _____

Name of Ins. Co: _____

Ins. Co Policy #: _____

Ins. Co Phone: _____

INSTITUTIONAL BILLING

Institution: _____

Address: _____

City, State, Zip: _____

Phone: _____ **Fax:** _____

Billing Contact: _____

MEDICAID / MEDICARE BILLING

Name of policy holder: _____

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

Address: _____

City, State, Zip: _____

SELF-PAY

Please contact us at 406-444-7532 for a copy of the Patient Self-Pay Agreement.

Name of responsible party: _____

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

Email: _____

Received	Date Received	Tracking #	Sender	Initials

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Call 406-444-7532 with questions.

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WHOLE EXOME SEQUENCING TEST REQUEST FORM

Patient Name: _____ **DOB:** _____

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

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 Indication
☐ Diagnostic ☐ Prenatal ☐ Carrier Screening ☐ Family History ☐ No Family History		

WHOLE EXOME SEQUENCING (WES)

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

☐ **Trio Whole Exome Sequencing (Patient + two additional family members)**

☐ **Duo Whole Exome Sequencing (Patient + one additional family member)**

☐ **Singleton Whole Exome Sequencing (Patient only)**

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

☐ **Reflex to Full Whole Exome Sequencing**

Specific Test Instructions

WHOLE EXOME SEQUENCING INFORMED CONSENT (REQUIRED)

Informed consent is required for the patient and each family member participating in whole exome sequencing prior to initiating testing. Please refer to our website or contact us at 406-444-7532 to obtain copies of the Patient Whole Exome Sequencing Consent Form and the Family Member Whole Exome Sequencing Consent Form. Shodair Genetics would be happy to provide consenting services to your patient and each participating family member. If you would like assistance with the consenting process, please indicate below and Shodair's scheduling team will contact the family to complete registration paperwork and coordinate a telemedicine visit with one of our genetic counselors. During this visit, the genetic counselor will review the patient's presentation and family history, and will discuss whole exome sequencing in detail. Please be aware that there will be a cost associated with this visit. Please mark your selection below. If you have any questions, please contact our office at 406-444-7532.

☐ Informed consent has **not** been completed. **I would like to refer the patient for a telemedicine visit with a genetic counselor to complete the whole exome sequencing consenting process. I agree to complete the below section providing demographic and contact information for each family member participating in whole exome sequencing if applicable. I agree to provide clinical records including recent visit notes and diagnostic test results along with this test request form.**

☐ Informed consent has been completed. The patient has completed the Patient Whole Exome Sequencing Consent Form, and each participating family member has completed the Family Member Whole Exome Sequencing Consent Form. **I agree to provide a copy of these signed consent forms along with this test request form.**

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

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WHOLE EXOME SEQUENCING TEST REQUEST FORM

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