THE UNIVERSITY OF OKLAHOMA HEALTH SCIENCES CENTER

GENETICS LABORATORY
WHOLE EXOME SEQUENCING (WES) FORM

PLEASE COMPLETE ALL FORMS AND
SEND WITH SAMPLE(S)

Ship To: O'Donoghue Research Bldg
1122 NE 13 Street, Suite 1400
Oklahoma City, OK 73104
Phone: 405-271-3589
Fax: 405-271-7117
After hours phone: 405-496-9514
www.genetics.ouhsc.edu

REFERRING PHYSICIAN/FACILITY

Physician Name _____

NPI_____

Phone(__)_____ Fax(__)_____

Genetic Counselor_____ Phone(__)_____

Laboratory/Institution_____

Address_____

City_____ State_____ Zip_____

Phone_(__)_____ Fax (__)_____

PATIENT INFORMATION

Patient Name (last,first,m.)_____

Parent Name (if patient is minor)_____

DOB_____ SSN_____ MRN_____

Sex: □ Male □ Female □ Ambiguous □ Other □ Inpatient □ Outpatient

Ethnicity of patient (check all that apply)
□ African-American □ Asian □ Caucasian/NW European □ E. Indian
□ Hispanic □ Jewish-Ashkenazi □ Jewish-Sephardic □ Native American
□ Native Hawaiian/Other Pacific Islander □ Other_____

Patient's Address_____

City_____ State_____ Zip _____

SPECIMEN INFORMATION

□ **Peripheral Blood** 3-5 cc larged EDTA tube (purple top), mix well keep at room temperature or cooler, do not freeze.

□ **Isolated DNA** qty 20ug Specimen Collection Date _____

If shipping materials via Fedex/UPS, packages can only be accepted Mon-Fri 9:00 AM to 5:00 PM. Our facilities are not accessible by delivery personnel on weekends or after hours. For courier service in Oklahoma City metro area call Rapid Transit 793-1122 for specimen pickup.

TEST OPTIONS

□	WES– Proband Only TAT 90 days	*CPT: 81415*	□	WES- Trio TAT 90 days	*CPT: 81416x2*

FAMILY MEMBER SAMPLE INFORMATION

Biological Mother:	_____	DOB: _____	□ Symptomatic	□ Asymptomatic	**All samples related to proband must be received within 10 days of proband
Biological Father:	_____	DOB: _____	□ Symptomatic	□ Asymptomatic	
Other:	_____	DOB: _____	□ Symptomatic	□ Asymptomatic	
	Relationship to proband: _____	Total # of samples sent	_____		

****A COMPLETED PATIENT/PROBAND CLINICAL INFORMATION FORM AND CLINIC NOTES MUST ACCOMPANY THE SAMPLES.**

PATIENT/GUARDIAN CONSENT– Please read the Informed Consent document before signing

I have read the Informed Consent document and I give OUHSC Genetics Lab my permission to perform the genetic testing listed above. I also give permission for [my/my child's] specimen and clinical information to be used in de-identified studies at OUHSC Genetics Lab to improve genetic testing and for publication, when appropriate. [My/my child's] name or other personal identifying information will not be used in or linked to the results of any studies and publications. I also give OUHSC Genetics Lab permission to inform me or [my/my child's] health care provider in the future about research opportunities, including treatments for the condition in [my/my child's] family.

□ Check this box to opt-**out** of receiving secondary findings.
□ Check this box to opt-**out** of research.

_____ Patient/Guardian Signature

_____ Date

ADDITIONAL REPORT

Physician/Facility_____

Phone_(___)_____ Fax_(___)_____

Address_____

GENETICS LABORATORY USE ONLY

Laboratory Number_____

Date/Time/Location of Pick-Up or Delivery_____

Initials_____ Check-in_____ Previous Lab Number_____

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Patient Name Last_____ First_____ MI_____

CONSENT AND AUTHORIZATION

I understand that my health care provider has ordered whole exome sequencing for myself/my child: _____
Sign Here

INFORMED CONSENT

What are potential results from this test?

There are three possible result outcomes:

1) Positive Result: A positive result indicates that a genetic change has been identified, and it explains the potential cause of [my/my child's] genetic condition or indicates that [I am/my child is] at increased risk to develop a disorder in the future. **It is possible to test positive for more than one genetic change.**
2) Negative Result: A negative result means that no disease-causing genetic change was identified in the test performed. **It does NOT guarantee that [I/ my child] will be healthy or free from genetic disorders or medical conditions.** If [I/my child] test negative for a genetic change known to cause a specific condition in other members of the family, a diagnosis of the same genetic disorder in [me/my child] due to this specific change has been ruled out.
3) Variant of Uncertain Significance (VUS): A variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with a genetic condition. **A VUS is not the same as a positive result** and does not specify whether [I am/my child is] at increased risk for a genetic condition. The variant may be a normal genetic change found in the human population, or it could be disease-causing. Further analysis may be warranted and include testing both parents (if not already tested) as well as other family members such as: siblings, grand parents, or aunts and uncles. Detailed medical records or additional information may be requested to re-classify this result .

Additional Positive Results

1) Secondary Findings: Secondary findings are genetic changes identified in genes that are unrelated to the individual's reported clinical features. **The American College of Medical Genetics and Genomics (ACMG) has recommended that secondary findings identified in a specific subset of medically actionable genes associated with various inherited disorders be reported for all probands undergoing whole exome sequencing.** Refer to the latest version of the *ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing* for a complete list of the genes and associated genetic disorders.
2) Incidental Findings: In rare instances, this test may reveal an unexpected, yet important genetic change that is not directly related to the reason for ordering this test. This test may tell me about the risk for another genetic condition [I am/my child is] not aware of, or it may indicate differences in the number or rearrangements of sex chromosomes. Information may be disclosed to the ordering health care provider if it will likely impact [your/ your child's] medical care.

What will be reported

1) For the proband: All known and/or expected disease-causing genetic changes identified in the coding exons of the genes correlating with the submitted phenotype or included in the recommended gene list issued by the ACMG.
2) For relatives: The presence or absence of any secondary findings reported for the proband will be provided for all relatives.

Limitations and Risks

1) Disease-causing genetic changes may be present in a portion of the gene not covered by this test and therefore are not reported. The absence of reportable secondary findings for any particular gene does not mean there are no disease-causing genetic changes.
2) Pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified or reported.
3) Only changes in the genetic sequence will be reported in secondary findings. Larger deletions/duplications, abnormal methylations, triplet repeats and other expansions, or other variants not routinely identified by whole exome sequencing will not be reported.
4) This test has the potential to identify non-paternity and consanguinity (relatedness) between the proband's parents. It may be necessary to report these results to the health care provider who ordered the test if it will affect medical management of the proband.
5) Inaccurate results may be reported due to mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, or the presence of mosaicism (change not present in every cell).

Interpretation

1) Results will be interpreted with available information in the medical literature, research databases, and scientific databases. Due to the change in medical/ scientific knowledge and published studies, new information that becomes available may replace or build upon the information utilized in the interpretation of [my/my child's] results.
2) Providers can contact the laboratory at any time to discuss the classification or re-classification of an identified genetic change.
3) [My/my child's] health care providers and I may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find updated information about the interpretation of [my/my child's] genetic change(s).
4) Results will be released in a single report in the proband's name. Additional reports will NOT be released in the proband's family members' names if a trio is submitted.
5) Occasionally, an additional sample may be needed if the initial specimen is not adequate for analysis.

Patient Confidentiality and Genetic Counseling

1) It is strongly recommended that [I/my child] receive genetic counseling before and after having this test. I can find a genetic counselor in my area at www.nsgc.org.
2) Further testing or consultations may be necessary based on results.
3) To maintain confidentiality, the test results will only be released to the referring health care provider, the ordering laboratory, me, other health care providers involved in [my/my child's] diagnosis and treatment, or to others as entitled by law.
4) The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand I can visit www.genome.gov/10002077.

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Genetics Laboratory

Patient/Proband Clinical Information Form

Last Name: _____First: _____ MI: ___ DOB: _____

Primary Indications for Testing

- ❑ Multiple Congenital Anomalies
- ☐ Developmental Delays
- ❑ Neurological/Muscular Disorder

Previous Testing

- ❑ Karyotype/FISH
- ❑ CMA
- ❑ Newborn Screen Result
- ❑ Other Results_____

Family History (provide pedigree)

- ❑ Consanguinity
- ❑ Family History of Genetic Disease/Disorder

Development & Cognition

- ❑ Autism Spectrum
- ❑ Fine Motor Delays
- ❑ Global Delay
- ❑ Gross Motor Delays
- ❑ Intellectual Delays
  - □ Mild
  - □ Moderate
  - □ Severe
- ❑ Learning Delays
- ❑ Speech Delay

Perinatal History

- ❑ IUGR / SGA

Growth

- ❑ Failure to thrive
- ❑ Macrocephaly
- ❑ Microcephaly
- ❑ Overgrowth/Tall
- ❑ Short stature
- ❑ Other:_____

Craniofacial Anomalies

- ❑ Cleft Lip
- ❑ Cleft Palate
- ❑ Craniosynostosis
- ❑ Dysmorphic Facies
- ❑ Ear Malformation
- ❑ Other:_____

Ear / Hearing Loss (HL)

- ❑ Conductive HL
- ❑ Microtia
- ❑ Sensorineural HL
- ❑ Other:_____

Eye Anomalies

- ❑ Aniridia
- ❑ Congenital Cataract
- ❑ Cortical Blindness/CVI
- ❑ Coloboma
- ❑ Glaucoma
- ❑ Optic Nerve Abnormality
- ❑ Ptosis
- ❑ Retinitis Pigmentosa
- ❑ Other:_____

Pulmonary

- ❑ Diaphragmatic Hernia
- ❑ TE Fistula
- ❑ Other:_____

Cardiac

- ❑ Arrhythmia
- ❑ ASD
- ❑ Cardiomyopathy
- ❑ Coarctation of aorta
- ❑ Dextrocardia
- ❑ Tetralogy of fallot
- ❑ Ventriculomegaly
- ❑ VSD
- ❑ Other:_____

GI

- ❑ Anal Atresia
- ❑ Chronic Obstruction
- ❑ Dysphagia
- ❑ Esophageal Atresia
- ❑ Gastroschisis
- ❑ Hirschsprung Disease
- ❑ Liver Disease
- ❑ Omphalocele
- ❑ Polysplenia
- ❑ Situs Inversus
- ❑ Other:_____

Genitourinary

- ❑ Ambiguous Genitals
- ❑ Cryptochidism
- ❑ Hydronephrosis
- ❑ Hypospadias
- ❑ Kidney Malformation
- ❑ Renal Agenesis
- ❑ Renal Tubulopathy
- ❑ Other:_____

Skeletal

- ❑ Arthrogryposis
- ❑ Club Foot/Feet
- ❑ Contractures
- ❑ Joint Hypermobility
- ❑ Kyphosis
- ❑ Limb Anomaly
- ❑ Osteopenia
- ❑ Pes Planus
- ❑ Polydactyly
- ❑ Scoliosis
- ❑ Skeletal Dysplasia
- ❑ Syndactyly
- ❑ Vertebral anomaly
- ❑ Other:_____

Endocrine

- ❑ Diabetes Insipidus
- ❑ Diabetes Mellitus
- ❑ Hyperthyroidism
- ❑ Hypothyroidism
- ❑ Hyperparathyroidsim
- ❑ Hypoparathyroidism

Hematologic/Immuno

- ❑ Anemia
- ❑ Immunodeficient
- ❑ Neutropenia
- ❑ Pancytopenia
- ❑ Thrombocytopenia
- ❑ Other:_____

Neurological & Muscular

- ❑ Ataxia
- ❑ Brain Anomaly
- ❑ Cerebellar anomaly
- ❑ Chorea/Dystonia
- ❑ Encephalopathy
- ❑ Holoprosencephaly
- ❑ Hydrocephalus
- ❑ Hypertonia
- ❑ Hypotonia
- ❑ Lissencephaly
- ❑ Leukodystrophy
- ❑ Muscle Weakness/Atrophy
- ❑ Peripheral Neuropathy
- ❑ Vermis Hypoplasia
- ❑ Other:_____

Cancer/Tumors

- ❑ Tumor (describe)_____

_____

Age of Onset_____

Skin, Hair & Nails

- ❑ Abnormal Hair
- ❑ Abnormal Nails
- ❑ Hyperpigmentation (describe)_____
- ❑ Hypopigmentation (describe)_____
- ❑ Lipoma
- ❑ Other:_____

Metabolic Abnormalities

- ❑ Hyperammonemia
- ❑ Ketosis
- ❑ Lactic Acidosis
- ❑ Metabolic Acidemia
- ❑ Other:_____

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Genetics Laboratory
Billing Information Form

Patient Name LAST_____ FIRST_____ MI_____

YOU MUST CHOOSE ONE OF THE THREE BILLING OPTIONS LISTED BELOW.
PLEASE FORWARD ALL BILLING QUESTIONS TO DANIELLE OTIS AT DOTIS@OUHSC.EDU OR CALL 405-271-3589 OPT 4
AT THIS TIME WE DO NOT ACCEPT OUT-OF-STATE MEDICAID

PAYMENT OPTION 1-INSTITUTION

INSTITUTION NAME _____

BILLING ADDRESS_____

CITY, STATE, ZIP _____ CONTACT NAME_____

PHONE NUMBER_____ FAX NUMBER_____ CONTACT EMAIL ADDRESS _____

PAYMENT OPTION 2-SELF PAY (PAYMENT MUST BE SENT WITH SAMPLE)

☐ **CREDIT CARD** (CIRCLE ONE) AMEX DISCOVER VISA MASTERCARD AMOUNT TO CHARGE_____

VALID CARD # _____ EXP DATE_____

CVV CODE _____ CARDHOLDER PRINTED NAME_____

BILLING ADDRESS _____ CITY, STATE, ZIP_____

CARDHOLDER SIGNATURE _____

☐ **CHECK #** _____ AMOUNT ENCLOSED_____

PAYMENT OPTION 3-INSURANCE PROVIDE A LEGIBLE COPY OF THE FRONT & BACK OF INSURANCE CARD
PLEASE NOTE: OUR FACILITY WILL CONFIRM COVERAGE AND VERIFY WHETHER OR NOT THE TEST(S) ORDERED ARE COVERED BY YOUR PLAN.
OUR OFFICE CAN ALSO OBTAIN PRE-AUTHORIZATION FROM THE INSURANCE PLAN.

PRIMARY INSURANCE POLICYHOLDER NAME _____ POLICYHOLDER DOB_____

PRIMARY POLICYHOLDER SS# _____ GENDER:○M ○F EMPLOYER_____

RELATIONSHIP TO PATIENT_____ POLICY # _____

GROUP #_____ INSURANCE CO. NAME _____

PHONE _____ CLAIMS ADDRESS _____

CITY, STATE, ZIP_____ INSURANCE AUTH #_____

SECONDARY INSURANCE POLICYHOLDER NAME _____ POLICYHOLDER DOB_____

SECONDARY POLICYHOLDER SS# _____ GENDER:○ M ⊙ F EMPLOYER_____

RELATIONSHIP TO PATIENT_____ POLICY # _____

GROUP #_____ INSURANCE CO. NAME _____

PHONE _____ CLAIMS ADDRESS _____

CITY, STATE, ZIP_____ INSURANCE AUTH #_____

I CONSENT TO HAVE THE TEST(S) LISTED ON THE PREVIOUS PAGE PERFORMED. I AUTHORIZE THE UNIVERSITY OF OKLAHOMA HSC GENETICS LABORATORY TO FURNISH ANY MEDICAL INFORMATION REQUESTED ON MYSELF, OR MY COVERED DEPENDENTS. IN CONSIDERATION OF SERVICES RENDERED, I TRANSFER AND ASSIGN ANY BENEFITS OF INSURANCE TO UNIVERSITY OF OKLAHOMA HSC GENETICS LABORATORY. I UNDERSTAND I AM RESPONSIBLE FOR ANY CO-PAY, DEDUCTIBLES, OR NON-AUTHORIZED SERVICES AND REMAINING BALANCES AFTER INSURANCE REIMBURSEMENT. I UNDERSTAND I AM FULLY RESPONSIBLE FOR PAYMENT OF MY ACCOUNT IF THE UNIVERSITY OF OKLAHOMA HSC GENETICS LABORATORY IS NOT A PARTICIPANT WITH MY HEALTH PLAN OR MY HEALTH PLAN DOES NOT FULLY REIMBURSE MY MEDICAL SERVICES DUE TO LACK OF AUTHORIZATION OR MEDICAL NECESSITY.

PRINTED NAME _____ SIGNATURE_____ DATE _____