Knight Diagnostic Laboratories

Fax: (855) 535-1329
Email: KDLClientServices@ohsu.edu
Shipping: 2525 SW 3rd Ave, Ste 350, Portland, OR 97201
Questions? (855) 535-1522

Exome Requisition

Complete Exome Orders Must Include All of the Following:

- ☑ Include completed Requitision Form (pages 1-2) or an Epic Order
- ☑ Clinical and Phenotypic History Form (pages 3-4)
- ☑ Signed Consent Form (pages 5-8)
- ☑ Fax all forms to KDL Client Services: (855) 535-1329

Exome Tests

Exome Sequencing Trio[1]		
	Code	**Test Name**
☐	2890	Trio

Exome Sequencing Duo[2]		
	Code	**Test Name**
☐	2850	Duo

Exome Sequencing Proband Only		
	Code	**Test Name**
☐	2800	Proband Only

[1]Trio testing requires blood/saliva from two biological relatives in addition to the proband.

[2]Duo testing requires blood/saliva from one biological relative in addition to the proband.

Patient Information

Patient Last Name
Patient First Name
Street Address
City, State, Zip
Phone DOB / /
Fax ☐ Male ☐ Female
ID/MRN #
Hospital In-Patient ☐ Yes ☐ No

Physician Signature Date

Ordering Physician Information

Full Name
NPI
Office/Facility Name
Address
City, State, Zip
Phone Fax
Account #
Notes

Send additional copies of test results to:

Physician Name Physician Phone Fax
Physician Name Physician Phone Fax

Billing Information - Select One Billing Method

Billing is done in accordance with the information provided below and OHSU policy. Appropriate areas must be completed or eferring laboratory/physician will be billed.

☐ Self Pay ☐ Bill Insurance
Attach Copy of Insurance Card or Billing Face Sheet

☐ Bill Referring Provider or Institution
Invoice will be sent to Client Account and Address Listed Above

Primary Insurance Name
Primary Policy #
Primary Group #
Preauthorization #

Relation to Insured ☐ Medicaid ☐ Medicare
☐ Self ☐ Child ☐ Spouse ☐ Other_____

Secondary Insurance Name
Secondary Policy #
Secondary Group #
Preauthorization #

Relation to Insured ☐ Medicaid ☐ Medicare
☐ Self ☐ Child ☐ Spouse ☐ Other_____

Clinical Information

Specimen Type
☐ Saliva
☐ DNA from* _____
☐ Whole Blood
☐ Other _____

Date of Specimen Collection
/ /

Time of Specimen Collection
: :

ICD-10 (required)

* DNA extraction must occur in a CLIA-certified lab or a lab meeting equivalent requirements.

Diagnosis

☐ African American
☐ Alaska Native
☐ Asian
☐ Caucasian / Non-Hispanic
☐ Hispanic American
☐ Jewish, Ashkenazi
☐ Jewish (Other)
☐ Native American Indian
☐ Other:_____

Result Release

Results will be immediately available to the patient unless you mark the box below

☐ Do not release (I reasonably believe that an Information Blocking exception applies)

Comments / Requests:

CLINICAL AND PHENOTYPIC HISTORY FORM

Indication for Testing	Known Familial Mutations
☐ Family History, Mutation Known: ☐ Yes* ☐ No *If Yes, please complete KNOWN FAMILIAL MUTATIONS ☐ Symptomatic ☐ Possible Diagnosis ☐ Definite Diagnosis ☐ Carrier Testing ☐ Presymptomatic Testing ☐ Prenatal Testing ☐ Predispositional Testing ☐ Other (Please Specify) Pregnancy: LMP:_____ GMP:_____	Please call Client Services at 1-800-KDL-1LAB and provide clinical report if proband testing was performed outside of OHSU. Patient Status: ☐ Symptomatic ☐ Asymptomatic Name of Gene: _____ Variants to be tested: _____ Name of Proband:_____ Relationship to Proband: _____ OHSU Sample # of Proband:_____ * If proband testing was performed outside of OHSU, please provide clinical report.

Family History

Please draw or attach patient's three generation pedigree

Please include any additional relevant family history information:

Phenotypic History

Instructions: Please provide the patient's phenotypic history by filling either Normal or an abnormality in a sub-category. In the space provided below each section, add any additional relevant clinician notes for that category. On the following pages is a list of phenotype terms to assist in completing the requisition form.

	Abnormality - Please provide detailed description	**Normal**
Nervous system	☐	☐
Voice or speech	☐	☐
Behavioral or neuropsychiatric	☐	☐
Growth and development	☐	☐
Metabolic system	☐	☐
Head, facial, ears or neck	☐	☐
Eye or vision	☐	☐
Skin, nails or hair	☐	☐
Skeletal system or limb extremities	☐	☐
Musculature and soft tissues	☐	☐
Connective or adipose tissues	☐	☐
Genitourinary system	☐	☐
Immune system	☐	☐
Blood and/or bloodforming tissues	☐	☐
Cardiovascular system	☐	☐
Endocrine system	☐	☐
Respiratory system	☐	☐
Abdomen or abdominal organs	☐	☐
Chest or thoracic	☐	☐
Prenatal development or abnormal birth	☐	☐
Neoplasm	☐	☐
Other	☐	☐

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Exome Sequencing Consent Form

The purpose of this form is to provide comprehensive information about Exome Sequencing. The following information should be used as a guide to provide informed consent to the patient and/or patient's family. A qualified Healthcare Provider must order testing.

For the purposes of this consent, "I," "my," and "your" will refer to you or your child, if your child is the person for whom the Healthcare Provider has ordered testing.

Exome Testing Options:
Please Note: Testing biological parents improves the analysis of the test and decreases the likelihood of receiving an inconclusive result.

SELECT ONE:

☐ **Proband-Only Testing (Patient Only)**

Previous Genetic Testing (If yes, specify): _____

☐ **Duo Testing (Patient plus One Biological Relative)**

Name: _____ DOB: _____

Relationship to Patient: _____

Affected? (If yes, specify): _____

Previous Genetic Testing (If yes, specify): _____

☐ **Trio Testing (Patient plus Two Biological Relatives)**

Name: _____ DOB: _____

Relationship to Patient: _____

Affected? (If yes, specify): _____

Previous Genetic Testing (If yes, specify): _____

Name: _____ DOB: _____

Relationship to Patient: _____

Affected? (If yes, specify): _____

Previous Genetic Testing (If yes, specify): _____

Overview of Exome Sequencing

- The purpose of this test is to look for genetic changes that might contribute to the cause of your health condition.
- In general, this test is used when your medical history and physical exam strongly suggest a genetic cause.
- This test is performed using Next-Generation Sequencing (NGS) technologies. In addition, Copy Number Variants (CNVs), also known as deletions/duplications, are detected from NGS data.
- A change in your DNA sequence compared to a reference DNA sequence is called a variant.
- This test requires a blood or saliva sample from you. A blood or saliva sample is also required from each biological parent/relative (if applicable).
- Genetic counseling and/or clinical genetics consultation before and after testing is recommended.

Test Results

The results of Exome Sequencing are provided in an individualized, written report transmitted directly to the ordering Healthcare Provider.

The main report may contain one or more of the following:

- Diagnostic Findings (Positive): A variant(s) was identified in a gene or region (for CNVs) interpreted to be responsible for, or contributing to, your condition. This interpretation is based on information available in the medical literature and scientific databases.
- No Diagnostic Findings (Negative): No variants were found that could explain your health condition. This does not rule out a genetic condition. It is possible that there is a genetic variant not found by the test.
- Inconclusive (Variant of Uncertain Significance (VUS)): A variant(s) was identified; however, it is uncertain whether this variant is the cause of your health condition. The impact of the genetic change is unknown due to a lack of (or conflicting) scientific information and therefore classified as a VUS.
- Secondary Findings (Optional): In some instances, this test may reveal a clinically relevant genetic change that is not directly related to your health condition. Please read the description below outlining Secondary Findings and indicate the reporting preference for each individual tested as appropriate.
- Incidental Findings: Very rarely, computational algorithms may incidentally discover a genetic variant that is unrelated to your health condition and is not included on the ACMG Secondary Findings list, but nevertheless may have clinical implications for you. The lab does not actively search for incidental findings; however, if there are medical implications, the Healthcare Provider will be contacted and the findings may be included in your report. There is no option to opt out of incidental findings.

Secondary Findings

Anyone being tested has the option of learning about genetic variants that can make you at higher risk for unrelated, but serious health conditions such as heart disease and certain inherited cancers. The genes included in the Secondary Findings list are considered medically actionable. If you wish to learn of these conditions, the results reported will be on a limited list of genes based on the American College of Medical Genetics (ACMG) recommendations (Miller *et al*. 2023 Genetics in Medicine https://doi.org/10.1016/j.gim.2023.100866 (v3.2)). If you choose to opt-in, all the genes on the Secondary Findings List will be analyzed and there is no option to choose the analysis for less than the complete list of genes.

Genetic changes in medically actionable genes might change the way your doctor treats you. Results might show a higher risk for a disease that either may not happen for many years or may not happen at all. Some of these diseases have no treatment, but they might change how you plan for the future. These results may also impact other family members, who may wish to know or not know about these conditions.

If a Secondary Finding is identified, genetic counseling and/or other medical evaluations may be recommended. These expenses may or may not be covered by your health insurance. This may also affect your ability to obtain certain types of insurance (see Limitations and Risk section).

Limitations and Risks

- This test may not answer all health-related questions for several reasons:
    - In some cases, testing may not identify a genetic variant even if one exists. This may be due to limitations in current medical knowledge or limitations of testing technology.
    - This test cannot accurately sequence repetitive regions, such as trinucleotide repeats.
    - Not all health issues have known genetic causes.
- The meaning of the results may be uncertain. One or more Variant(s) of Uncertain Significance (VUS) may be identified.
- This test may find unexpected changes in a family tree. Testing multiple family members may reveal that familial/biological relationships are not what they were assumed to be. The test may show that a child has a different biological father (non-paternity), that a donor egg or sperm was used for a pregnancy, or that a child has been adopted. If you are aware of any such issues in the family, they should be discussed confidentially with your genetic counselor or ordering Healthcare Provider. If results suggest a different biological relationship, the Healthcare Provider will be contacted.
- This test may show that the biological mother and father are close relatives. This can cause conflicts in some families. If the results suggest relatedness, the Healthcare Provider may be contacted.
- This test may affect your ability to obtain certain types of insurance. Test results will be in the medical record. You and your family may have concerns about genetic discrimination, including health insurance, life insurance, long-term disability insurance, and employment. These concerns should be addressed according to federal and state laws. The Federal Genetic Information Non-Discrimination Act (GINA) prohibits the use of genetic information for discrimination in health insurance and employment.

Exome Reanalysis

- Our understanding of genetic changes will improve over time. It is possible that a more certain interpretation of the identified genetic variants will be possible in the subsequent years following the initial report. For this reason, the lab reviews genetic variants reported as “uncertain significance” periodically. If new information is discovered and it is thought to be important to your health and/or the health of your relatives, your Healthcare Provider will be contacted. Secondary Findings are not included in this reanalysis process.
- A Healthcare Provider can request reanalysis. Please see Whole Exome Reanalysis and Interpretation Services for more information.

Specimen Retention

The Knight Diagnostic Laboratory does not return the remaining sample to individuals or physicians, and does not guarantee the future availability of isolated DNA. Any requests for additional studies must be ordered by the Healthcare Provider and charges will be incurred.

Signatures

I hereby authorize the Knight Diagnostic Laboratories to conduct genetic testing for the Exome Sequencing test as recommended by my physician. Please note that if the patient (proband) is a child, parents or legal guardians may authorize predictive genetic testing for asymptomatic children at risk of childhood-onset conditions. Ideally, the assent of the child should be obtained.

Patient's (Proband) Name: _____

☐ Opting IN to receive Secondary Findings

☐ Opting OUT of receiving Secondary Findings

Signature: _____ Date: _____
Patient or Legal Guardian

Biological Relative's Name: _____

☐ Opting IN to receive Secondary Findings

☐ Opting OUT of receiving Secondary Findings

Signature: _____ Date: _____

Biological Relative's Name: _____

☐ Opting IN to receive Secondary Findings

☐ Opting OUT of receiving Secondary Findings

Signature: _____ Date: _____

Witness Signature: _____ Date: _____