

WHOLE EXOME SEQUENCING (WES) REQUISITION

PATIENT INFORMATION (COMPLETE ONE FORM FOR EACH PERSON TESTED)

Patient Last Name _____ Patient First Name _____ MI _____ Date of Birth (MM / DD / YYYY) _____ / _____ / _____
 Address _____ City _____ State _____ Zip _____ Phone _____
 Accession # _____ Hospital / Medical Record # _____
 Genetic Sex: Female Male Unknown
 Gender identity (if different from above): _____

Note: All reports will be sent via fax except for international recipients.

ORDERING PHYSICIAN

ADDITIONAL REPORTS

Ordering Physician _____	Institution Code _____	Name _____	Name _____
Institution Name _____		Email _____	Email _____
Email (Required for International Clients) _____		Phone _____	Phone _____
Phone _____ Fax _____		Fax _____	Fax _____

Note: Reports will be sent by FAX except for international recipients

PAYMENT (FILL OUT ONE OF THE OPTIONS BELOW)

SELF PAYMENT
 Pay With Sample Bill To Patient

INSTITUTIONAL BILLING

Institution Name _____ Institution Code _____ Institution Contact Name _____ Institution Phone _____ Institution Contact Email _____

INSURANCE
 Do Not Perform Test Until Patient is Aware of Out-Of-Pocket Costs (excludes prenatal testing)

REQUIRED ITEMS	1. Copy of the Front/Back of Insurance Card(s)	2. ICD10 Diagnosis Code(s)	ICD10 Diagnosis Code(s) (Required)
	3. Name of Ordering Physician	4. Insured Signature of Authorization	

Primary Insurance Co. Name _____	Primary Insurance Co. Phone _____	Secondary Insurance Co. Name _____	Secondary Insurance Co. Phone _____
Primary Member Policy # _____	Primary Member Group # _____	Secondary Member Policy # _____	Secondary Member Group # _____
Name of Insured _____	Insured Date of Birth (MM / DD / YYYY) _____	Name of Insured _____	Insured Date of Birth (MM / DD / YYYY) _____
Patient's Relationship to Insured _____	Phone of Insured _____	Patient's Relationship to Insured _____	Phone of Insured _____
Address of Insured _____		Address of Insured _____	
City _____ State _____ Zip _____		City _____ State _____ Zip _____	

By signing below, I hereby authorize Baylor Genetics to provide my insurance carrier any information necessary, including test results, for processing my insurance claim. I understand that I am responsible for any co-pay, co-insurance, and unmet deductible that the insurance policy dictates. If self-pay is selected, I agree to pay for the cost of testing ordered and billed by Baylor Genetics as outlined in the Good Faith Estimate I received. I understand that I am responsible for sending Baylor Genetics any and all payments that I receive directly from my insurance company in payment for this test. Please note, Medicare may not cover certain screening tests.

Patient / Guardian Printed Name _____ Patient / Guardian Signature _____ Date (MM / DD / YYYY) _____ / _____ / _____

STATEMENT OF MEDICAL NECESSITY AND CONSENT TO TERMS & CONDITIONS FOR TEST ORDER (REQUIRED)

This requisition hereby incorporates the Terms and Conditions of the Laboratory Services found at <https://www.baylorgenetics.com/lab-terms-conditions/> or, in the case of international entities, <https://www.baylorgenetics.com/terms-conditions-of-the-laboratory-services-international/>. This test is medically necessary for the risk assessment, diagnosis, or detection of a disease, illness, impairment, symptom, syndrome, or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Physician is authorized by law to order the test(s) requested herein. I confirm that I have provided genetic testing information to the patient, and they have consented to genetic testing.

Physician's Printed Name _____ Physician's Signature _____ Date (MM / DD / YYYY) _____ / _____ / _____

WHOLE EXOME SEQUENCING (WES) REQUISITION

Patient Last Name

Patient First Name

MI

Date of Birth (MM / DD / YYYY)

Genetic Sex

INSTRUCTIONS FOR ORDERING

Any combination of Chromosomal Microarray Analysis (CMA), mtDNA Analysis, or Global MAPS® can be ordered along with a WES test, however the turnaround time for results will differ from exome sequencing. Parental samples are required for Trio WES and Duo WES, and optional for Proband WES.

Please contact the laboratory if placing test orders for different members of the family other than the proband or their parents.

TRIO WES TEST OPTIONS

- 1600 Trio Whole Exome Sequencing
- 1532 Trio Whole Exome Sequencing + Comprehensive mtDNA Analysis
- 1722 Rapid Trio Whole Exome Sequencing
- 1533 Rapid Trio Whole Exome Sequencing + Comprehensive mtDNA Analysis

CORRESPONDING PARENTAL TESTS
(Both Biological Parents Are Required)

- 1550 Parental WES - Maternal
- 1550 Parental WES - Paternal
- 1602 WES - Additional Affected Sibling

NOTE: Please use separate *Additional Affected Sibling* for Trio requisition for additional family members.

DUO WES TEST OPTIONS

- 1603 Duo Whole Exome Sequencing
- 1723 Rapid Duo Whole Exome Sequencing

CORRESPONDING PARENTAL TESTS
(One Parent Is Required)

- 1550 Parental WES - Maternal
- 1550 Parental WES - Paternal
- 1602 WES - Additional Affected Sibling

NOTE: Please use separate *Additional Affected Sibling* for Trio requisition for additional family members.

PROBAND WES TEST OPTIONS

- 1500 Proband Whole Exome Sequencing
- 1530 Proband Whole Exome Sequencing + Chromosomal Microarray Analysis (CMA) (Comprehensive)
- 1531 Proband Whole Exome Sequencing + Comprehensive mtDNA Analysis
- 1729 Rapid Proband Whole Exome Sequencing

CORRESPONDING PARENTAL TESTS

- 6997 Parental Control

OPT-IN TESTING OPTIONS

Opt-In for RNA Sequencing (RNAseq) as Reflex to WES

- If WES identifies a qualified variant that might be reclassified through RNA sequencing, please reflex to RNAseq if possible.

GLOBAL MAPS® TESTS

- 4900 Global Metabolomic Assisted Pathway Screen - Plasma from EDTA
Was plasma extracted from EDTA? Yes No
- 4901 Global Metabolomic Assisted Pathway Screen - Urine

Skin biopsy sample type not available for Global Maps Tests

ADD-ON TESTS

- 8665 Chromosomal Microarray Analysis (CMA)-HR+SNP Screen (Comprehensive)
- 2055 Comprehensive mtDNA analysis by NGS
- 9815 Exome Raw Data Release

ADDITIONAL REPORTING OPTIONS

If a box is not checked the lab will default to No / Not Report.

Option for Reporting of Incidental Findings

Pathogenic and likely pathogenic variants in genes covered under Category II of the Incidental Findings section of the consent form will be reported.

- Please report pathogenic and likely pathogenic variants in genes associated with Incidental Findings.

Trio Orders Only – Option for Reporting of Research Findings

For variants in genes with no known disease association, these variants will be reported if biallelic or de novo.

- Please report biallelic and de novo variants in genes with no known disease association.

WHOLE EXOME SEQUENCING (WES) REQUISITION

Patient Last Name _____ Patient First Name _____ MI _____ Date of Birth (MM / DD / YYYY) _____ Genetic Sex _____

PROBAND SAMPLE(S)

Please refer to www.baylorgenetics.com for full sample requirements.

- | | | | |
|---|--|---|---|
| <input type="radio"/> Blood in EDTA (preferred) | <input type="radio"/> Saliva | <input type="radio"/> mtDNA analysis only | <input type="radio"/> Global MAPS® only |
| <input type="radio"/> Buccal Swab | <input type="radio"/> Skin Biopsy† | <input type="radio"/> Skeletal Muscle | <input type="radio"/> Plasma from EDTA |
| <input type="radio"/> Cord Blood | <input type="radio"/> Extracted DNA from _____ | <input type="radio"/> Liver | <input type="radio"/> Urine |
| <input type="radio"/> Cultured Skin Fibroblast | | <input type="radio"/> Tissue | Date of Collection (MM / DD / YYYY) _____ |

NOTE: Extracted DNA/RNA will only be accepted if the isolation of nucleic acids for clinical testing occurs in a CLIA-certified laboratory or a laboratory meeting equivalent requirements as determined by the CAP and/or the CMS.

BIOLOGICAL PARENTS INFORMATION

BIOLOGICAL PARENTS SAMPLES ARE REQUIRED FOR TRIO WES; Other family members cannot be substituted for either parent. Be sure to label parental samples with full name and date of birth - DO NOT LABEL WITH CHILD'S NAME. Parent(s) must sign the parental testing authorization on consent.

MATERNAL INFORMATION

- Asymptomatic Symptomatic (Attach summary of findings)

Maternal Last Name _____ Maternal First Name _____ MI _____
 Maternal Date of Birth (MM / DD / YYYY) _____ / _____ / _____ Sample Type:
 Blood in EDTA (preferred)
 Buccal Swab
 Saliva
 Date of Collection (MM / DD / YYYY) _____ / _____ / _____

PATERNAL INFORMATION

- Asymptomatic Symptomatic (Attach summary of findings)

Paternal Last Name _____ Paternal First Name _____ MI _____
 Paternal Date of Birth (MM / DD / YYYY) _____ / _____ / _____ Sample Type:
 Blood in EDTA (preferred)
 Buccal Swab
 Saliva
 Date of Collection (MM / DD / YYYY) _____ / _____ / _____

ITEM CHECKLIST FOR TESTING

- | | | |
|--|--|---|
| <input type="checkbox"/> Proband Sample (Required) | <input type="checkbox"/> Signed WES Consent Form | <input type="checkbox"/> Indication for Study |
| <input type="checkbox"/> Maternal Sample (Required for Trio WES) | <input type="checkbox"/> Clinical Note/Summary | <input type="checkbox"/> Pedigree (Optional) |
| <input type="checkbox"/> Paternal Sample (Required for Trio WES) | <input type="checkbox"/> Requisition | |

* This sample type incurs an additional fee and typically adds 14 days to the turnaround time, depending on sample quality.
 † Baylor Genetics will store this sample for up to 14 days after the report is issued, allowing for follow-up testing if needed.

WHOLE EXOME SEQUENCING (WES) REQUISITION

Patient Last Name _____

Patient First Name _____

MI _____

Date of Birth (MM / DD / YYYY) _____ / _____ / _____

Genetic Sex _____

INDICATION FOR TESTING (REQUIRED)

Please provide the following clinical information regarding the patient to be tested. Please also submit a clinic note and pedigree, if available. Phenotypes listed are in HPO terms with the corresponding HPO number (<http://human-phenotype-ontology.github.io/>). This information is needed to facilitate interpretation of whole exome sequencing results. If the laboratory requires additional information, please indicate the health care provider to be contacted:

PRE/PERINATAL HISTORY

- 0001622 Prematurity - GA at birth _____
- 0001511 Intrauterine Growth Restrictions
- 0001562 Oligohydramnios
- 0001561 Polyhydramnios
- 0000476 Cystic Hygroma
- 0000776 Congenital Diaphragmatic Hernia
- 0001508 Failure to Thrive
- 0001539 Omphalocele
- 0002084 Encephalocele
- 0010880 Increased Nuchal Translucency
- _____

EYE DEFECTS & VISION

- 0000505 Visual Impairment
- 0000618 Blindness
- 0000589 Coloboma
- 0000526 Aniridia
- 0000528 Anophthalmia
- 0000568 Microphthalmia
- 0000508 Ptosis
- 0000486 Strabismus
- 0000519 Cataract Congenital Bilateral
- _____
- _____

MOTOR/COGNITIVE DEVELOPMENT

- 0000750 Delayed Speech & Language Development
- 0001270 Delayed Motor Milestones
- 0002376 Developmental Regression
- Intellectual Disability
 - 0001256 Mild
 - 0002342 Moderate
 - 0010864 Severe
- 0000729 Autistic Spectrum Disorder
- _____
- _____

STRUCTURAL BRAIN ABNORMALITIES

- 0001360 Holoprosencephaly
- 0001339 Lissencephaly
- 0002084 Encephalocele
- 0000238 Hydrocephalus
- 0002119 Ventriculomegaly
- 0001273 Abnormality of Corpus Callosum
- 0002539 Cortical Dysplasia
- 0012444 Brain Atrophy
- 0002352 Leukoencephalopathy
- 0002269 Abnormality of Neuronal Migration
- 0002126 Polymicrogyria
- 0001302 Pachgyria
- 0002500 Abnormality of Cerebral White Matter
- 0007266 Cerebral Dysmyelination
- 0006808 Cerebral Hypomyelination
- 0002134 Abnormality of the Basal Ganglia
- 0002363 Abnormality of the Brainstem
- 0007360 Aplasia/Hypoplasia of the Cerebellum
- 0006817 Aplasia/Hypoplasia of the Cerebellar Vermis
- _____

NEUROLOGICAL

- 0001284 Areflexia
- 0200134 Epileptic Encephalopathy
- 0001250 Seizures
 - 0002373 Febrile Seizures
 - 0012469 Infantile Spasms
 - 0002123 Generalized Myoclonic Seizures
 - 0002069 Generalized Tonic-clonic Seizures
 - 0010818 Generalized Tonic Seizures
 - 0010819 Atonic Seizures
 - 0002121 Absence Seizures
 - 0011169 Generalized Clonic Seizures
 - 0001251 Ataxia
 - 0001332 Dystonia
 - 0002072 Chorea
 - 0001257 Spasticity
 - 0009830 Neuropathy
- _____
- _____

CRANIOFACIAL

- 0000256 Macrocephaly
- 0000252 Microcephaly
- 0001363 Craniosynostosis
- 0000204 Cleft Upper Lip
- 0000175 Cleft Palate
- 0000316 Hypertelorism
- 0000601 Hypotelorism
- 0008050 Abnormality of the Palpebral Fissures
- 0000286 Epicanthal Folds
- 0000288 Abnormality of the Philtrum
- 0010938 Abnormality of the External Nose
- _____
- _____

Indications continued on next page



WHOLE EXOME SEQUENCING (WES) REQUISITION

Patient Last Name _____

Patient First Name _____

MI _____

Date of Birth (MM / DD / YYYY) _____ / _____ / _____

Genetic Sex _____

INDICATION FOR TESTING (REQUIRED) - CONTINUED

HAIR & SKIN

- 0000957 Cafe-Au-Lait Spots
- 0001034 Hypermelanotic Macule
- 0001010 Hypopigmentation of the Skin
- 0008066 Abnormal Blistering of the Skin
- 0008064 Ichthyosis
- 0000988 Skin Rash
- 0001581 Recurrent Skin Infections
- 0005306 Capillary Hemangiomas
- 0001597 Abnormality of the Nail
- 0004554 Generalized Hypertrichosis
- 0001596 Alopecia
- 0002208 Coarse Hair
- 0002299 Brittle Hair
- _____
- _____

CARDIAC

- 0001631 Atria Septal Defect
- 0001629 Ventricular Septal Defect
- 0001655 Patent Foramen Ovale
- 0001713 Abnormality of Cardiac Ventricle
- 0001636 Tetralogy of Fallot
- 0001680 Coarctation of Aorta
- 0001647 Bicuspid Aortic Valve
- 0002616 Aortic Root Dilatation
- 0001638 Cardiomyopathy
- 0011675 Arrhythmia
- _____
- _____

GENITOURINARY

- 0000113 Polycystic Kidney Dysplasia
- 0000107 Renal Cyst
- 0008738 Partially Duplicated Kidney
- 0000104 Renal Agenesis
- 0000085 Horseshoe Kidney
- 0000069 Abnormality of the Ureter
- 0000795 Abnormality of the Urethra
- 0000047 Hypospadias
- 0000028 Cryptorchidism
- 0000035 Abnormality of the Testis
- 0000062 Ambiguous Genitalia
- _____
- _____

RESPIRATORY

- 0002093 Respiratory Insufficiency
- 0002878 Respiratory Failure
- 0002104 Apnea
- 0002791 Hypoventilation
- 0002883 Hyperventilation
- 0002788 Recurrent Upper Respiratory Tract Infections
- _____
- _____

METABOLIC

- 0001946 Ketosis
- 0003074 Hyperglycemia
- 0001943 Hypoglycemia
- 0001941 Acidosis
- 0003128 Lactic Acidosis
- 0003215 Dicarboxylic Aciduria
- 0002490 Increased CSF lactate
- 0001992 Organic Aciduria
- 0030085 Abnormal CSF Lactate Level
- 00003542 Increased Serum Pyruvate
- 0003535 3-Methylglutaconic aciduria
- 0001942 Metabolic acidosis
- 0100493 Hypoammonemia
- 0001987 Hyperammonemia
- 0004923 Hyperphenylalaninemia
- 0003234 Decreased Plasma Carnitine
- 0003236 Elevated Serum Creatine Phosphokinase
- Abnormal Newborn Screen
- Unusual Color/Odor
- _____
- _____

MUSCULOSKELETAL

- 0011398 Hypotonia
- 0001276 Hypertonia
- 0000098 Tall Stature
- 0004322 Short Stature
- 0001382 Joint Hypermobility
- 0001371 Flexion Contracture
- 0002804 Arthrogryposis Multiplex Congenita
- 0001161 Hand Polydactyly
- 0001829 Foot Polydactyly
- 0006101 Finger Syndactyly
- 0001770 Toe Syndactyly
- 0100490 Camptodactyly of Finger
- 0012165 Oligodactyly
- 0001762 Talipes Equinovarus
- 0002757 Recurrent Fractures
- 0002650 Scoliosis
- 0002808 Kyphosis
- 0003307 Hyperlordosis
- 0001528 Hemihypertrophy
- 0001513 Obesity
- 0001548 Overgrowth
- 0002652 Skeletal Dysplasia
- _____
- _____

GASTROINTESTINAL

- 0002021 Pyloric Stenosis
- 0002575 Tracheoesophageal Fistula
- 0002032 Esophageal Atresia
- 0002020 Gastroesophageal Reflux
- 0001733 Pancreatitis
- 0002014 Diarrhea
- 0002019 Constipation
- 0002037 Inflammatory Bowel Disease
- 0004389 Intestinal Pseudo-Obstruction
- 0001399 Hepatic Failure
- 0002572 Episodic Vomiting
- 0001744 Splenomegaly
- 0002240 Hepatomegaly
- 0001508 Postnatal Failure to Thrive
- 0002578 Gastroparesis
- _____
- _____

Indications continued on next page

WHOLE EXOME SEQUENCING (WES) REQUISITION

Patient Last Name _____ Patient First Name _____ MI _____ Date of Birth (MM / DD / YYYY) _____ Genetic Sex _____

INDICATION FOR TESTING (REQUIRED) - CONTINUED

ENDOCRINE

- 0000819 Diabetes Mellitus
- 0000873 Diabetes Insipidus
- 0000821 Hypothyroidism
- 0000829 Hypoparathyroidism
- 0000834 Abnormality of the Adrenal Glands
- 0001738 Exocrine Pancreatic Insufficiency
- 0002721 Immunodeficiency
- _____
- _____

EAR DEFECTS & HEARING

- 0000407 Sensorineural Hearing Impairment
 - 0008619 Bilateral
- 0000405 Conductive Hearing Impairment
- 0000410 Mixed Hearing Impairment
- 0004467 Preauricular Pit
- 0000384 Preauricular Skin Tag
- 0000369 Low-set Ears
- 000037 Abnormality of the Pinna
- _____
- _____

HEMATOLOGY

- 0001875 Neutropenia
 - 0005549 Congenital
 - Chronic
 - Cyclic
- 0001873 Thrombocytopenia
- 0040185 Macrothrombocytopenia
- 0005537 Decreased Mean Platelet Volume
- 0005518 Erythrocyte Macrocytosis
- 0004444 Spherocytosis
- 0012410 Pure Red Cell Aplasia
 - Aplastic
 - Hypoplastic
- 0001903 Anemia
- 0005528 Bone Marrow Hypocellularity
- _____
- _____

CANCER

- Type of Cancer _____
- Age of Diagnosis _____
- Family History of Cancer and Affected Relatives _____
- _____
- _____

OTHER

- Organomegaly
- Chronic Infections
- 0004311 Abnormality of Macrophages
- 0001954 Episodic Fever
- 0004313 Hypogammaglobulinemia
- 0010701 Abnormal Immunoglobulins
- 0002721 Immunodeficiency
- 0012088 Abnormal urinary odor
- 0012537 Food intolerance
- 0008067 Abnormally lax or hyperextensible skin
- Abnormal Movements
- Family History of Similar Disorder
- 0001254 Lethargy
- 0002415 Leukodystrophy
- _____
- _____

GENES OF INTEREST

ADDITIONAL CLINICAL INFORMATION

DIFFERENTIAL DIAGNOSIS

Consent on next page

WHOLE EXOME SEQUENCING (WES) CONSENT

Patient Last Name Patient First Name MI Date of Birth (MM / DD / YYYY) Genetic Sex

TEST INFORMATION

This consent form will provide you with information regarding Whole Exome Sequencing (WES), which you should discuss with your healthcare provider or a genetic counselor. To assist you in understanding the reason for this testing, we have provided information about the testing process and potential results below. This testing can be performed on you or your child. "Your child" can also mean your unborn child, for the purposes of this consent.

The WES test may identify changes, called variants, in a person's DNA that cause genetic diseases or medical conditions. DNA is the genetic material that we receive from our parents. Genes are made of DNA and are the instructions for maintaining the health of our bodies. The WES test provides a comprehensive analysis of the exome, which is the part of the human genome that helps the body make proteins. The WES test will analyze the important regions of thousands of genes at the same time. Based on the symptoms that are known for you/your child, genes with changes associated with these symptoms will be reported. It is possible that even if WES identifies the underlying genetic cause for a disease in a family, this information may not help in predicting medical outcomes or changing medical management or treatment of disease. In addition, WES testing may identify information about genes and diseases that have a clear and immediate medical significance to your health or the health of your family members, even if that information is not related to the currently known symptoms. After you have received your results, you should discuss the significance of these results with your healthcare provider or genetic counselor.

RESULTS

There are several types of test results that may be reported including:

- **Positive:** Positive or "abnormal" results mean a variant in the DNA was detected that is related to your/your child's medical issues or that you/your child are at an increased risk of developing a disease in the future. It is possible to test positive for more than one variant. Positive results might include pathogenic variants (variants known to be associated with disease) and likely pathogenic variants (variants that are likely to be associated with disease).
- **Negative:** Negative or "normal" results mean that no relevant variants were detected that are related to your/your child's medical issues or that would increase your/your child's risk for developing a disease in the future. This might indicate that there are no variants associated with disease in the genes tested. Genetic testing, while highly accurate, might not detect a variant present in the genes tested. This can be due to limitations of the information available about the genes being tested, or limitations of the testing technology.
- **Variant of Uncertain Clinical Significance:** Testing can detect variant(s) in DNA which we do not yet fully understand. These are also referred to as variants of uncertain clinical significance (VUS). Additional testing may be recommended for you/your child or your family if a VUS is identified in a gene that may be associated with your/your child's medical condition.
- **Secondary / Incidental Findings:** Testing can sometimes detect a variant in a person's DNA unrelated to the reason for testing. If this variant is expected to have medical or reproductive significance, it is called a secondary or incidental finding.

INCIDENTAL FINDINGS

This test may find changes in genes that cause symptoms or diseases not related to the reason for having the test. These are called Secondary or Incidental Findings, and are associated with a clear and immediate medical significance to your/your child's health or the health of your family members.

CATEGORY I: ACMG SECONDARY FINDINGS

The American College of Medical Genetics (ACMG) has published a series of guidelines for the reporting of these types of medically actionable or secondary findings (including PMID: 34012068). These guidelines include a list of genes, which are updated occasionally, that are considered medically actionable and indicate laboratories should report pathogenic (disease-causing) and likely pathogenic findings in these genes. In accordance with an update to this policy statement (PMID: 25356965), you and your provider may choose to opt-in to have these findings reported — please indicate this selection in the Patient Reporting Options and Release of Updated Results section below.

CATEGORY II: OTHER INCIDENTAL FINDINGS

Medically actionable variants are changes found in genes known to be associated with disease but not associated with your/your child's current symptoms or clinical presentation. These variants are reported as they may cause severe, early-onset disease or may have implications for treatment and prognosis. You and your provider may choose to opt-in to have these findings reported — this selection is on page 2 of the test requisition form.

ADDITIONAL REPORTING INFORMATION

The report will NOT include findings in genes causing adult-onset neurodegenerative syndromes for which there is presently no prevention or cure unless directly related to the phenotype. If specific genes causing adult-onset neurodegenerative syndromes should be considered for reporting, these genes should be marked in the Genes of Interest section on the requisition. For each gene, please indicate whether findings should be reported for only the proband (patient) or both the proband and their parents.

Additional reporting for Proband WES: Samples from biological parents may help facilitate interpretation of Proband (patient-only) WES results. After the proband report is issued, parental samples can be tested by WES or targeted testing for the variants detected in the proband's exome data at an additional charge. Free testing for variants of uncertain clinical significance for immediate family members is available with prior written approval.

Additional considerations for Duo/Trio WES: As part of the Duo/Trio WES test, a sample from one (for Duo) or both (for Trio) biological parent(s) is required. WES will be performed on the proband (patient) and parental sample(s) at the same time and the sequence data will be analyzed in the context of the family relationships. The parental data will be used to help interpret the proband's data. Follow up testing is available for other family members at an additional charge. Free testing for variants of uncertain clinical significance is available with prior written approval. A separate report for each parent will be issued regarding any secondary findings that are identified.

Your physician may order a test that includes WES in combination with another type of testing. These tests include other methodologies which may help identify changes that the WES alone cannot. Testing of parents with other methodologies may or may not be necessary to interpret the proband's results. Any results obtained from these additional tests will be included in a separate report from the WES report. Please visit the Baylor Genetics website for further information about these tests and their associated consent forms.

WHOLE EXOME SEQUENCING (WES) CONSENT

Patient Last Name Patient First Name MI / / _____
Date of Birth (MM / DD / YYYY) Genetic Sex

RNASEQ INFORMATION

For variants that meet certain criteria ("qualified variants"), a comprehensive analysis of the RNA can be performed by RNAseq. RNA is made from DNA and is used by the body to create many different proteins. RNAseq can help clarify the clinical significance of the qualified variant(s) being assessed. It is possible that even if RNAseq identifies additional information it may not be enough to clarify the clinical significance of any or all qualified variants.

The results of RNAseq may help to clarify the clinical significance of one or more variant(s) identified via WES. An updated version of your WES report may be issued with information obtained from RNAseq. Possible test results may include:

- **Reclassification of the variant to pathogenic/likely pathogenic ("upgrade"):** One or more previously identified variant(s) are now classified as pathogenic or likely pathogenic. These variants are now considered to be related to your/your child's medical issues or indicate that you/your child are at an increased risk of developing a disease in the future.
- **Reclassification of the variant to benign ("downgrade"):** One or more previously identified variants are now classified as benign (unlikely to be associated with disease). These variants are now considered unrelated to your/your child's medical issues and not expected to be associated with an increased risk of developing a disease in the future.
- **Classification of the variant remains the same:** One or more previously identified variant(s) was not able to be upgraded or downgraded. These variants still have the same classification. Additional testing may be recommended to further clarify the clinical significance of these variants.

CONSIDERATIONS AND LIMITATIONS

- This consent form can only be used for WES. Consent forms for other tests are located at Baylor Genetics' website (<https://www.baylorgenetics.com/consent/>).
- Results may indicate you/your child have a genetic disease, are at increased risk to develop a genetic disease, and/or be at an increased risk to have a child with a genetic disease. It is important to understand that genetic tests, even if negative, cannot rule out every variant. Genetic testing, while highly accurate, might not detect a variant present in the gene(s) tested. This can be due to limitations of the information available about the gene(s) being tested, or limitations of the testing technology. It is not possible to exclude risks for all genetic diseases for you/your child and your family members.
- It is possible that even if the test identifies the underlying genetic cause for the disease in your family, this information may not help in predicting the progression of disease or change management or treatment of disease.
- Depending on the type of genetic testing performed and the results, additional genetic testing or other testing may be needed to fully understand the likelihood of you/your child developing the disease or the severity of the disease. This additional testing might be needed for you/your child or other members of your family. This information will be discussed by your healthcare provider and additional consent obtained as required.
- In many instances, WES will not identify a qualified variant. If no qualified variant is identified by WES, RNAseq will not be performed.
- It is recommended that you discuss genetic testing with your healthcare provider or genetic counselor before signing this consent and again after results are made available.
- It may not always be possible to complete testing as sometimes the sample does not have enough DNA/RNA to perform testing or other reasons. In these cases, another sample may need to be sent to the laboratory to perform testing.

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION

- If several family members are tested, the correct interpretation of the results depends on the information provided about the relationships among family members. In rare cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. If a difference is identified, it may be necessary to share this information with the healthcare provider who ordered the testing.
- Genetic testing is highly accurate, however, in rare cases, inaccurate results may occur. Reasons for this include, but are not limited to, mislabeled samples, inaccurate reporting of clinical/medical information, or rare technical errors.
- If you sign this consent form, but you no longer wish to have your/your child's sample(s) tested, you can contact the healthcare provider who ordered the test to cancel the test. If you wish to cancel testing, the laboratory must be notified of the cancellation request before 5 PM CST the business day after the sample has been received by Baylor Genetics. If the laboratory is not notified of your cancellation request until after this time, you will be charged for the full cost of the test.
- Only Baylor Genetics and Baylor Genetics contracted partners will have access to the sample(s) provided to conduct the requested testing. Results will only be released to the following person(s): (i) a licensed healthcare provider, (ii) those authorized in writing, (iii) the patient or their personal representative, and (iv) those allowed access to test results by law. I understand that I have the right to access my test results directly from Baylor Genetics by providing a written request. I also understand that laboratory raw data can be requested by providing a written request or HIPAA Authorization Form.
- In rare cases, persons with genetic diagnoses have experienced problems with insurance coverage and employment. The U.S. 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, you can visit www.genome.gov/10002077.
- Samples will be retained in the laboratory in accordance with the laboratory retention policy.
- After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring healthcare providers unless specific prior arrangements have been made.
- Samples from residents of New York State will not be included in general research studies without your written consent and will not be retained for more than 60 days after receipt of the sample, unless specifically authorized by your selection below. No tests other than those authorized shall be performed on the biological sample.

FOR SAMPLES SUBMITTED FROM NEW YORK STATE

_____ I understand that no genetic test other than those I have authorized shall be performed on my biological sample, and the sample will be destroyed at the end of testing or not more than 60 days after the sample was taken. However, by initialing here, I hereby authorize the lab to retain my sample(s) for longer retention in accordance with the laboratory retention policy for internal laboratory quality assurance studies and possible research testing.

- By signing this Consent form, I understand and agree that information identified may also be submitted to public databases, such as ClinVar. Such submission serves to contribute knowledge to the medical community. I understand that limited clinical information is also required for the submission of information to ClinVar's database and further that the contents of this limited clinical information may, although unlikely, include information that may identify me or members of my family.

WHOLE EXOME SEQUENCING (WES) CONSENT

Patient Last Name Patient First Name MI Date of Birth (MM / DD / YYYY) Genetic Sex

PATIENT REPORTING OPTIONS AND RELEASE OF UPDATED RESULTS

Please read the statements below carefully and check the appropriate box. Due to the nature of the methodology of this testing we are unable to guarantee that all pathogenic (disease-causing) variants in each option will be detected by WES.

For all options below: If no selection is made, this will default to the NO option.

FOR ALL WES:

REPORTING OF CATEGORY I (ACMG) SECONDARY FINDINGS FOR THE PATIENT

Pathogenic and likely pathogenic variants in genes included in the ACMG policy statement regarding recommendations for reporting of secondary findings will be reported as medically actionable on the WES report.

- YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.
- NO - Please do NOT report pathogenic and likely pathogenic variants in genes included in the ACMG policy statement.

OPTION TO ALLOW RELEASE OF UPDATED RESULT

If a possible diagnosis can be made with new information, we would like to issue an updated report to the physician who ordered your WES. This updated report will NOT include a complete review of all of your/your child's data.

- YES - If new information regarding the clinical significance of changes in my/my child's WES becomes known, I would like Baylor Genetics to issue an updated report which includes this information to my physician who ordered this WES testing.
- NO - Please do NOT issue an updated report if there is new information regarding the clinical significance of my/my child's WES that becomes known.

FOR DUO AND TRIO WES ONLY:

We understand that our samples will be utilized for Duo or Trio WES as ordered by our healthcare provider. This will be analyzed to help interpret the sequence data of our child. A separate parental report will be issued regarding the below category of secondary findings. Testing of parental status for this category of results will be initiated independently of our child's data. It may be possible to infer information about a family member's results based on our child's or other family member's results.

REPORTING OF MATERNAL CATEGORY I (ACMG) SECONDARY FINDINGS

Pathogenic and likely pathogenic variants in genes included in the ACMG policy statement regarding recommendations for reporting of incidental findings will be reported as medically actionable on the maternal WES report.

- YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.
- NO - Please do NOT report pathogenic or likely pathogenic variants in genes included in the ACMG policy statement.

REPORTING OF PATERNAL CATEGORY I (ACMG) SECONDARY FINDINGS

Pathogenic and likely pathogenic variants in genes included in the ACMG policy statement regarding recommendations for reporting of incidental findings will be reported as medically actionable on the paternal WES report.

- YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.
- NO - Please do NOT report pathogenic or likely pathogenic variants in genes included in the ACMG policy statement.

FOR WES PERFORMED ON ANOTHER FAMILY MEMBER BESIDES THE PROBAND OR PARENTS ONLY:

We understand that our samples will be utilized for WES as ordered by our healthcare provider. This will be analyzed to help interpret the sequence data of my other family members being tested. A separate report will be issued regarding the below category of secondary findings. Testing of familial status for these categories of results will be initiated independently of my family member's data. It may be possible to infer information about a family member's results based on the results obtained.

REPORTING OF CATEGORY I (ACMG) SECONDARY FINDINGS FOR OTHER FAMILY MEMBER

Pathogenic and likely pathogenic variants in genes included in the ACMG policy statement regarding recommendations for reporting of incidental findings will be reported as medically actionable on the family member's WES report.

- YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.
- NO - Please do NOT report pathogenic or likely pathogenic variants in genes included in the ACMG policy statement.



WHOLE EXOME SEQUENCING (WES) CONSENT

Patient Last Name

Patient First Name

MI

_____/_____/_____
Date of Birth (MM / DD / YYYY)

Genetic Sex

FINANCIAL AGREEMENT AND GUARANTEE

By signing this consent form, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider. For insurance billing, I hereby authorize Baylor Genetics to bill my health insurance plan on my behalf, and further authorize Baylor Genetics to release any information to my insurance carrier which is reasonably required for billing. I additionally designate Baylor Genetics as my designated representative for purposes of appealing any denial of benefits by my insurance carrier. I irrevocably assign associated payment to Baylor Genetics, and direct that payment be made directly to Baylor Genetics. I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by Baylor Genetics as part of a verification of benefits investigation. I agree to be financially responsible for all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for unpaid services performed by Baylor Genetics on my behalf, I agree to endorse the insurance check as appropriate and forward such check to Baylor Genetics within thirty (30) days of receipt thereof, as payment towards Baylor Genetics' claim for services rendered. If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by Baylor Genetics.

If my health insurer does not cover the test or I do not have health insurance, I have received a good faith estimate of the cost for the genetic testing ordered by my provider and agree to pay for the cost of the genetic testing billed to me by Baylor Genetics based on that good faith estimate. More information is available in Baylor Genetics' No Surprises Act and Good Faith Estimate Notice located at <https://www.baylorgenetics.com/no-surprises-act/>.

I understand that a completed Advance Beneficiary Notice (ABN) is required for Medicare fee for service patients if the service is not payable by Medicare as not medically necessary or reasonable.

RECONTACT FOR RESEARCH CONSENT

Baylor Genetics participates in research relating to health, disease prevention, drug development, and other scientific purposes. Baylor Genetics may contact patients directly as part of this research. I agree to allow Baylor Genetics to contact me about possible research involving the sample(s) and/or information associated with this testing. I understand that patients generally receive no compensation for this participation in research. For more information on research at Baylor Genetics, please visit baylorgenetics.com.

If I wish to opt out of being recontacted for research purposes by Baylor Genetics, I understand that I may check the box below:

Please do not contact me regarding any research that uses information obtained from this testing.

For any research I may be contacted about, I prefer contact through the following methods (please check all that apply – if no choices are selected, contact via secure email will be made if an email address is provided):

Email Phone Mail

PATIENT AUTHORIZATION

By signing this statement of consent, I acknowledge that I have read, understand, and hereby grant my informed consent for genetic testing. I have received appropriate explanations from my healthcare provider about the planned genetic test(s) and possible results. I have been informed by my healthcare provider about the availability and importance of genetic counseling and have been provided with written information identifying a genetic counselor or medical geneticist who can provide such counseling services. All my questions have been answered and I have had the necessary time to make an informed decision about the genetic test(s).

Note: If Prenatal WES was ordered, please leave the Patient section blank and complete only the Maternal and Paternal section below.

I hereby give permission to Baylor Genetics to conduct genetic testing as recommended by my physician.

Patient Name

Patient's Signature

_____/_____/_____
Date Signed (MM / DD / YYYY)

Patient's Parent / Personal Representative* Name

Patient's Parent / Personal Representative Signature

_____/_____/_____
Date Signed (MM / DD / YYYY)

Relationship of Personal Representative* to the Patient

Ordering Provider's Signature

_____/_____/_____
Date Signed (MM / DD / YYYY)



WHOLE EXOME SEQUENCING (WES) CONSENT

Patient Last Name Patient First Name MI Date of Birth (MM / DD / YYYY) Genetic Sex

PATIENT AUTHORIZATION

FOR DUO, TRIO, AND PRENATAL TRIO WES ONLY

Maternal Name Maternal Signature Date Signed (MM / DD / YYYY)

Paternal Name Paternal Signature Date Signed (MM / DD / YYYY)

Maternal Personal Representative* Name Maternal Personal Representative* Signature Date Signed (MM / DD / YYYY)

Relationship of Maternal Personal Representative* Date Signed (MM / DD / YYYY)

Paternal Personal Representative* Name Paternal Personal Representative* Signature Date Signed (MM / DD / YYYY)

Relationship of Paternal Personal Representative* Date Signed (MM / DD / YYYY)

FOR AFFECTED SIBLING OR OTHER FAMILY MEMBER WES ONLY

Affected Sibling/Other Family Member Name Affected Sibling/Other Family Member Signature Date Signed (MM / DD / YYYY)

Affected Sibling/Other Family Member Parent /
Personal Representative* Name Affected Sibling/Other Family Member Parent /
Personal Representative* Signature Date Signed (MM / DD / YYYY)

Relationship of Personal Representative* to Affected Sibling /
Other Family Member Date Signed (MM / DD / YYYY)