BAYLOR GENETICS

BAYLOR GENETICS
2450 HOLCOMBE BLVD.
GRAND BLVD. RECEIVING DOCK
HOUSTON, TX 77021-2024

PHONE
1.800.411.4363
FAX
1.800.434.9850

CONNECT

ADDITIONAL AFFECTED SIBLING FOR TRIO 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 #

Patient discharged from the hospital/facility: ◯ Yes ◯ No

Biological Sex: ◯ Female ◯ Male ◯ Unknown

Gender identity (if different from above):

REPORTING RECIPIENTS

Ordering Physician | Institution Name

Email (Required for International Clients) | Phone | Fax

ADDITIONAL RECIPIENTS

Name | Email | Fax

Name | Email | Fax

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) 3. Name of Ordering Physician 4. Insured Signature of Authorization

Name of Insured | Insured Date of Birth (MM / DD / YYYY)

Patient's Relationship to Insured | Phone of Insured

Address of Insured

City | State | Zip

Primary Insurance Co. Name | Primary Insurance Co. Phone

Primary Member Policy # | Primary Member Group #

Name of Insured | Insured Date of Birth (MM / DD / YYYY)

Patient's Relationship to Insured | Phone of Insured

Address of Insured

City | State | Zip

Secondary Insurance Co. Name | Secondary Insurance Co. Phone

Secondary Member Policy # | Secondary Member Group #

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, as well as any amounts not paid by my insurance carrier for reasons including, but not limited to, non-covered and non-authorized services. 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 that Medicare does not cover routine screening tests.

Patient's Printed Name | Patient's Signature | Date (MM / DD / YYYY)

STATEMENT OF MEDICAL NECESSITY (REQUIRED)

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)

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ETHNICITY

- ◯ African American
- ◯ Ashkenazi Jewish
- ◯ East Asian (China, Japan, Korea)
- ◯ Finnish
- ◯ French Canadian
- ◯ Hispanic American
- ◯ Mennonite
- ◯ Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)
- ◯ Native American
- ◯ Northern European Caucasian (Scandinavian, UK, Germany)
- ◯ Pacific Islander (Philippines, Micronesia, Malaysia, Indonesia)
- ◯ South Asian (India, Pakistan)
- ◯ Southeast Asian (Vietnam, Cambodia, Thailand)
- ◯ Southern European Caucasian (Spain, Italy, Greece)
- ◯ Other (Specify): _____

TEST OPTION

● 1602 Additional Affected Sibling for Trio

SAMPLE

SAMPLE TYPE

- ◯ Blood
- ◯ Buccal Swab
- ◯ Cord Blood
- ◯ Cultured Skin Fibroblast
- ◯ Extracted DNA from: _____

Date of Collection: ____ / ____ / ____ (MM / DD / YYYY)

For Trio WES Orders (1600 and 1532)

Parental samples are REQUIRED. Testing cannot proceed unless both biological parental samples are received. If all three familial samples cannot be sent together, the samples will be prepped and held until all necessary samples are received. Testing will be cancelled if all three samples do not arrive within 8 weeks after receipt of the 1st sample. Please consider Proband Whole Exome Sequencing (Test Code 1500) if all three familial samples cannot be collected (See separate requisition on our website at www.BMGL.com).

For Additional Affected Sibling for Trio Orders (1602)

A complete Trio WES order should be submitted along with or prior to the submission of the Additional Affected Sibling for Trio order. The sibling must be the full biological sibling (same mother and father) as the proband for Trio Whole Exome Sequencing; substitutions for other family members, or healthy siblings will not be accepted. The sibling submitted for this test (1602) does NOT need to have the same phenotype/symptoms as the other family member(s). The Additional Affected Sibling for Trio test will be referred to as the "Sibling Trio". Turnaround time for test code 1602 is 8 weeks AFTER financial responsibility has been verified by the Baylor Genetics billing office.

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.

ITEM CHECKLIST

- ☐ Patient Sample (EDTA Required)
- ☐ Signed WES Consent Form
- ☐ Clinical Note/Summary
- ☐ Pedigree
- ☐ Requisition
- ☐ Indication for Study

FAMILY INFORMATION

TRIO Previously Sent?

- ◯ Yes
- ◯ Concurrently

Lab # _____

Family # _____

NOTE: If sent concurrently, please complete and send Trio Whole Exome Sequencing requisition which can be found at www.bmgl.com

1600 | PROBAND INFORMATION

Proband Last Name

Proband First Name | MI

Proband Date of Birth: ____ / ____ / ____ (MM / DD / YYYY)

1550 | MATERNAL INFORMATION

Maternal Last Name

Maternal First Name | MI

Maternal Date of Birth: ____ / ____ / ____ (MM / DD / YYYY)

1550 | PATERNAL INFORMATION

Paternal Last Name

Paternal First Name | MI

Paternal Date of Birth: ____ / ____ / ____ (MM / DD / YYYY)

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ADDITIONAL AFFECTED SIBLING FOR TRIO REQUISITION

Patient Last Name | Patient First Name | MI | Date of Birth (MM / DD / YYYY) | Biological 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:

Physician Name | Physician Phone | ICD-10 Diagnosis Code(s)

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

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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 Polydactly
- ☐ 0001829 Foot Polydactly
- ☐ 0006101 Finger Syndactly
- ☐ 0001770 Toe Syndactly
- ☐ 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 Tracheoesophogeal 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

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INDICATION FOR TESTING (REQUIRED) - CONTINUED

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

_____

_____

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
- ☐ _____
- ☐ _____

OTHER

- ☐ Organomegaly
- ☐ Chronic Infections
- ☐ 0004311 Abnormality of Macrophages
- ☐ 0001954 Episodic Fever
- ☐ 0004313 Hypogammaglobulinemia
- ☐ 0010701 Abnormal Immunoglobulins
- ☐ 0002721 Immunodeficiency
- ☐ _____
- ☐ _____

GENES OF INTEREST

_____

_____

_____

_____

_____

_____

_____

_____

_____

ADDITIONAL CLINICAL INFORMATION

DIFFERENTIAL DIAGNOSIS

Consent on next page

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INFORMATION AND CONSENT FOR TESTING

The WES test is a highly complex test that is developed for the identification of changes in an individual's DNA that are causative or related to their medical concerns. The exome refers to the portion of the human genome that contains functionally important sequences of DNA that direct the body to make proteins essential for the body to function properly. These regions of DNA are referred to as exons. It is known that most of the errors that occur in DNA sequences that then lead to genetic disorders are located in the exons. In contrast to other sequencing tests that analyze one gene or small groups of related genes at a time, WES will analyze the important regions of tens of thousands of genes at the same time. Therefore, sequencing of the exome is thought to be an efficient method of analyzing a patient's DNA to discover the genetic cause of diseases or disabilities. However, it is possible that even if WES identifies the underlying genetic cause for the disorder in your family this information may not help in predicting prognosis or change medical management or treatment of disease.

DESCRIPTION OF WHOLE EXOME SEQUENCING TEST

The WES test is a highly complex test that is developed to identify changes in an individual's DNA that cause or contribute to their medical concerns. The exome refers to -the portion of the human genome that contains functionally important sequences of DNA that direct the body to make proteins essential for the body to work properly. These regions of DNA are called exons. It is known that most of the errors that occur in DNA sequences that then lead to genetic disorders are located in the exons. In contrast to other sequencing tests that analyze one gene or small groups of related genes at a time, WES will analyze the important regions of tens of thousands of genes at the same time. Therefore, sequencing of the exome is thought to be an efficient method of analyzing a person's DNA to discover the underlying genetic cause of disease.

TESTING REPORTING

When your exome sequence is compared to a normal reference sequence, many variations or differences are expected to be found. Based on currently available medical and scientific information, we will decide whether any of these variations are predicted to be causative or related to your medical concerns. The report will contain results that may explain the cause of your current medical problems. It may also contain information about genes and diseases that have clear and immediate medical significance to your health or the health of family members, whether or not they relate to your current symptoms.

You may recieve any of the following types of results:

- Positive: Positive or “abnormal” results mean there is a change in the genetic material related to your medical issues.
- Negative: Negative or “normal” results mean no relevant genetic change could be detected using WES. This does not mean there is no genetic change, but it may mean that WES could not detect it.
- Results of Unclear Significance: WES can detect change(s) in DNA that do not have clear meaning. These alterations are also referred to to as variants of uncertain significance (VUS). Additional studies may be indicated if a VUS is identified in a gene that may be associated with your medical concerns.
- SecondaryFindings: WES testing can sometimes detect a change in a person's DNA unrelated to the reason for testing. If this change has medical or reproductive significance, it is called a secondary finding.

SECONDARY FINDINGS

You have the choice to OPT-IN or OPT-OUT of the following categories of secondary findings:

Category I: Medically Actionable

The report may also contain information regarding genes and diseases that are considered medically actionable because they have clear and immediate medical significance to your health or the health of family members, whether or not they relate to your current symptoms. The American College of Medical Genetics (ACMG) has published guidelines for the reporting of theses types of medically actionable or secondary findings (PMID: 23788249, 27854360). These guidelines include a list of genes (updated periodically) that are considered medically actionable and thus, laboratories should seek and report pathogenic (disease causing) findings in these genes. In acccordance with an update to this policy statement (PMID: 25356965), there is the choice to opt-out of recieving this information.

Category II: Carrier Status

This testing can determine if an individual is a carrier of a genetic variant(s) that may not impact your health directly but may impact reproductive decision making. Carrier status will include disorders recommended for reproductive screening by professional societies such as ACMG or ACOG.These conditions include cystic fibrosis (CFTR), sickle cell anemia (S allele, HBB), familial dysautonomia (ELP1), Tay-Sachs disease (HEXA), Canavan disease (ASPA), Fanconi anemia group C (FANCC), Niemann- Pick type A, B (SMPD1), Bloom syndrome (BLM), mucolipidosis IV (MCOLN1), Gaucher disease type I (GBA).

ADDITIONAL REPORTING

The report will NOT include findings in genes causing adult onset dementia syndromes for which there is presently no prevention or cure. If the reason for testing includes features that clearly indicate such a disorder, we recommend pursuing targeted testing based on specific symptoms and not WES testing. However, if the reason for testing includes a clinical presentation that could include such a disorder or a mixed neurological phenotypes, then results may be reported in the proband (patient) and the parents for genes that have an allelic association with dementia or is a component of the phenotype. The interpretation of the variants is based on information available at the time of testing and may change in the future as medical knowledge advances. As determined necessary by the laboratory, the proband's sample will have the findings confirmed by a second methodology (Sanger sequencing). We expect to find hundreds of variations when comparing the DNA to the reference sequence, Most of these do not relate to disease and therefore, will not be reported. The raw sequence data generated by WES is available for request once a WES report has been issued. Pleasde see our website for further information regarding this.

Additional reporting for Trio WES (test codes 1600, 1722, 1532, 1533): As part of the Trio WES analysis, we will report findings in genes that have occurred in the affected individual, but not in the asymptomatic parents. This category of results caused by new (de novo) findings may be significant in determining the cause of your medical condition. Thus, this category of changes will be reported for genes with or without a known current association with disease. We will also report compound heterozygous, homozygous and hemizygous variants in genes where each parent has one change and the affected individual has inherited both changes,

continue on next page

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			/ /	

INFORMATION AND CONSENT FOR TESTING

for genes with or without a known association with disease. It is important to note that the Trio WES report may contain information about diseases and genes that do not relate to your current condition, or may develop many years from now, or do not have any known link to disease, according to current knowledge. As part of the Trio WES test, blood samples from the biological parents of the proband are required. Trio WES will be performed on the proband and parental samples 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. A separate parental report will be issued regarding the two categories of secondary findings.

Additional reporting for Proband WES (test codes 1500, 1530,1531): We will also include variants in possible candidate disease genes that might potentially contribute to patient phenotype on the focused report. Further research studies are needed to clarify the clinical relevance of those variants/genes. In discussion with your physician, an expanded report can be ordered (no additional charge) for up to six months after the focused report is recieved. The expanded report may contain information about diseases and genes that do not relate to your current condition, or may develop many years from now, or do not have any known link to disease, according to current knowledge Information included in the expanded report is not Sanger confirmed (unless determined necessary by the laboratory). A requisistion for ordering the expanded report is available on our website. Biological parental samples are requested to facilitate interpretation of Proband WES results. The parental samples will NOT be tested by whole exome sequencing; instead they will be tested by targeted methods such as Sanger sequencing for changes in genes that are highly likely to be causative of disease (related to patient indication for testing) to confirm mode of inheritance, de novo status, ect. as determined necessary by the laboratory. Additionally, if opted-in to receive carrier status for reproductive screening and medically actionable findings, this information will be issued in a separate parental report. Testing of parental status will ONLY be initiated if there is a variant identified in the proband.

Your physician may order additional tests along with WES. Further test code specific information is as follows:

Test codes1531, 1532 and 1533: In addition to WES analysis as detailed above, this order will also include a separate analysis of the mitochondrial DNA.

Test code 2055: This is the evaluation of the entire mitochondrial genome for point mutations and deletions. This will be reported separately from the WES results with a turnaround time of 50 days. If an mtDNA change is identified, the report will indicate recommendations for familial follow-up. Baylor Genetics will NOT automatically initiate testing on the maternal sample. If this is desired, please contact client services for assistance.

Test code 1530: This order will also include a separate analysis for detection of deletions and duplications plus a screen for detection of uniparental disomy (UPD) andabsence of heterozygosity (AOH).

Test code 8665: This will be reported separately from the WES results with a turnaround time of 14 days. If a copy number change is identified, the report will indicate recommendations for familial follow-up. Baylor Genetics will NOT automatically initiate testing on the parental smple(s). If this is desired, please contact client services for assistance.

Test codes 4900, 4901 and 4902 (Global MAPS): This is a large scale, semi-quantitative screening test that looks at changes in both individual analytes and pathways related to biochemical abnormalities, including (but not limited to) amino acid, organic acid, lipid and nucleotide metabolism. It should be used as a screening tool for individuals who have an undifferentiated phenotype or as supportive evidence in individuals with equivocal mutations in genes related to metabolic processes. It is not intended to supplant current diagnostic testing for specific conditions, nor is it intended for monitoring therapy. Any abnormalities detected using Global MAPS should be confirmed by diagnostic biochemical or molecular diagnostic testing. Consent for testing below is for WES and does not need to be completed if only Chromosomal Microarray Analysis, mtDNA Analysis or Global MAPS is ordered. Please visit our website for further information about these tests.

POTENTIAL RISKS, LIMITATIONS, AND DISCOMFORTS

1. It is possible that you could have a variant in a gene included in the WES test, but the WES test was unable to detect the variant. Therefore, it is possible that you may be affected with one of the conditions tested by WES, but that the test did not detect the condition.
2. The WES test does not analyze 100% of the genes in the human genome. There are some genes that cannot be included in the test due to technical reasons.
3. Results may be unclear or indicate the need for further testing on other family members.
4. It is possible that additional information may come to light during these studies regarding family relationships. For example, data may suggest that family relationships are not as reported, such as non-paternity (the father of the individual is not the biological father) or consanguinity (marriage or reproductive partners are blood relatives). Since the accurate assignment of family relationships is critical to the analysis of WES, we may perform a separate genetic test to confim that the samples that were submitted from the parents were correctely identified. If a discrepancy is identified, we will proceed with testing for the individual(s) who are correctly identified.
5. If you sign the consent form, but you no longer wish to have your samples tested by WES, you can contact your doctor to cancel the test. If testing is complete, but you have not received your results yet, you can inform your doctor that you no longer wish to recieve the results. However, if you withdraw consent for testing after 5p.m. CST, the next business day following sample receipt by the laboratory, you will be charged for the full cost of the test.
6. Information including results, indications for testing and clinical status obtained from the WES test may be shared with health care providers, scientists and health care databases or used in scientific publications or presentations, but the personal identifying information of all persons studied will not be revealed in such data sharing or publications/presentations.
7. Variants identified by WES may also be submitted to public databases, such as ClinVar, to contribute knowledge to the medical profession. Usually limited clinical information is also required for the submission. However, it is unlikely that contents of the database submissions will include any information that will identify you personally.
8. Due to the fact that many different genes and conditions are being analyzed, there is a risk that you will learn genetic information about yourself or your family that is not directly related to the reason for ordering the WES. This information might relate to diseases with symptoms that may develop in the future in yourself or other family members as well as conditions that have no current treatment.
9. It is possible that even if WES identifies the underlying genetic cause for the disorder in your family, this information may not help in predicting prognosis or change management or treatment of disease.

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CONNECT

ADDITIONAL AFFECTED SIBLING FOR TRIO REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

PROBAND REPORTING OPTIONS AND AUTHORIZATION

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

For Options 1 & 2: If neither box is checked, or if form is not signed, the lab will default to the NO/ do not report option.

INITIAL 1. MEDICALLY ACTIONABLE

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 WES report.

_____ ◯ **YES** Please report pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

_____ ◯ **NO** Please do NOT report pathogenic variants in genes included in the ACMG policy statement.

INITIAL 2. CARRIER STATUS FOR AUTOSOMAL RECESSIVE CONDITIONS RECOMMENDED FOR REPRODUCTIVE CARRIER SCREENING

_____ ◯ **YES** Please report carrier status. By checking this box, I choose to receive information regarding carrier status.

_____ ◯ **NO** Please do NOT report carrier status. By checking this box, I choose to NOT receive information regarding carrier status.

For option 3: if neither box is checked, or the form is not signed, the lab will default to the YES/ release updated report option.

INITIAL 3. OPTION TO ALLOW RELEASE OF UPDATED RESULTS

We may periodically review old cases when new information is learned regarding the significance of changes in a particular gene. If a possible diagnosis can be made with this information we would like to issue an updated report to the physician who ordered your WES test. The current schedule for this review is every six months, but is subject to change and does NOT include a complete review of all of your data.

_____ ◯ **YES** If new information is known regarding clinical significance of information that may not have previously been included in my WES report I would like for you to issue an updated report 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 WES data that may not have been previously reported.

I hereby authorize Baylor Genetics to conduct genetic testing for myself (or my child) for the Whole Exome Sequencing test as recommended by my physician.

Printed Name | Signature | Date (MM / DD / YYYY)

Relationship to Patient | Proband Name | Proband DOB (MM/DD/YY)

Physician's/Counselor's Signature | Date (MM / DD / YYYY)

FOR SAMPLES SUBMITTED FROM NEW YORK STATE

INITIAL _____ 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.

Consent authorization on next page

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ADDITIONAL AFFECTED SIBLING FOR TRIO REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

Trio WES: (test codes 1600, 1722, 1532, 1533) We understand that our samples will be subjected to Trio WES, and will be analyzed to help interpret the sequence data of our child. A separate parental report will be issued regarding the below two categories of incidental findings. Testing of parental status for these categories of results will be initiated independent of the proband's data. It may be possible to infer information about family member's results based on the proband's or other family member's results. Turnaround time to receive this report is up to 8 weeks.

Proband WES (test codes 1500, 1530, 1531) We understand that our samples will be subjected to targeted testing only (such as Sanger sequencing) and will NOT have WES testing. The laboratory will decide which changes will need parental studies. Testing of parental status for the below two categories of incidental findings will ONLY be initiated if there is a variant identified in the proband.

Please read the below statements carefully and check the appropriate box and initial. Due to the nature of the methodology of this testing we are unable to guarantee that all pathogenic variants in each option will be detected by the WES testing. For options 1 & 2 below: if neither box is checked, or the form is not signed, the lab will default to the NO/ do NOT report option.

MATERNAL REPORTING OPTIONS AND AUTHORIZATION

INITIAL

1. MEDICALLY ACTIONABLE

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 WES report.

_____ ◯ **YES** Please report pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

_____ ◯ **NO** Please do NOT report pathogenic variants in genes included in the ACMG policy statement.

2. CARRIER STATUS FOR AUTOSOMAL RECESSIVE CONDITIONS RECOMMENDED FOR REPRODUCTIVE CARRIER SCREENING

_____ ◯ **YES** Please report carrier status. By checking this box, I choose to receive information regarding carrier status.

_____ ◯ **NO** Please do NOT report carrier status. By checking this box, I choose to NOT receive information regarding carrier status.

Mother's Printed Name	Date of Birth (MM / DD / YYYY)	Mother's Signature	Date (MM / DD / YYYY)
	/ /		/ /

PATERNAL REPORTING OPTIONS AND AUTHORIZATION

INITIAL

1. MEDICALLY ACTIONABLE

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 WES report.

_____ ◯ **YES** Please report pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

_____ ◯ **NO** Please do NOT report pathogenic variants in genes included in the ACMG policy statement.

2. CARRIER STATUS FOR AUTOSOMAL RECESSIVE CONDITIONS RECOMMENDED FOR REPRODUCTIVE CARRIER SCREENING

_____ ◯ **YES** Please report carrier status. By checking this box, I choose to receive information regarding carrier status.

_____ ◯ **NO** Please do NOT report carrier status. By checking this box, I choose to NOT receive information regarding carrier status.

Father's Printed Name	Date of Birth (MM / DD / YYYY)	Father's Signature	Date (MM / DD / YYYY)
	/ /		/ /

FOR SAMPLES SUBMITTED FROM NEW YORK STATE

MOTHER'S INITIAL	FATHER'S INITIAL	
_____	_____	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.

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ADDITIONAL AFFECTED SIBLING FOR TRIO REQUISITION

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RAW DATA CONSENT

☐ By checking this box, I agree to allow Baylor Genetics to provide the raw data such as FASTQ or VCF sequencing files from my genetic test, only upon request, to me, my physician, or the requesting laboratory.

RESEARCH & RECONTACT CONSENT

For more information on research at Baylor Genetics, please visit baylorgenetics.com. Please read the below statements carefully and check the appropriate box.

Note: If left blank, consent is interpreted as "NO."

☐ I agree to use of my de-identified specimen for research to improve genetic testing for all patients and contribute to scientific research.

☐ In addition to agreeing above, I agree to be contacted by Baylor Genetics regarding research opportunities.

CONTACT INFORMATION

Phone # | Alternative Phone # | Email

Address | City | State | Zip

Preferred Method of Contact: ☐ Email ☐ Mail ☐ Phone

INITIAL ☐ **NO** I DO NOT wish to be contacted regarding participation in research studies.

PATIENT AUTHORIZATION

Printed Name | Signature | Date (MM / DD / YYYY)

Relationship to Patient | Patient Name | Patient Date of Birth (MM/DD/YY)

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ADDITIONAL STUDIES - RESEARCH

There may be research studies that you may be eligible for and may be of interest to you. Please read the following statements carefully and check the appropriate box. If the "YES"/contact option is chosen please complete the additional information requested. Please note that if neither box is checked the lab will default to the "NO"/ no contact option.

INITIAL ☐ **YES** Baylor Genetics may share my contact information with researchers who have a Baylor College of Medicine Institutional Review Board (IRB) approved research study for which I may be eligible for participation. There is no obligation to participate if contacted. No information, other than the contact information below, will be provided to the researcher.

Authorization and contact information MUST be completed, or we will not be able to reach you regarding these opportunities.

AUTHORIZATION

Printed Name | Signature | Date (MM / DD / YYYY)

Relationship to Patient | Patient Name | Patient Date of Birth (MM/DD/YY)

CONTACT INFORMATION

Phone # | Alternative Phone # | Email

Address | City | State | Zip

Preferred Method of Contact: ☐ Email ☐ Mail ☐ Phone

INITIAL ☐ **NO** I DO NOT wish to be contacted regarding participation in research studies.

ORDERING PHYSICIAN CONTACT INFORMATION

INITIAL

Physician Last Name | Physician First Name

☐ **YES** Baylor Genetics may contact my/my child's doctor who ordered the Trio Whole Exome Sequencing test to discuss research studies that I/my child may be eligible for. There is no obligation to participate if contacted. If choosing YES, please make sure that the "Authorization" section above is completed.

Phone # | Phone #

Address

☐ **NO** I DO NOT want my/my child's doctor contacted regarding research studies.

City | State | Zip