BAYLOR GENETICS

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

PHONE
713.798.6555
FAX
713.798.2787

CONNECT

WES ADVANTAGE 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 #

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

INSTRUCTIONS FOR ORDERING

Listed below are test codes that when ordered together allow for the most comprehensive assessment to increase the diagnostic yield for patients with an undifferentiated phenotype. Any combination of Chromosomal Microarray Analysis, mtDNA Analysis or Global MAPS can be ordered along with an exome test. For exome testing, select either Trio WES or Proband WES since ordering both Trio and Proband WES is considered duplicate ordering. Parental samples are required for Trio WES and optional for Proband WES.

EXOME TESTS

TRIO WES TESTS

- ☐ 1600 Trio Whole Exome Sequencing
- ☐ 1532 Trio Whole Exome Sequencing + Comprehensive mtDNA Analysis (Send 2 separate EDTA tubes of blood)
- ☐ 1722 Critical Trio Whole Exome Sequencing
- ☐ 1533 Critical Trio Whole Exome Sequencing + Comprehensive mtDNA Analysis (Send 2 separate EDTA tubes of blood)

Add CMA to your Trio test by selecting below

- ☐ 8665 Chromosomal Microarray Analysis (CMA) - HR + SNP Screen (Comprehensive)

CORRESPONDING PARENTAL TESTS
(Both Parents Are Required)

- ☐ 1550 Parental WES - Maternal
- ☐ 1550 Parental WES - Paternal

PROBAND WES TESTS

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

CORRESPONDING PARENTAL TESTS
(Send within 2 weeks of proband sample)

- ☐ 1505 Parental Sanger - Maternal
- ☐ 1505 Parental Sanger - Paternal

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
- ☐ 4902 Global Metabolomic Assisted Pathway Screen - Cerebrospinal Fluid

Add CMA or Mitochondrial testing to your Global MAPS test by selecting below

- ☐ 8665 Chromosomal Microarray Analysis (CMA) - HR + SNP Screen (Comprehensive)
- ☐ 2055 Comprehensive mtDNA Analysis by NGS

SAMPLE

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

- ◯ Blood in EDTA
- ◯ Cerebrospinal Fluid (Global MAPS only)
- ◯ Cord Blood (Call lab for sample specification)
- ◯ Cultured Skin Fibroblast
- ◯ Extracted DNA from ____
- ◯ Liver (mtDNA Analysis only)
- ◯ Plasma from EDTA (Global MAPS only)
- ◯ Saliva (CMA only)
- ◯ Skeletal Muscle (mtDNA Analysis only)
- ◯ Tissue (mtDNA Analysis only)
- ◯ Urine (Global MAPS only)

BIOLOGICAL PARENTS INFORMATION

BIOLOGICAL PARENTS SAMPLES ARE REQUIRED FOR TRIO WES; Other family members cannot be substituted for either parent. Send 10 cc blood in an EDTA tube for each parental sample. Be sure to label parental samples with full name and date of birth - DO NOT LABEL WITH CHILD'S NAME. Must sign 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) ____ / ____ / ____

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

Sample Type:
- ◯ Blood
- ◯ Saliva (only for 1505)

PATERNAL INFORMATION

☐ Asymptomatic ☐ Symptomatic (Attach summary of findings)

Paternal Last Name | Paternal First Name | MI

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

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

Sample Type:
- ◯ Blood
- ◯ Saliva (only for 1505)

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

ITEM CHECKLIST FOR TESTING

- ☐ Proband Sample (Required)
- ☐ Maternal Sample (Required for Trio)
- ☐ Paternal Sample (Required for Trio)
- ☐ Signed WES Consent Form
- ☐ Clinical Note/Summary
- ☐ Requisition
- ☐ Indication for Study

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

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

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			/ /	

INFORMATION AND CONSENT FOR TESTING

-- Also available in other languages at BMGL.com under the Testing tab.

Your physician has advised you (or your child) to undergo the genetic test called Whole Exome Sequencing (referred to WES). The purpose of this document is to provide information about the test. This information is meant to be used as a supplement to your discussion with your health care professional. The decision to undergo WES is made by you and your physician. In general, the test is used when your medical history and physical exam findings strongly suggest that there is a genetic cause for your medical issues. If you agree to have the WES test, you will be asked to sign the pages that follow in this document, indicating that you understand the information provided and wish to have testing. You will be given a copy of this document for your records.

Test order 1531, 1532 and 1533, in addition to WES analysis as detailed below, will also include a separate analysis of the mitochondrial DNA. To learn more about this testing please visit our website, test code 2055 Comprehensive mtDNA Analysis by Massively Parallel Sequencing (MitoNGSSM). This is the evaluation of the entire mitochondrial genome for point mutations and deletions. The detection threshold of massively parallel sequencing analysis for heteroplasmic mitochondrial DNA point mutations is approximately 1.5%. 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. BMGL will NOT automatically initiate testing on the maternal sample, if this is desired please contact client services for assistance.

Test order 1530, in addition to Proband WES analysis as detailed below, will also include a separate analysis for detection of deletions and duplications plus a screen for detection of uniparental disomy (UPD) and absence of heterozygosity (AOH). To learn more about this testing please visit our website, test code 8665 Chromosomal Microarray Analysis - HR + SNP Screen (Comprehensive). 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. BMGL will NOT automatically initiate testing on the parental sample(s), if this is desired please contact client services for assistance.

Test order 4900, 4901 and 4902 (Global MAPS) is a large scale, semi-quantitative screening test that looks at perturbations in both individual analytes and pathways related to biochemical abnormalities, including but not limited to amino acid, organic acid, lipid, and nucleotide metabolism. This is a small molecule screen for compounds ranging in size from 50-1500 Da. 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 on the Metabolomic Profile should be confirmed by diagnostic biochemical or molecular diagnostic testing. Consent for testing below is only for WES and does not need to be completed if only Global MAPS is being ordered.

DESCRIPTION OF WHOLE EXOME SEQUENCING TEST

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.

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 information in the medical literature and in scientific databases, we will decide whether any of these variations are predicted to be causative or related to your medical condition. The report will contain results that may explain the cause of your current medical problems. It may also contain information on 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.

In addition, it may also contain information in the following categories:
Category I: Medically Actionable. The report may also contain information on 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 these types of medically actionable or incidental findings (PMID: 23788249, 27854360). These guidelines include a list of genes, which may be updated periodically, that have been determined to be considered medically actionable and therefore laboratories should seek and report pathogenic variants in these genes. In accordance with an update to this policy statement (PMID: 25356965), there is the option to opt-out of receiving pathogenic variants if identified in the genes listed in ACMG policy statement.

Category II: Carrier Status. Carrier status for autosomal recessive conditions will include disorders recommended for reproductive screening by professional societies such as ACMG or ACOG, which includes: Cystic fibrosis (CFTR), Sickle cell anemia (S allele, HBB), Familial dysautonomia (IKBKAP), 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), Hemolytic anemia due to G6PD deficiency (G6PD* X-linked inheritance).

See the following pages for options regarding receipt of certain categories of results in the report.

Consent continued on next page

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WES ADVANTAGE REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

Because medical information continues to advance, it is important to know that the interpretation of the variants is based on information available at the time of testing and may change in the future. As determined necessary by the laboratory the patient's sample will have certain findings confirmed by a second methodology (Sanger sequencing). The report will not include findings in genes causing adult onset dementia syndromes for which there is presently no prevention or cure. If the proband has a phenotype that clearly indicates such a disorder we recommend pursuing targeted testing based on phenotype and not WES testing. However, please note that if the patient has a clinical presentation that could indicate such a disorder or a mixed neurological phenotype, then results may be reported in the proband and the parents for genes that have an allelic association with dementia or dementia is a component of the phenotype. 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. Please 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 de novo findings may be significant in determining the cause of you/your child's 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 or homozygous variants in genes where each parent has one change and the affected individual has inherited both changes, for genes with or without a known association with disease. It is important to note that the Trio WES report may contain information on 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.

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. Once the focused report is received, the expanded report can be ordered (no additional charge). The expanded report may contain information on 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). In discussion with your physician, the expanded report can be ordered for up to 6 months after the focused report is received, for no additional charge. A requisition for ordering the expanded report is available on our website.

BIOLOGICAL PARENTAL SAMPLES

TRIO (test codes 1600, 1722, 1532, 1533) As part of the Trio WES test, blood samples from the biological parents of the proband are required. Trio Whole Exome Sequencing (Trio WES) will be performed on the proband and parental samples concurrently 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 two categories of incidental findings. See the following pages for options regarding receipt of certain categories of results in the report, and see the previous sections "medically actionable" and "carrier status" for descriptions of these two categories.

PROBAND WES (test codes 1500, 1530, 1531) 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, etc. 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 for these categories of results will ONLY be initiated if there is a variant identified in the proband. See the following pages for options regarding receipt of certain categories of results in the report, and see the previous sections "medically actionable" and "carrier status" for descriptions of these two categories.

POTENTIAL RISKS 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 the WES, we may perform a separate genetic test to confirm that the samples that were submitted from the parents were correctly 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 sample 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 receive the results. However, if you withdraw consent for testing after 5 p.m. the next business from the day of sample receipt by the laboratory, you will be charged for the full cost of the test.
(6) The cumulative results of WES testing on many samples may be published in the medical literature to contribute knowledge to the medical profession. It is unlikely that these publications will include any information that will identify you personally.
(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.

Due to the complex nature of the WES testing it is recommended that families seek genetic counseling in conjunction with testing.

Consent continued on next page

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CONNECT

WES ADVANTAGE 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.

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

_____ Specimen Retention: My sample shall be destroyed at the end of the testing process or not more than 60 days after completion of testing. However, I hereby authorize the lab to retain my sample(s) for a longer retention in accordance to the laboratory retention policy for internal laboratory quality assurance studies and possible research testing.

Consent authorization on next page

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WES ADVANTAGE 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 _____

Specimen Retention: My sample shall be destroyed at the end of the testing process or not more than 60 days after completion of testing. However, I hereby authorize the lab to retain my sample(s) for a longer retention in accordance to the laboratory retention policy for internal laboratory quality assurance studies and possible research testing.

SEE NEXT PAGE FOR POTENTIAL RESEARCH OPPORTUNITY

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

ADDITIONAL STUDIES - RESEARCH

The ordering physicain may be contacted regarding research opportunities regarding your results/data. Additionally there may be instances in which the laboratory or other researchers would contact you directly regarding 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. Other than the contact information below, the researcher will only be provided with limited geneotype and phenotype information.

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.