

## WHOLE GENOME SEQUENCING (WGS) 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): \_\_\_\_\_

### ORDERING PHYSICIAN

### ADDITIONAL REPORTS

Ordering Physician \_\_\_\_\_  
 Institution Name \_\_\_\_\_  
 Email (Required for International Clients) \_\_\_\_\_  
 Phone \_\_\_\_\_ Fax \_\_\_\_\_

Name \_\_\_\_\_  
 Email \_\_\_\_\_  
 Phone \_\_\_\_\_  
 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) 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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### 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. Global MAPS® can be ordered along with a genome test, however the turnaround time for results will differ from genome sequencing. Parental samples are required for Trio WGS, Duo WGS, and optional for Proband WGS.

### TRIO WGS TEST OPTIONS

- 1800 Trio Whole Genome Sequencing
- 1822 Rapid Trio Whole Genome Sequencing

- CORRESPONDING PARENTAL TESTS (Both Biological Parents Are Required)
- 1850 Parental WGS - Maternal
  - 1850 Parental WGS - Paternal

### DUO WGS TEST OPTIONS

- 1803 Duo Whole Genome Sequencing
- 1823 Rapid Duo Whole Genome Sequencing

- CORRESPONDING PARENTAL TESTS (One Biological Parent Is Required)
- 1850 Parental WGS - Maternal
  - 1850 Parental WGS - Paternal

### PROBAND WGS TEST

- 1810 Proband Whole Genome Sequencing
- 1829 Rapid Proband Whole Genome Sequencing

### ADD-ON TEST

**GLOBAL MAPS® TESTS**

- 4900 Global Metabolomic Assisted Pathway Screen - Plasma from EDTA
- 4901 Global Metabolomic Assisted Pathway Screen - Urine

Was plasma extracted from EDTA?       Yes       No

### PROBAND SAMPLE(S)

Please refer to [www.baylorgenetics.com](http://www.baylorgenetics.com) for full sample requirements.

- Blood in EDTA (preferred)
- Buccal Swab
- Saliva
- Cord Blood
- Cultured Skin Fibroblast
- Extracted DNA from Blood
- Extracted DNA from Cultured Skin Fibroblasts

\_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_  
 Date of Collection  
 (MM / DD / YYYY)

**NOTE:** For ultra-rapid WGS testing options, only blood in EDTA will be accepted. 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 WGS. 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)      \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Date of Collection  
 (MM / DD / YYYY)      \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

- Sample Type:
- Blood in EDTA (preferred)
  - Buccal Swab
  - Saliva

#### 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 in EDTA (preferred)
  - Buccal Swab
  - Saliva

## WHOLE GENOME SEQUENCING (WGS) REQUISITION

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### ITEM CHECKLIST FOR TESTING

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

### 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 genome 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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MI \_\_\_\_\_

Date of Birth (MM / DD / YYYY) \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Biological 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 Atrial 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

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### INDICATION FOR TESTING (REQUIRED) - CONTINUED

**ENDOCRINE** ..... **HEMATOLOGY** ..... **OTHER** .....

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

- 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
- \_\_\_\_\_
- \_\_\_\_\_

- 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
- \_\_\_\_\_
- \_\_\_\_\_

**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
- \_\_\_\_\_
- \_\_\_\_\_

**CANCER** .....

- Type of Cancer \_\_\_\_\_
- Age of Diagnosis \_\_\_\_\_
- Family History of Cancer and Affected Relatives \_\_\_\_\_
- \_\_\_\_\_
- \_\_\_\_\_

**GENES OF INTEREST** .....

\_\_\_\_\_  
 \_\_\_\_\_  
 \_\_\_\_\_  
 \_\_\_\_\_

### ADDITIONAL CLINICAL INFORMATION

### DIFFERENTIAL DIAGNOSIS

Consent on next page



## WHOLE GENOME SEQUENCING CONSENT

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

### INFORMATION AND CONSENT FOR TESTING

The Whole Genome Sequencing (WGS) test is a highly complex test developed for the identification of changes in an individual's DNA that are causative or related to their medical concerns. The WGS test provides a comprehensive analysis of the human genome by assessing for a wide range of errors in DNA, ranging from single nucleotide variants to alterations involving large segments of genetic information. In contrast to other sequencing tests that analyze a single gene or groups of related genes, WGS will analyze the complete genetic code. Therefore, WGS is thought to be an efficient method of analyzing a patient's DNA to evaluate for a genetic cause of diseases or disabilities. You may consider discussing the significance of your results with your healthcare provider or genetic counselor.

### TESTING REPORTING

When your genome sequence is compared to a standard reference sequence, many variations or differences are expected to be found. In addition to comparison to a standard reference sequence, the proband's (affect individual) sequence will be compared to the sequence of other family members who undergo the same testing (comparators). Currently available medical and scientific information will be used to decide whether any of these variations are predicted to be causative or related to your medical concerns. The report may contain results that 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 the findings relate to your current symptoms.

**You may receive 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 WGS. This does not mean there is no genetic change, but it may mean that WGS could not detect it.
- **Results of Unclear Significance:** WGS can detect change(s) in DNA that do not have clear meaning. These alterations are also referred to as variants of uncertain significance (VUS). Additional studies may be recommended if a VUS is identified in a gene that may be associated with your medical concerns.
- **Secondary Findings:** WGS 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 for secondary findings:

The report may also contain information regarding genes and diseases that are considered secondary findings 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 secondary findings (PMID: 34012068). These guidelines include a list of genes (updated periodically) that are considered medically actionable and thus, laboratories should seek and report pathogenic (disease causing) or likely pathogenic findings in these genes. In accordance with an update to this policy statement (PMID: 25356965), there is the choice to opt-out of receiving this information.

### ADDITIONAL REPORTING

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. 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 WGS is available for request once a WGS report has been issued. Please see our website for further information regarding this.

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 WGS testing. However, if the reason for testing includes a clinical presentation that could include such a disorder or a mixed neurological phenotype, then results may be reported in the proband (patient), and the patient's parents for genes that have an association with dementia or are a component of the phenotype.

#### ADDITIONAL REPORTING FOR TRIO WGS (TEST CODES 1800, 1822): .....

As part of the Trio WGS analysis, we will report findings in genes that have occurred in the affected individual, but not in the asymptomatic parents or comparators. 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 of potential deleterious effects 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 WGS 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 WGS test, blood samples from the biological parents of the patient are required. Trio WGS 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 secondary findings.

#### ADDITIONAL REPORTING FOR DUO WGS (TEST CODES 1803, 1823): .....

As part of the Duo WGS analysis, we will report findings in genes that have occurred in the affected individual, but not in the asymptomatic parent or comparator. 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 of potential deleterious effects in genes where the parent or comparator has one change and the affected individual has inherited the change, for genes with or without a known association with disease. It is important to note that the Duo WGS 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 Duo WGS test, a blood sample from the biological parent of the proband are required. Duo WGS will be performed on the proband and parental sample 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 secondary findings.

## WHOLE GENOME SEQUENCING CONSENT

\_\_\_\_\_  
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Date of Birth (MM / DD / YYYY)                      Biological Sex

### CONSIDERATIONS AND LIMITATIONS

1. It is possible that you could have a variant in a gene included in the WGS test, but the WGS test was unable to detect the variant. Therefore, it is possible that you may be affected with one of the conditions tested by WGS, but that the test did not detect the condition.
2. The WGS test does not analyze 100% of the genes in the human genome. There are some genes and non-coding regions that cannot be included in the test due to technical reasons.
3. Results may be inconclusive 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 misidentified parentage (the father or mother of the individual is not as believed) or may detect consanguinity (reproductive partners are blood relatives). Since the accurate assignment of family relationships is critical to the analysis of WGS, we will 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 contact the referring provider prior to proceeding with testing.
5. If you sign the consent form, but you no longer wish to have your samples tested by WGS, 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 5pm 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 WGS 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. Because 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 WGS. 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.
8. It is possible that even if WGS identifies the underlying genetic cause for the disorder in your family, this information may not help in predicting prognosis, management, or treatment of disease.
9. Results will only be released to a licensed healthcare provider, to those allowed access to test results by law, and to those authorized in writing.
10. Samples will be retained in the laboratory in accordance with the laboratory retention policy.

### PATIENT 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 WGS testing. Please refer to the Baylor Genetics website for up-to-date information on the detectable range of the WGS test for various mutation types. For Options 1 & 2: If neither box is checked, or if form is not signed, consent is interpreted as "NO."

#### 1. SECONDARY FINDINGS

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

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

#### 2. 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 WGS test. The current schedule for this review is every 12 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 the information that may not have previously been included in my WGS report, I would like for you to issue an updated report to my physician who ordered this WGS testing.  
 NO Please do NOT issue an updated report if there is new information regarding the clinical significance of my WGS 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 Genome 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)

**WHOLE GENOME SEQUENCING CONSENT**

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

**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.

**TRIO WGS: (TEST CODES 1800, 1822)**

We understand that our samples will be subjected to Trio WGS and 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 these categories of results will be initiated independently of the patient's data. It may be possible to infer information about a family member's results based on the patient's or other family member's results.

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 WGS testing. For the parental options below: if neither box is checked, or the form is not signed, consent is interpreted as "NO."

**DUO WGS: (TEST CODES 1803, 1823)**

I understand that my sample will be subjected to Duo WGS and will be analyzed to help interpret the sequence data of my child. A separate parental report will be issued regarding the below category of secondary findings. Testing of parental status for these categories of results will be initiated independently of the patient's data. It may be possible to infer information about a family member's results based on the patient's or other family member's results.

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 WGS testing. For the parental options below: if neither box is checked, or the form is not signed, consent is interpreted as "NO."

**MATERNAL REPORTING OPTIONS AND AUTHORIZATION**

**1. SECONDARY FINDINGS**

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

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

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

**PATERNAL REPORTING OPTIONS AND AUTHORIZATION**

**1. SECONDARY FINDINGS**

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

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

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

**FOR SAMPLES SUBMITTED FROM NEW YORK STATE**

\_\_\_\_\_  
Mother's Initial      \_\_\_\_\_  
Father's Initial      **Specimen Retention:** By leaving this section blank, my sample shall be destroyed at the end of the testing process or not more than 60 days after completion of testing. 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.



