BAYLOR GENETICS

**BAYLOR GENETICS**
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**CONNECT**

# WHOLE GENOME SEQUENCING (WGS) CONSENT

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

## TEST INFORMATION

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

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

## RESULTS

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

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

## INCIDENTAL FINDINGS

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

### CATEGORY I: ACMG SECONDARY FINDINGS

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

### CATEGORY II: OTHER INCIDENTAL FINDINGS

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

## ADDITIONAL REPORTING INFORMATION

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

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

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

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

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## RNASEQ INFORMATION

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

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

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

## CONSIDERATIONS AND LIMITATIONS

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

## PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION

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

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### PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION CONTINUED

**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 initiating here, I hereby authorize the lab to retain my sample(s) for longer retention in accordance with the laboratory retention policy for internal laboratory quality assurance studies and possible research testing.

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

### PATIENT REPORTING OPTIONS AND RELEASE OF UPDATED RESULTS

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

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

**FOR ALL WGS:**

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

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

☐ YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

☐ NO - Please do NOT report pathogenic and likely pathogenic variants in genes included in the ACMG policy statement.

**OPTION TO ALLOW RELEASE OF UPDATED RESULT**

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

☐ YES - If new information regarding the clinical significance of changes in my/my child's WGS becomes known, I would like Baylor Genetics to issue an updated report which includes this information 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/my child's WGS that becomes known.

**FOR DUO AND TRIO WGS ONLY:**

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

**REPORTING OF MATERNAL CATEGORY I (ACMG) SECONDARY FINDINGS**

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

☐ YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

☐ NO - Please do NOT report pathogenic or likely pathogenic variants in genes included in the ACMG policy statement.

**REPORTING OF PATERNAL CATEGORY I (ACMG) SECONDARY FINDINGS**

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

☐ YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

☐ NO - Please do NOT report pathogenic or likely pathogenic variants in genes included in the ACMG policy statement.

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

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

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

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

☐ YES - Please report pathogenic and likely pathogenic variants in genes determined to be medically actionable by the ACMG policy statement.

☐ NO - Please do NOT report pathogenic or likely pathogenic variants in genes included in the ACMG policy statement.

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## FINANCIAL AGREEMENT AND GUARANTEE

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

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

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

## RECONTACT FOR RESEARCH CONSENT

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

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

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

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

☐ Email ☐ Phone ☐ Mail

## PATIENT AUTHORIZATION

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

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

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

Patient's Parent / Personal Representative* Name | Patient's Parent / Personal Representative Signature | Date Signed (MM / DD / YYYY)

Relationship of Personal Representative* to the Patient

Ordering Provider's Signature | Date Signed (MM / DD / YYYY)

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| | | | / / | |

### PATIENT AUTHORIZATION

**FOR DUO AND TRIO WGS ONLY**

| | | |
|---|---|---|
| Maternal Name | Maternal Signature | Date Signed (MM / DD / YYYY) |
| Paternal Name | Paternal Signature | Date Signed (MM / DD / YYYY) |
| Maternal Personal Representative* Name | Maternal Personal Representative* Signature | Date Signed (MM / DD / YYYY) |
| Relationship of Maternal Personal Representative* | | Date Signed (MM / DD / YYYY) |
| Paternal Personal Representative* Name | Paternal Personal Representative* Signature | Date Signed (MM / DD / YYYY) |
| Relationship of Paternal Personal Representative* | | Date Signed (MM / DD / YYYY) |

**FOR AFFECTED SIBLING OR OTHER FAMILY MEMBER WGS ONLY**

| | | |
|---|---|---|
| Affected Sibling/Other Family Member Name | Affected Sibling/Other Family Member Signature | Date Signed (MM / DD / YYYY) |
| Affected Sibling/Other Family Member Parent / Personal Representative* Name | Affected Sibling/Other Family Member Parent / Personal Representative* Signature | Date Signed (MM / DD / YYYY) |
| Relationship of Personal Representative* to Affected Sibling / Other Family Member | | Date Signed (MM / DD / YYYY) |