



PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

PATIENT INFORMATION (COMPLETE ONE FORM FOR EACH PERSON TESTED)

Fetus of: _____		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:		Biological Sex:		
				<input type="radio"/> Yes	<input type="radio"/> No	<input type="radio"/> Female	<input type="radio"/> Male	<input type="radio"/> 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
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☐ **INSURANCE**

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

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)
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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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PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

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

ETHNICITY

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

TESTING OPTION

- ☒ 1622 Prenatal Trio Whole Exome Sequencing

GESTATIONAL INFORMATION

NOTE: Providing U/S dating allows for the best handling of the specimen in the lab and improves performance of AFAFP analysis.

U/S Date (MM / DD / YYYY)

LMP Date (MM / DD / YYYY)

Gestational Age on U/S Date:

Weeks Days

SAMPLE

Performing Physician Date of Collection (MM / DD / YYYY)

SAMPLE TYPE

- ☐ Cultured Amniocytes ☐ Amniotic Fluid¹ _____ cc
- ☐ Cultured CVS ☐ CVS¹ _____ mg ☐ TA ☐ TC
- ☐ Extracted DNA² from: _____

1: If direct specimen is submitted, it will be cultured.

2: Extracted DNA is only acceptable from cultured fetal specimen.

Prior to ordering Prenatal Trio WES testing, you must call the lab and discuss the clinical history and sample requirements with a genetic counselor. Please call 713-798-6555.

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

Specimen Requirements/Order Discussed with: Name of Baylor Genetics Genetic Counselor Date of Collection (MM / DD / YYYY)

Additional Cultures to be sent later: ☐ Yes ☐ No Cultures will be sent from (Name of Laboratory)Has prior testing been performed at Baylor Genetics? ☐ Yes ☐ No If YES, provide Baylor Genetics Family #

BIOLOGICAL PARENTS INFORMATION

BOTH BIOLOGICAL PARENTS SAMPLES ARE REQUIRED. Testing cannot proceed unless BOTH parental samples are received. If BOTH biological parents are not available, then this test CANNOT be ordered. Please call 713-798-6555 to discuss other testing options. Send 10 cc blood in an EDTA tube for each parental sample OR collect with ORAcollection Dx (OCD-100) self-collection kit. 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. Turnaround time is 3 weeks AFTER completion of sample culture.

1550 | MATERNAL INFORMATION

- ☐ Asymptomatic ☐ Symptomatic (Attach summary of findings)

Maternal Last Name Maternal First Name MI

Sample Type:
☐ Blood
☐ Buccal Swab

Maternal Date of Birth (MM / DD / YYYY)

Date of Collection (MM / DD / YYYY)

1550 | PATERNAL INFORMATION

- ☐ Asymptomatic ☐ Symptomatic (Attach summary of findings)

Paternal Last Name Paternal First Name MI

Sample Type:
☐ Blood
☐ Buccal Swab

Paternal Date of Birth (MM / DD / YYYY)

Date of Collection (MM / DD / YYYY)

ITEM CHECKLIST

- ☐ Fetal Sample ☐ Consent Form Signed by All Individuals Tested ☐ Maternal Sample (EDTA Required)
- ☐ Requisition ☐ Clinical Note/Summary ☐ Paternal Sample (EDTA Required)
- ☐ Indication for Study Checklist ☐ Pedigree

02.27.20

PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

Your physician has advised you to undergo the genetic test called the Prenatal Trio Whole Exome Sequencing (referred to as Prenatal Trio WES) for your pregnancy. 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. If you agree to have the Prenatal Trio WES test, the mother of the fetus will be asked to sign the last page(s) of this document, indicating that she understand the information provided and wish to have testing. You will be given a copy of this document for your records.

DESCRIPTION OF THE PRENATAL TRIO WHOLE EXOME SEQUENCING TEST

The Prenatal Trio WES test is a highly complex test that is newly developed for the identification of changes in an individual's DNA that are causative or related to their medical concerns. This test differs from other genetic tests in that a sample from your baby (fetal sample) is tested together with his or her parents and the results interpreted as a family. This approach to testing can be helpful in identifying genetic causes of a medical condition. Analyzing the data for changes that occur in the fetus, but not in the parents, can help to identify new mutations in genes that may be causative of fetus' disease (de novo changes). In other cases, following the inheritance of changes from parent(s) to fetus can also aid in the identification of potentially causal disease genes.

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 current sequencing tests that analyze one gene or small groups of related genes at a time, the Prenatal Trio Whole Exome Sequencing test 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 an individual's DNA to discover the genetic cause of diseases or disabilities. However, it is possible that even if the Prenatal Trio 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.

INDICATIONS FOR TESTING

The decision to undergo the Prenatal Trio Whole Exome Sequencing test is made by you and your physician. In general, the test is used when fetal imaging and family medical history strongly suggest that there is a genetic cause for the fetal medical issues.

TESTING REPORTING

When the fetal 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 baby's medical condition.

The report will contain results that may explain the cause of your baby's current medical concerns. As part of the Prenatal Trio WES analysis, we will report genetic variants/changes currently with a known association with disease that may be significant in determining the cause of the fetus' medical condition. Those genetic changes include de novo changes, i.e. changes that have occurred in the fetus, but not in the asymptomatic parents and compound heterozygous or homozygous variants in genes where each parent has one change and the affected individual has inherited both changes. It is important to note that the Prenatal Trio WES report may contain information on diseases and genes that do not relate to the fetus' current condition, but the disease may develop years from now during childhood, according to current knowledge.

In addition, an incidental findings report can be requested regarding medically actionable and carrier status information once the baby is born (see the parental section for details regarding these two categories). A separate test order must be submitted to receive this information.

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).

REPORT EXCLUSIONS

The report will not include findings in genes causing adult onset such as dementia syndromes for which there is presently no prevention or cure. We expect to find a large number 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 the Prenatal Trio WES is available for request once a Prenatal Trio WES report has been issued. Please see our website for further information regarding this.

REQUIREMENT FOR BIOLOGICAL PARENTAL SAMPLES

As part of the Prenatal Trio WES test, blood samples from the biological parents of the proband are required. Prenatal Trio Whole exome sequencing (Prenatal 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, with a turnaround time of 10 weeks. See the following pages for options regarding receipt of these categories of results in parental report.

Consent continued on next page



PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

As part of the Prenatal Trio WES test, blood samples from the biological parents of the proband are required. Prenatal Trio Whole exome sequencing (Prenatal 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.

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. The American College of Medical Genetics (ACMG) has published guidelines for the reporting of these types of medically actionable or incidental findings (PMID: 23788249). 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 (ACMG.net), there is the option to opt out of receiving pathogenic variants information, 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), Mucopolidosis IV (MCOLN1), Gaucher disease Type I (GBA).

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, with a turnaround time of 10 weeks. See the following pages for options regarding receipt of these categories of results in parental report.

Potential Risks and Discomforts

- (1) It is possible that fetus could have a variant in a gene included in the Prenatal Trio WES test, but the Prenatal Trio WES test was unable to detect the variant. Therefore, it is possible that fetus may be affected with one of the conditions tested by Prenatal Trio WES, but that the test did not detect the condition.
- (2) The Prenatal Trio 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 fetus 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 Prenatal Trio WES, 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, you will be notified through your physician and the Prenatal Trio WES testing will be cancelled.
- (5) If you sign the consent form, but you no longer wish to have your families sample tested by Prenatal Trio 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 Prenatal Trio WES testing on many samples may be published in the medical literature. These publications will not include any information that will identify your family personally.
- (7) Due to the fact that many different genes and conditions are being analyzed, there is a risk that you will learn genetic information about your fetus, yourself or your family that is not directly related to the reason for ordering the Prenatal Trio WES. This information might relate to diseases with symptoms that may develop in the future in your fetus, yourself or your family members as well as conditions that have no current treatment. If you have concerns about learning about other diseases unrelated to the current medical problems, please tell your doctor so that the results will not include this information.

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

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 continued on next page



PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

FETAL REPORTING OPTION AND AUTHORIZATION FOR TESTING

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 Prenatal Trio 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.

If neither box is checked the lab will default to the YES/Report option.

INITIAL

- _____ ☐ **YES** If new information is known regarding clinical significance of information that may not have previously been included in my Prenatal Trio WES report I would like for you to issue an updated report to my physician who ordered this Prenatal Trio WES testing.
- _____ ☐ **NO** Please do NOT issue an updated report if there is new information regarding the clinical significance of my Prenatal Trio WES data that may not have been previously reported.

Mother's Signature

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

Mother's Printed Name

_____/_____/_____
Maternal DOB (MM/DD/YY)

Physician's/Counselor's Signature

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

PARENT REPORTING OPTIONS AND AUTHORIZATION

Confirmation of Parentage:

I understand that the accurate assignment of family relationships is critical to the analysis of the Trio WES, therefore the laboratory will perform a separate genetic test to confirm that the samples that were submitted from the parents and child were correctly identified. If a discrepancy is identified, we will proceed with testing our child's sample with a revised test order to Proband WES (test code 1500) with expedited turnaround time.

_____ Mother's Initials

_____ Father's Initials

We hereby authorize Baylor Genetics to conduct genetic testing on our samples (biological parents) for the purposes of clarifying results for the Prenatal Trio Whole Exome Sequencing test (Prenatal Trio WES) that is being performed on our baby's prenatal sample as recommended by our child's physician. We understand that our samples will be subjected to Trio WES, and will be analyzed to help interpret the sequence data of our baby's prenatal sample. A separate parental report will be issued regarding the below two categories of incidental findings, with a turnaround time of up to 10 weeks. It may be possible to infer information about family member's results based on the proband's or other family member's results.

Consent continued on next page



PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

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

INFORMATION AND CONSENT FOR TESTING

MATERNAL 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 Trio 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.

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 Trio 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 Signature

Date (MM / DD / YYYY)

Mother's Printed Name

Maternal 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.

PATERNAL 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 Trio WES testing.

For options 1 & 2 below: if neither box is checked, or if the 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 Trio 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 Signature

Date (MM / DD / YYYY)

Father's Printed Name

Paternal 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.

SEE NEXT PAGE FOR POTENTIAL RESEARCH OPPORTUNITY



PRENATAL TRIO WHOLE EXOME SEQUENCING REQUISITION

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

ADDITIONAL STUDIES - RESEARCH

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

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

INITIAL

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

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

INITIAL

ORDERING PHYSICIAN CONTACT INFORMATION

_____ INITIAL

_____ Physician Last Name

_____ Physician First Name

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

☐ **YES**

_____ Phone #

_____ Fax #

_____ Address

☐ **NO**

I DO NOT want my/my child's doctor contacted regarding research studies.

_____ City

_____ State

_____ Zip