Prenatal Exome Request Form – Maternal/Fetal

Greenwood Genetic Center
106 Gregor Mendel Circle • Greenwood, SC 29646
Toll Free: (800) 473-9411 • Fax: (864) 941-8141
Website: www.ggc.org Highlighted boxes are required

LAB USE ONLY

Patient Information (Please Print):

Last Name First MI			Address
Race ☐ B ☐ W ☐ Other:	Sex ☐ M ☐ F	Maternal DOB MM/DD/YYYY	City, State, Zip
Specimen Collection Date MM/DD/YYY	Type of specimen	Numeric Identifier (Medical record # or SSN)	Home telephone

Referring Physician:

Name	Address	
Institution	City, State, Zip	
NPI#	Telephone	Fax
Email Address:	Preferred Method to Receive Results: ☐ Secure Email ☐ Fax ☐ Regular Mail	

Additional report to: ☐ **Genetic Counselor** ☐ **Institution** ☐ **Care Coordinator** ☐ **Other:**

Name			Address
Telephone	Fax	Email:	City, State, Zip

Additional report to: ☐ **Genetic Counselor** ☐ **Institution** ☐ **Care Coordinator** ☐ **Other:**

Name			Address
Telephone	Fax	Email:	City, State, Zip

Billing: Select how the test(s) will be billed & complete the billing information on the next page. The BILLING FORM on page 2 is required.

☐ **Institutional Billing: Complete section 1 on the separate BILLING FORM (page 2)**
☐ **Insurance: Complete section 2 on the BILLING FORM (page 2). No out-of-state (non-SC) insurance or Medicaid will be accepted.**
☐ **Self-pay: Complete section 3 on the separate BILLING FORM (page 2).**

Indication for Study

☐ Prenatal Findings; ICD-10 Code(s): _____

☐ Family History; ICD-10 Code(s): _____

Please complete the required *Clinical Information Form* (page 3).

Is the mother affected: ☐ Yes ☐ No EDC (MM/DD/YYYY): _____

If Yes, please list clinically relevant features: _____

_____

☐ **Prenatal Exome Sequencing (DUO analysis)**

☐ **Prenatal Exome Sequencing (TRIO analysis)**

☒ **Maternal Cell Contamination Studies** (required)

Specimen Requirements

Amniotic Fluid: Direct amniotic fluid will be accepted for analysis given there is sufficient volume for back-up culture to be established. A back-up culture at reference lab OR Greenwood lab is required.
Cultured amniocytes: 2x T25 confluent flasks
Extracted DNA: A minimum of 3 micrograms of extracted DNA is required.
Maternal Sample: A maternal sample is required. Accepted sample types include whole blood in an EDTA/lavender top tube, extracted DNA, or saliva/saliva swab.
For **CVS sample**, please contact the laboratory to determine if sample can be accepted for PES.

Ordering Checklist:

- ☐ Test Requisition & Informed consent for prenatal/maternal analysis
- ☐ Fetal Sample & Maternal Sample
- ☐ Clinical information page & Pedigree

If Trio Analysis is being requested, these are also required:

- ☐ Paternal Request and Consent
- ☐ Paternal Sample

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EDTA RT / R / F	Na Hep RT / R / F	Plasma RT / R / F	Urine / Flasks / Other RT / R / F	Serum / Tissue RT / R / F	DBS / DNA RT / R / F

June 2020

Greenwood Genetic Center

Diagnostic Laboratory Billing Form

This page is required to process any test requests.

LAB USE ONLY

- **Out of State (non-SC) commercial insurance can only be filed for NGS Panels.**
- **No out of state Medicaid will be accepted for any tests.**
- **The following items are needed in order to bill the patient's insurance directly. We will not be able to file the claim if we are missing information.**
  - ☐ **This form must be completed with ALL requested information.**
  - ☐ **A legible copy of both sides of the insurance card**
  - ☐ **Authorization number, authorization letter, or letter of agreement from insurance company**

Patient Information:

Last Name First MI		Address	
Numeric Identifier (Medical record # or SSN)	Maternal DOB MM/DD/YYYY	City, State, Zip	Telephone
ICD10 Code(s)			

Section 1: Institutional Billing

Complete section below with institution information. *New clients must complete an INSTITUTIONAL ACCOUNT REQUEST FORM when submitting the order.* Please contact the GGC Billing Office at 864-941-8117 or billing@ggc.org with any questions about your account.

Institution/Organization	Contact Name:	Email:
Billing Address	City, State, Zip	
Account Number:	Telephone	Fax

Section 2: Insurance Information

MUST INCLUDE LEGIBLE COPY OF INSURANCE CARD (FRONT & BACK)
All information required to file insurance claims.

Primary

Insured/Policy Holder Name:	Policy Holder DOB:	Policy Holder Gender ☐ Male ☐ Female
Relationship to Patient ☐ Self ☐ Spouse ☐ Dependent ☐ Other:	Policy #	
Insurance Company Name:	Insurance ID #:	
Group #:	Insurance Address	
Authorization Number: (attach copy of authorization letter)	Insurance City, State, Zip	Phone

Secondary

Insured/Policy Holder Name:	Policy Holder DOB:	Policy Holder Gender ☐ Male ☐ Female
Relationship to Patient ☐ Self ☐ Spouse ☐ Dependent ☐ Other:	Policy #	
Insurance Company Name:	Insurance ID #:	
Group #:	Insurance Address	
Authorization Number (attach copy of authorization letter)	Insurance City, State, Zip	Phone

I authorize Greenwood Genetic Center (GGC) Diagnostic Laboratories to furnish any medical information requested of me, or my covered dependents. In consideration of services rendered, I transfer and assign any benefits of insurance to GGC Diagnostic Laboratories. I understand I am responsible for any co-pay, deductibles, non-authorized, or non-covered services and remaining balances after insurance reimbursement. I understand I am fully responsible for payment of my account if the GGC Diagnostic Laboratories is not a participant with my health plan, or my health plan does not fully reimburse my medical services due to lack of authorization for medical necessity.

Printed Name:_____ Signature:_____ Date (MM/DD/YY):_____

Section 3: Self-pay

We accept check/Visa/MasterCard. All information required to process credit card payments.
Payments will be processed prior to initiation of testing.

Payment Method: ☐ Check ☐ Visa ☐ MasterCard	Credit Card Number:	
Amount: (with discount applied if applicable)	Exp. Date	CVV
Cardholder Name(print as it appears on the card):	Cardholder Signature:	Date
Billing address	City, State, Zip	Telephone

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Prenatal Exome Clinical Information

LAB USE ONLY

Last Name	First	MI	Maternal DOB	Numeric Identifier

Please provide the following clinical information regarding the proband being tested. This clinical information is crucial for an accurate interpretation of results. Check all that apply. If a feature is selected, please provide an additional description of the finding. Use blank space on right to provide other relevant details.

Growth

- ☐ Intrauterine Growth Restriction (IUGR)
- ☐ Microcephaly (OFC <3rd centile) _____
- ☐ Macrocephaly (OFC >97th centile) _____
- ☐ Other growth concerns: _____

Congenital Anomalies, please specify

- ☐ Heart malformations _____
- ☐ Kidney abnormalities _____
- ☐ Genital abnormalities _____
- ☐ Brain malformations _____
- ☐ Gastrointestinal anomalies _____
- ☐ Other _____

Craniofacial

- ☐ Cleft lip +/-cleft palate_____
- ☐ Dysmorphic facies _____
- ☐ Eye anomalies _____
- ☐ Ear anomalies _____

Skeletal & Limb Anomalies

- ☐ Limb anomalies _____
- ☐ Joint contractures _____
- ☐ Abnormal short/long bone measurements_____
- ☐ Other skeletal findings:_____

Other

- ☐ Polyhydramnios _____
- ☐ Oligohydramnios _____
- ☐ Non-immune hydrops _____
- ☐ Decreased fetal movement _____
- ☐ Other _____

Additional Information

Please draw pedigree below with any clinically relevant family history:

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Prenatal Exome Patient Consent Form

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Last Name First MI	Maternal DOB	**Numeric Identifier** (MR # or SS #)

The purpose of this document is to provide information about the *Prenatal Exome Sequencing (PES) Test.* Due to the complexity of this testing, genetic counseling is required prior to ordering the testing to discuss the possible outcomes and after the testing is completed to discuss the results.

ABOUT THE PRENATAL EXOME SEQUENCING TEST

The goal of this test is to identify the underlying genetic cause of birth defects and anomalies identified by ultrasound during pregnancy. Prenatal exome sequencing, abbreviated as PES, is a comprehensive and complex genetic test. A specimen of amniotic fluid or placental tissue [chorionic villus sampling (CVS)] will collected from the patient in order to test the baby's genetic material. Blood samples from the parent(s) will also be collected for analysis. DNA will be isolated from the specimens for genetic testing.

The whole ***genome*** is made up of DNA and includes the entire set of human genes (approximately 20,000) and other genetic material contained in the human chromosomes. The genes make up only a small fraction of the genome and are segments of DNA that serve as the "code" (i.e. the "recipe" or "blueprints") for the body by telling the cells of the body how to make proteins that have certain jobs. The segments of genes that help to make proteins are called ***exons,*** and the full collection of the exons from all of the genes is called the ***exome***. Exome sequencing is a single genetic test designed to analyze the important regions of many thousands of genes simultaneously. This test is different from other genetic tests in which only one gene or a selected group of genes is analyzed.

POTENTIAL OUTCOMES OF PES

- DIAGNOSTIC: There may be a change identified as the cause of the patient's concerns. These types of changes are mutations and would be the most helpful in understanding the underlying genetic diagnosis for the patient.
- NORMAL: There may not be any changes identified in the exome that are important to report. This does not mean that the patient does not have a genetic condition since PES is not able to detect all types of genetic mutations and does not analyze every part of every gene.
- PARENTAL BLOOD RELATIONSHIP OR MISTAKEN PARENTAGE: PES could reveal a potential blood relationship between the parents and could also detect mistaken parentage or non-paternity. These findings will typically not be reported unless it is necessary for understanding the patient's results.

OTHER IMPORTANT INFORMATION

- PES is not able to completely analyze the entire exome. Therefore, there may be mutations in genes that will not be identified by this test. PES is not able to detect all types of genetic mutations such as large missing or extra pieces of the chromosomes or expanded triplet repeats. This test may not detect changes in the mitochondrial DNA which is separate from the chromosomal DNA.
- There are certain changes that will not be reported during this analysis. The PES report **will not** include the following types of results:
    - BENIGN VARIANTS: Changes that are benign or do not cause disease will not be reported.
    - CARRIER STATUS: Carrier status for conditions not relevant to the clinical presentation and findings will not be reported. If a single variant is identified in a gene that could be important to the reason for testing, this will be reported.
    - VARIANT OF UNCERTAIN CLINICAL SIGNIFICANCE: Changes may be identified on exome sequencing that have uncertain implications for the patient. These changes may or may not be related to the patient's concerns. These changes will not be reported.
    - Changes that may cause a slight increased risk for common and easily diagnosable conditions such as diabetes and high blood pressure, or changes that can give information about drug metabolism (pharmacogenetics) will not be reported.
    - SECONDARY FINDINGS: There may be unanticipated changes identified in genes that are not related to the patient's current concerns but are medically important for the patient's health or the family's health. Secondary findings will not be included in the prenatal report.

Last Name First MI	Maternal DOB	**Numeric Identifier (MR # or SS #)**

OTHER IMPORTANT INFORMATION CONTINUED

- Genetic information often changes because information about all genes is not complete at this time. Therefore, variants in new genes will not be reported if the function of the gene is currently unknown.

REANALYSIS

Since our understanding of genes and variants changes constantly, it can be helpful to go back and review the patient's genetic data again when new information may be available. This is called a reanalysis. The reanalysis of the patient's genetic information is not done automatically. Usually the physician or genetic counselor will initiate the reanalysis by contacting the lab to request the patient's data be reviewed again and will provide the lab with any new medical information. A reanalysis of the data from the Prenatal Exome Sequencing can be requested at no additional charge after the baby is born. This reanalysis may include additional types of results that were not reportable during the initial prenatal analysis such as variants of unclear clinical significance (VUS) or secondary findings. A new consent form will be required to initiate this reanalysis. If any genetic changes are identified that are thought to be the cause of the concerns identified during pregnancy, this information will be stated in an updated report from the lab. A new, postnatal sample will be required for confirmation. The referring doctor or genetic counselor will contact the patient with this new information.

By signing below, I give consent to the Greenwood Genetic Center to conduct prenatal exome sequencing for my current pregnancy as recommended by my physician.

I also give consent to the Greenwood Genetic Center to conduct exome sequencing for myself in order to help interpret the results of prenatal exome sequencing that is being performed for my current pregnancy. I understand that a separate test report will not be issued for me, but that potentially significant genetic changes that are found in my DNA sample will be listed in the prenatal test report.

Signature: _____ Date:_____

Physician/Counselor Statement: I have provided genetic counseling to this individual regarding the clinical prenatal exome sequencing test. We have discussed the potential genetic findings, implications of the genetic test results, and limitations as outlined in this consent document. I have answered her questions about this testing.

Physician/Counselor Signature: _____ Date:_____

Prenatal Exome Sequencing
Paternal Request Form

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Last Name First MI			Address
Race ☐ B ☐ W ☐ Other:	Sex ☐ M ☐ F	DOB MM/DD/YYYY	City, State, Zip

Specimen Collection Date MM/DD/YYYY	Type of specimen	ICD10 Code	Numeric Identifier (Medical record # SS #)	Home telephone

Referring Physician: ****Printed name and Physician Signature Required****

Physician Name		Address	
Physician Signature		City, State, Zip	
Institution	NPI#	Telephone	Fax

Genetic Counselor/Care coordinator:

Name		Address
Telephone	Fax	City, State, Zip

NO CHARGE for paternal samples submitted as part of PES trio.

Indication for Study

Please provide the following information so this sample can be appropriately linked to the correct prenatal and maternal samples.
Mother's Name:_____ Mother's DOB:_____ Father of Pregnancy: ☐ Ye s ☐ No

☐ Unaffected

☐ Affected**;** Please list any relevant clinical features: _____

_____

☐ **Prenatal Exome Sequencing Paternal Sample**

☐ **PES Reanalysis** (postnatal analysis)

☐ **Follow-up Studies for Variant of Unknown Significance**
Specify gene(s) & variant(s):
_____

☐ **Targeted Analysis for Known Mutation**
Specify gene(s) & variant (s):
_____

Specimen Requirements

Paternal Sample: Blood, extracted DNA, or saliva/saliva swab is accepted for analysis.
Blood: 3-5 mL whole blood in an EDTA, lavender top tube
Extracted DNA: A minimum of 3 micrograms of extracted DNA is required.
Saliva/Saliva Swab: Saliva samples must be submitted in an approved saliva collection kit. Contact the lab to request a saliva kit.

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EDTA RT / R / F	Na Hep RT / R / F	Plasma RT / R / F	Urine / Flasks / Other RT / R / F	Serum / Tissue RT / R / F	DBS / DNA RT / R / F

5/18/20

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Paternal Consent

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By signing below, I give consent to the Greenwood Genetic Center to conduct exome sequencing for myself in order to help interpret the results of the prenatal exome sequencing that is being performed for my child. I understand that a separate test report will not be issued for me, but that potentially significant genetic changes that are found in my DNA sample will be listed in the prenatal report test report.

Signature:_____ Date:_____

Physician/Counselor Statement: I have provided genetic counseling to this individual regarding the clinical prenatal exome sequencing test. We have discussed the potential genetic findings, implications of the genetic test results and limitations as outlined in the patient's consent document. I have answered his questions about this testing.

Physician/Counselor Signature:_____ Date:_____