



Exome Request Form - Proband

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/Ethnicity			Sex <input type="checkbox"/> M <input type="checkbox"/> F		DOB MM/DD/YYYY		City, State, Zip
Specimen Collection Date MM/DD/YYYY		Type of specimen*		Numeric Identifier (Medical record # or SSN)		Home telephone	
*DNA samples only: Please identify where DNA extraction was performed. <input type="checkbox"/> CAP/CLIA Accredited Lab: _____ <input type="checkbox"/> Research Lab: _____ <input type="checkbox"/> Unknown							

Referring Physician:

Name		Address					
Institution		City, State, Zip					
NPI#		Telephone			Fax		
Email Address:		Preferred Method to Receive Results: <input type="checkbox"/> Secure Email <input type="checkbox"/> Fax <input type="checkbox"/> 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.**

<input type="checkbox"/> Institutional Billing: Complete section 1 on the separate BILLING FORM (page 2)
<input type="checkbox"/> Insurance: Complete section 2 on the BILLING FORM (page 2). No out-of-state (non-SC) insurance or Medicaid will be accepted.
<input type="checkbox"/> Self-pay: Complete section 3 on the separate BILLING FORM (page 2).

Indication for Study

<input type="checkbox"/> Symptomatic
<input type="checkbox"/> Family History (please attach a copy of the pedigree.)
Please complete the <u>required</u> Clinical Information Form (page 3) OR submit phenotype via Face2Gene.
<input type="checkbox"/> Clinical information submitted via Face2Gene. Case Number: _____

Parental & Family Member testing

Are family member samples being submitted along with proband sample? Yes No

Please note that a separate requisition form is required for each family member's sample submitted.

If yes, which sample(s) is/are being submitted?

Mother: _____ Father: _____ Other family members: _____

Whole Exome Sequencing (SINGLETON analysis)

Whole Exome Sequencing (DUO analysis)

Whole Exome Sequencing (TRIO analysis)

Specimen Requirements: 3-5 ml of peripheral blood collected in an EDTA (lavender top) Vacutainer tube. The specimen should be kept at room temperature and delivered via overnight shipping. Extracted DNA and saliva are also accepted.

LAB USE ONLY		Accessioned By:		Event Codes:		FedEx		Eagle		UPS		DHL		WC		Other:	
EDTA	Na Hep	Plasma / Serum	Urine	Flasks / Tissue	DBS / DNA	Saliva / Swab Buccal	PAX	ACD									
RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F

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)		DOB MM/DD/YYYY	City, State, Zip
ICD10 Code(s)			Telephone

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 <input type="checkbox"/> Male <input type="checkbox"/> Female
Relationship to Patient <input type="checkbox"/> Self <input type="checkbox"/> Spouse <input type="checkbox"/> Dependent <input type="checkbox"/> 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 <input type="checkbox"/> Male <input type="checkbox"/> Female
Relationship to Patient <input type="checkbox"/> Self <input type="checkbox"/> Spouse <input type="checkbox"/> Dependent <input type="checkbox"/> 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: <input type="checkbox"/> Check <input type="checkbox"/> Visa <input type="checkbox"/> MasterCard <input type="checkbox"/> AmEx <input type="checkbox"/> Discover	Credit Card Number:
Amount: (with discount applied if applicable)	Exp. Date
Cardholder Name(print as it appears on the card):	CVV
Billing address	Cardholder Signature:
	Date
	City, State, Zip
	Telephone



Exome Clinical Information Form

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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.

<p>Growth</p> <p><input type="checkbox"/> Failure to thrive _____</p> <p><input type="checkbox"/> Microcephaly (OFC <3rd centile) _____</p> <p><input type="checkbox"/> Macrocephaly (OFC >97th centile) _____</p> <p><input type="checkbox"/> Short stature (Ht <3rd centile) _____</p> <p><input type="checkbox"/> Tall stature (Ht >97th centile) _____</p> <p><input type="checkbox"/> Obesity/Overgrowth _____</p> <p>Neurological/Muscular</p> <p><input type="checkbox"/> Spasticity/Hyperreflexia _____</p> <p><input type="checkbox"/> Ataxia _____</p> <p><input type="checkbox"/> Tremors _____</p> <p><input type="checkbox"/> Hypotonia _____</p> <p><input type="checkbox"/> Seizures _____</p> <p><input type="checkbox"/> Abnormal movements _____</p> <p>Development, Physical & Cognitive</p> <p><input type="checkbox"/> Delayed motor milestones _____</p> <p><input type="checkbox"/> Intellectual disability _____</p> <p><input type="checkbox"/> Speech/Language delay _____</p> <p><input type="checkbox"/> Developmental regression _____</p> <p>Craniofacial, Ophthalmologic, Auditory</p> <p><input type="checkbox"/> Vision Loss _____</p> <p><input type="checkbox"/> Hearing loss/Deafness _____</p> <p><input type="checkbox"/> Dysmorphic facies _____</p> <p><input type="checkbox"/> Eye anomalies _____</p> <p><input type="checkbox"/> Ear anomalies _____</p> <p>Skeletal & Limb Anomalies</p> <p><input type="checkbox"/> Limb malformation _____</p> <p><input type="checkbox"/> Joint contractures _____</p> <p><input type="checkbox"/> Craniosynostosis _____</p> <p><input type="checkbox"/> Hyperextensibility _____</p> <p>Congenital Anomalies</p> <p><input type="checkbox"/> Heart malformations _____</p> <p><input type="checkbox"/> Kidney abnormalities _____</p> <p><input type="checkbox"/> Genital abnormality _____</p> <p><input type="checkbox"/> Brain malformations _____</p> <p><input type="checkbox"/> Gastrointestinal anomalies _____</p> <p><input type="checkbox"/> Other _____</p> <p>Other Features</p> <p><input type="checkbox"/> Prematurity _____</p> <p><input type="checkbox"/> Intrauterine growth restriction _____</p> <p><input type="checkbox"/> Autism/Autism Spectrum Disorders _____</p> <p><input type="checkbox"/> Metabolic abnormalities _____</p> <p><input type="checkbox"/> Mitochondrial abnormalities _____</p> <p><input type="checkbox"/> Pigmentary abnormalities _____</p> <p><input type="checkbox"/> Other skin findings _____</p> <p><input type="checkbox"/> Organomegaly _____</p> <p><input type="checkbox"/> Cancer/tumor formation _____</p>	<p style="text-align: center;">Additional Information</p>
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Exome Patient Consent Form

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The purpose of this document is to provide information about the *Whole Exome Sequencing (WES) 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 WHOLE EXOME SEQUENCING TEST

The goal of this test is to identify the underlying genetic cause of intellectual or other developmental disabilities, birth defects, or unexplained medical concerns. Whole exome sequencing, abbreviated as WES, is a comprehensive and complex genetic test. A sample of blood will be collected from the patient, and when necessary, from his/her parents or other appropriate family members. DNA will be isolated from the blood 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 whole **exome**. Whole 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 WES

- **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.
- **VARIANT OF UNCERTAIN CLINICAL SIGNIFICANCE** – there may be a change identified that we cannot be certain about what it means for the patient. It may or may not be related to the patient’s concerns.
- **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 WES is not able to detect all types of genetic mutations and does not analyze every part of every gene.
- **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.
- **PARENTAL BLOOD RELATIONSHIP OR MISTAKEN PARENTAGE** - WES could reveal a potential blood relationship between the parents and could also detect mistaken parentage. These findings will typically not be reported unless it is necessary for understanding the patient’s results.

OTHER IMPORTANT INFORMATION

- WES is not able to completely analyze the entire exome. Therefore, there may be mutations in genes that will not be identified by this test. WES 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.

Last Name	First	MI	DOB	Numeric Identifier (MRN or SSN)
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- There are certain changes that will not be reported including changes that are benign or do not cause disease, even if the change could be important for future reproductive decisions (carrier status). 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 or incidental findings in genes that cause certain adult onset conditions that cannot be prevented (such as Alzheimer disease or Parkinson disease) will not be reported.
- Genetic information often changes because information about all genes is not complete at this time. Therefore, although a variant may be found by doing WES, it may not be reported if the function of the gene is currently unknown.

SECONDARY FINDINGS REPORTING

Secondary or incidental findings are changes in genes that are not related or not thought to be related to the patient’s concerns. The purpose of WES is not to detect incidental findings but rather to identify the cause of the patient’s current condition. However, the American College of Medical Genetics and Genomics (ACMG) recommends that all laboratories performing WES report mutations identified in a specific set of genes. These genes were chosen because they are considered medically important for the health of the patient or the patient’s family members. Our laboratory may also feel compelled to report secondary findings in additional genes not included in the set of genes recommended by the ACMG. Secondary findings will only be reported for the patient, and only mutations or variants that are expected to harm the function of the gene will be reported. It is important to remember that a normal result for these genes does not mean that there is no mutation present, since not all of these genes are being completely analyzed simply due to the nature of WES. In addition, a normal result for the patient does not mean that the parents, or other family members tested, would also have a normal result. The patient and/or parent has the option to receive or *not* to receive information about the patient’s changes that are considered secondary findings by initialing below.

_____ Yes I **do** choose to have secondary findings included in the results report

_____ No I **do not** choose to have secondary findings included in the results report

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 and typically can only be requested at least one year after the initial whole exome sequencing analysis has been reported. 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. Sometimes when we learn about a new gene, the lab will review patients’ genetic information for mutations in that specific gene in case it could be related to their health concerns. If any genetic changes are identified that are thought to be the cause of the patient’s concerns, this information will be stated in an updated report from the lab. The 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 whole exome sequencing for my child or myself as recommended by my physician.

Signature: _____ Date: _____

Printed Name: _____ Relationship to Patient: _____

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

Physician/Counselor Signature: _____ Date: _____



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Patient Information (Please Print):

Last Name		First	MI	Address		
Race/Ethnicity		Sex <input type="checkbox"/> M <input type="checkbox"/> 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:

Name		Address				
Institution		City, State, Zip				
NPI#		Telephone			Fax	
Email Address:		Preferred Method to Receive Results: <input type="checkbox"/> Secure Email <input type="checkbox"/> Fax <input type="checkbox"/> 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		

Indication for Study

Proband Name: _____ Proband DOB: _____

Relationship of family member to the proband: _____

Unaffected – *Clinic Information Form* is not needed

Affected – **Please complete the required *Clinical Information Form* (page 7)**

- Whole Exome Sequencing (DUO analysis)**
- Whole Exome Sequencing (TRIO analysis)**
- Whole Exome Sequencing (OTHER analysis)**
- Targeted Analysis for Known Mutation** (submit first page only – no consent needed)

Specify gene(s) & variant(s): _____

Specimen Requirements: 3-5 ml of peripheral blood collected in an EDTA (lavender top) Vacutainer tube. The specimen should be kept at room temperature and delivered via overnight shipping. Extracted DNA and saliva are also accepted.

<p>Ordering Checklist:</p> <ul style="list-style-type: none"> <input type="checkbox"/> Test Requisition Form <input type="checkbox"/> Informed consent for proband <input type="checkbox"/> Copy of Pedigree <input type="checkbox"/> Clinical information page 	<p><u>Family Member Information</u></p> <ul style="list-style-type: none"> <input type="checkbox"/> Maternal Sample & Requisition <input type="checkbox"/> Paternal Sample & Requisition <input type="checkbox"/> Other Family Member: _____ <input type="checkbox"/> Family Member Consents
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EDTA	Na Hep	Plasma / Serum	Urine	Flasks / Tissue	DBS / DNA	Saliva / Swab Buccal	PAX	ACD	
RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	RT / R / F	

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Please provide the following clinical information regarding the individual 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.

<p>Growth</p> <p><input type="checkbox"/> Failure to thrive _____</p> <p><input type="checkbox"/> Microcephaly (OFC <3rd centile) _____</p> <p><input type="checkbox"/> Macrocephaly (OFC >97th centile) _____</p> <p><input type="checkbox"/> Short stature (Ht <3rd centile) _____</p> <p><input type="checkbox"/> Tall stature (Ht >97th centile) _____</p> <p><input type="checkbox"/> Obesity/Overgrowth _____</p> <p>Neurological/Muscular</p> <p><input type="checkbox"/> Spasticity/Hyperreflexia _____</p> <p><input type="checkbox"/> Ataxia _____</p> <p><input type="checkbox"/> Tremors _____</p> <p><input type="checkbox"/> Hypotonia _____</p> <p><input type="checkbox"/> Seizures _____</p> <p><input type="checkbox"/> Abnormal movements _____</p> <p>Development, Physical & Cognitive</p> <p><input type="checkbox"/> Delayed motor milestones _____</p> <p><input type="checkbox"/> Intellectual disability _____</p> <p><input type="checkbox"/> Speech/Language delay _____</p> <p><input type="checkbox"/> Developmental regression _____</p> <p>Craniofacial, Ophthalmologic, Auditory</p> <p><input type="checkbox"/> Vision Loss _____</p> <p><input type="checkbox"/> Hearing loss/Deafness _____</p> <p><input type="checkbox"/> Dysmorphic facies _____</p> <p><input type="checkbox"/> Eye anomalies _____</p> <p><input type="checkbox"/> Ear anomalies _____</p> <p>Skeletal & Limb Anomalies</p> <p><input type="checkbox"/> Limb malformation _____</p> <p><input type="checkbox"/> Joint contractures _____</p> <p><input type="checkbox"/> Craniosynostosis _____</p> <p><input type="checkbox"/> Hyperextensibility _____</p> <p>Congenital Anomalies</p> <p><input type="checkbox"/> Heart malformations _____</p> <p><input type="checkbox"/> Kidney abnormalities _____</p> <p><input type="checkbox"/> Genital abnormality _____</p> <p><input type="checkbox"/> Brain malformations _____</p> <p><input type="checkbox"/> Gastrointestinal anomalies _____</p> <p><input type="checkbox"/> Other _____</p> <p>Other Features</p> <p><input type="checkbox"/> Prematurity _____</p> <p><input type="checkbox"/> Intrauterine growth restriction _____</p> <p><input type="checkbox"/> Autism/Autism Spectrum Disorders _____</p> <p><input type="checkbox"/> Metabolic abnormalities _____</p> <p><input type="checkbox"/> Mitochondrial abnormalities _____</p> <p><input type="checkbox"/> Pigmentary abnormalities _____</p> <p><input type="checkbox"/> Other skin findings _____</p> <p><input type="checkbox"/> Organomegaly _____</p> <p><input type="checkbox"/> Cancer/tumor formation _____</p>	<p>Additional Information</p>
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Exome Family Member Consent Form

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Last Name	First	MI	DOB	Numeric Identifier (MRN or SSN)
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ABOUT THE WHOLE EXOME SEQUENCING TEST

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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 whole **exome**. Whole 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 at the same time. Please refer to the Whole Exome Sequencing Patient Consent Form or the Patient Information Brochure for more information.

By signing below, I give consent to the Greenwood Genetic Center to conduct whole exome sequencing for myself in order to help interpret the results of whole exome sequencing that is being performed for my child/other family member (i.e. the patient). 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 my child's/other family member's (i.e. the patient's) test report. I understand that I will not receive information regarding secondary findings for myself.

Signature: _____ Date: _____

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

Physician/Counselor Signature: _____ Date: _____