



LRS 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	

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Name		Address			
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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.)
<input type="checkbox"/> Please complete the <u>required</u> <i>Clinical Information Form</i> (page 3)

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: _____

Long-Read Sequencing Singleton (proband only) Long-Read Sequencing (DUO analysis)

Long-Read Sequencing (TRIO analysis)

Ordering Checklist: Test requisition form, informed consent for proband, copy of pedigree, clinical information page, family member requisition and consent for family member samples being submitted.

Specimen Requirements: The accepted sample type is 3-4 ml of fresh or frozen blood collected in an EDTA (purple top) tube. Ultra-high molecular weight DNA is required for long-read sequence analysis and can only be extracted if specimen and transport requirements are met. Fresh blood can be shipped at ambient temperature. Frozen blood should be shipped on dry ice.

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	

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	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 <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	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

Last Name	First	MI	DOB	Numeric Identifier (MRN or SSN)
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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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LRS Patient Consent Form

106 Gregor Mendel Circle • Greenwood, SC 29646
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Website: www.ggc.org

LAB USE ONLY

Last Name	First	MI	DOB	Numeric Identifier (MRN or SSN)
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The purpose of this document is to provide information about the *Long Read Sequencing 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 LONG-READ 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. Long-Read Sequencing, (abbreviated LRS) 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.

Long-read sequencing is a specific type of genetic test used to analyze a patient’s whole genome. 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 the genes is called the whole *exome*. The exome is only part of the genome. The exome is the part of the genome that we have the most information and understand the best.

Long-read sequencing utilizes larger pieces of DNA than other methods. Using longer strands of DNA can make it easier to analyze the data and allow the lab to detect more types of genetic changes. LRS can detect both small changes such as spelling mistakes in the DNA as well as larger changes such as missing or extra pieces of genetic information, called deletions or duplications. While this test can analyze most of the genome, there are some regions of the genome that we are still learning about.

POTENTIAL OUTCOMES OF LRS

- **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 with this test that are important to report. This does not mean that the patient does not have a genetic condition since LRS 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** – LRS 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

- This test cannot detect all types of genetic mutations and there may be genetic changes not identified by this test.
- 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) and are not related to the patient’s current medical conditions 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 this test, it may not be reported if the function of the gene is currently unknown.

Last Name	First	MI	DOB	Numeric Identifier (MRN or SSN)
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MITOCHONDRIAL VARIANT REPORTING

LRS analysis also captures sequence data from the mitochondrial genome. You may choose to include this information as part of the analysis. Please note that only variants currently known to be associated with mitochondrial disorders will be reported, and copy number status of the mitochondrial genome is not included.

_____ Yes I **do** choose to have mitochondrial variant analysis included in the results report
 _____ No I **do not** choose to have mitochondrial variant analysis included in the results report

TRIPLET REPEAT EXPANSION REPORTING

LRS analysis can detect conditions caused by triplet repeat expansions, such as Fragile X syndrome or myotonic dystrophy. You may choose to include this information as part of the analysis.

_____ Yes I **do** choose to have repeat expansion analysis included in the results report
 _____ No I **do not** choose to have repeat expansion analysis included in the results report

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 the LRS 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 exome or genome sequencing 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 LRS. 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, variants, and all genetic information 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 this test has been reported. Usually the healthcare provider 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. One of the genetics healthcare providers will contact the patient with this new information.

By signing below, I give consent to the Greenwood Genetic Center to conduct long-read 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 long-read 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: _____



LRS – Family Member

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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: _____

Affected – Specify: _____

Unaffected

Long-Read Sequencing Family Member

Targeted Analysis for Known Mutation (submit first page only – no consent needed)

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

Ordering Checklist:

- Test Requisition Form
- Informed consent for proband
- Copy of Pedigree
- Clinical information page

Family Member Information

- Maternal Sample & Requisition
- Paternal Sample & Requisition
- Other Family Member: _____
- Family Member Consents

Specimen Requirements The accepted sample type is 3-4 ml of fresh or frozen blood collected in an EDTA (purple top) tube. Ultra-high molecular weight DNA is required for long-read sequence analysis and can only be extracted if specimen and transport requirements are met. Fresh blood can be shipped at ambient temperature. Frozen blood should be shipped on dry ice.

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



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By signing below, I give consent to the Greenwood Genetic Center to conduct long-read sequencing for myself in order to help interpret the results of long-read 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: _____

Healthcare Provider Statement: I have provided genetic counseling to this individual/this individual’s family regarding the clinical long-read 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: _____



LRS – Family Member

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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	

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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



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Signature: _____ Date: _____

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Physician/Counselor Signature: _____ Date: _____