

Exome Request Form - Proband

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

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Patient Information (Please Print): Address Race/Ethnicity DOB MM/DD/YYYY City, State, Zip Пм Specimen Collection Date MM/DD/YYY Numeric Identifier (Medical record # or SSN) Type of specimen Home telephone Referring Physician: Address Name Institution City, State, Zip NPI# Telephone Email Address: Preferred Method to Receive Results: ☐ Fax ☐ Regular Mail ☐ Secure Email ☐ Care Coordinator Additional report to: Genetic Counselor ☐ Institution Other: Address Telephone City, State, Zip Additional report to: Genetic Counselor Institution ☐ Care Coordinator Other: 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. ☐ <u>Institutional Billing</u>: Complete section 1 on the separate <u>BILLING FORM</u> (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 Symptomatic** Family History (please attach a copy of the pedigree.) Please complete the required Clinical Information Form (page 3) OR submit phenotype via Face2Gene. 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? Father: Other family members: Mother: Whole Exome Sequencing (proband only) WES Reanalysis (submit second page with Whole Exome Sequencing (TRIO analysis) clinical information if phenotype has changed) 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 unless otherwise indicated. Ordering Checklist: Family Member Information Maternal Sample & Requisition **Test Requisition Form** Informed consent for proband Paternal Sample & Requisition Other Family Member: Copy of Pedigree Clinical information page Family Member Consents LAB USE ONLY Accessioned By: Eagle UPS DHL WC Other: **Event Codes:** FedEx DBS / DNA **EDTA** Na Hep Plasma / Urine Flasks / Saliva / Swab PAX ACD Serum Tissue Buccal RT / R / F RT / R / F



Greenwood Genetic Center Diagnostic Laboratory Billing Form This page is required to process any test requests.

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Out of State (non-SC) commercial insurance can only be filed for NGS Panels.

No out of state Medicaid will be accepted for the following items are needed in order to bil information. This form must be completed witted the completed witted the completed of the Alegible copy of both sides of the Authorization number, authorization.	II the pation th ALL rec ne insuran	ent's insurand quested infori nce card	mation.		claim if we are missing	
Patient Information:						
Last Name First	M	11	Address			
Numeric Identifier (Medical record # or SSN)	/DD/YYYY	City, State, Zip		Telephone		
ICD10 Code(s)						
Section 1: Institutional Billing Complete section below with institution information. *I submitting the order.* Please contact the GGC Billing (Institution/Organization	Office at 8					
				Liliali.		
Billing Address		City, State, Zip				
Account Number:		Telephone		Fax		
Section 2: Insurance Information						
			RANCE CARD (FRONT &	BACK)		
Primary	ormation	required to ii	le insurance claims.			
Insured/Policy Holder Name:		Policy Holde	er DOB:	Policy H	lolder Gender ☐ Male ☐ Female	
Relationship to Patient		Policy#			□ Iviale □ I elliale	
☐ Self ☐ Spouse ☐ Dependent ☐ Other: Insurance Company Name:		Insurance II	D #:			
Group #:		Insurance A	ddress			
Authorization Number: (attach copy of authorization letter)		Insurance City, State, Zip		Phon	Phone	
Secondary						
Insured/Policy Holder Name:		Policy Holde	er DOB:	Policy H	older Gender ☐ Male ☐ Female	
Relationship to Patient Self Spouse Dependent Other:		Policy#		•		
Insurance Company Name:		Insurance II	D #:			
Group #:		Insurance Address				
Authorization Number (attach copy of authorization letter)		Insurance C	Insurance City, State, Zip		e	
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: Date (MM/DD/YY):						
Section 3: Self-pay						
We accept check/Visa/Maste Payments			required to process credi or to initiation of testing.	t card payments	S.	
Payment Method:	sterCard	Credit Card N				
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Cardholder Name(print as it appears on the card):		Cardholder Si	gnature:		Date	

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Payment Method:			Credit Card Number:			
☐ Check	☐ Visa	☐ MasterCard				
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Cardholder Name(print as it appears on the card):		Cardholder Signature:		Date		
Billing address			City, State, Zip	Telep	hone	



Last Name

Other skin findings _ Organomegaly _____

Cancer/tumor formation____

Exome Clinical Information Form

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DOB

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1	ΔR	LISE	UNI	V

Numeric Identifier (MRN or SSN)

Plea	ise provide the following clinical information regarding the proband being tested. This clinical	al information is crucial for an accurate
	rpretation of results. Check all that apply. If a feature is selected, please provide an additional	
	ce on right to provide other relevant details.	in description of the infamily, ose stank
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	Macrocephaly (OFC >97 th centile)	
\Box	Short stature (Ht <3 rd centile)	
_	Tall stature (Ht >97 th centile)	
\Box	Obesity/Overgrowth	
	rological/Muscular	
	Spasticity/Hyperreflexia	
	Ataxia	
	Tremors	
	Hypotonia	
	Seizures	
	Abnormal movements	
Dev	elopment, Physical & Cognitive	
	Delayed motor milestones	
	Intellectual disability	
	Speech/Language delay	
	Developmental regression	
Cran	iofacial, Opthalmalogic, Auditory	
	Vision Loss	
	Hearing loss/Deafness	
	Dysmorphic facies	
	Eye anomalies	
	Ear anomalies	
Skel	etal & Limb Anomalies	
	Limb malformation	
	Joint contractures	
	Craniosynostosis	
Ш	Hyperextensibility	
Con	genital Anomalies	
닏	Heart malformations	
닏	Kidney abnormalities	
	Genital abnormality	
	Brain malformations	
	Gastrointestinal anomalies	
	Other	
_	er Features	
	Prematurity	
	Intrauterine growth restriction	
	Autism/Autism Spectrum Disorders	
	Metabolic abnormalities	
	Mitochondrial abnormalities	



Exome Patient Consent Form

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

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.

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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 <i>do not</i> choose to have secondary findings included in the results report

REANALYSIS

Physician/Counselor Signature:

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 <u>not</u> 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 Green my child or myself as recommended by my phy	wood Genetic Center to conduct whole exome sequencing for ysician.
Signature:	Date:
Printed Name:	Relationship to Patient:
regarding the clinical whole exome sequencing	ed genetic counseling to this individual/this individual's family ng test. We have discussed the potential genetic findings, g secondary findings, and limitations as outlined in this consent tions about this testing.

Date:



EDTA

RT / R / F

Na Hep

RT / R / F

Plasma /

RT / R / F

Serum

Urine

RT / R / F

Flasks /

Tissue

RT / R / F

DBS / DNA

RT / R / F

Saliva / Swab

RT / R / F

Buccal

PAX

Exome Request Form – Family Member

Greenwood
Genetic Center

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Patient Information (Please Prin	+ \•				2.0000
Last Name First	i. L	MI	Addr	ess	
Race/Ethnicity	Sex	DOB MM/DD/YYY	Y City,	State, Zip	
Specimen Collection Date MM/DD/YYYY	Type of specimen	ICD10 Code	Numeric Id	entifier (Medical record # SS #)	Home telephone
Referring Physician:					
Name			Address		
Institution			City, State	, Zip	
NPI#			Telephone		Fax
Email Address:				Method to Receive Results:	Fax ☐ Regular Mail
Additional report to: Genetic Co	ounselor 🗌 Instit	ution Care	Coordinat		Tax Tregular Mail
Name			Address		
Telephone Fax	Email:			City, State, Zip	
Additional report to: Genetic Co	ounselor 🗌 Instit	ution 🗌 Care	Coordinat	or	
Name			Address		
Telephone Fax	Email:			City, State, Zip	
Billing: Select how the test(s) will be	e billed & complete	the billing inforn	nation on t	he next page. The BILLING	FORM on page 2 is required.
☐ Institutional Billing: Complete se					роде за тако
☐ Insurance: Complete section 2 o	n the <u>BILLING FOR</u>	(page 2). No o	ut-of-state	(non-SC) insurance or Medi	caid will be accepted.
☐ <u>Self-pay</u> : Complete section 3 on	the separate BILLI	NG FORM (page 2	2).		
NO C	HARGE for fam	ily members	submitte	d as part of WES duo	or trio.
Indication for Study		_		-	
Proband Name:		Prob	and DOB	·	
Relationship of family member to	the proband:				
Unaffected – Clinic Inform	mation Form is no	t needed			
			··	(man = 7)	
Affected – Please comple	ete the <u>required</u> (Jinicai informat	tion Form	(page /)	
Whole Exome Sequencing	Family Membe	er			
WES Reanalysis (submit se	cond page with cl	inical informatio	n if phen	otype has changed for affe	ected family members)
Follow-up Studies for Var	iant of Unknow	n Significance			
Specify gene(s) & variant	(s):				
☐ Targeted Analysis for Kno	wn Mutation (s	ubmit first pag	e only –	no consent needed)	
Specify gene(s) & variant	(s):				
Ordering Checklist:				Family Member Informatio	nn
☐ Test Requisition Form				☐ Maternal Sample	
☐ Informed consent for pro	band			Paternal Sample	
☐ Copy of Pedigree				☐ Other Family Me	
☐ Clinical information page				☐ Family Member (Consents
LAB USE ONLY Accessioned By:	Event	Codes:	FedE	x Eagle UPS DHL W	C Other:

ACD



First

Last Name

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MI

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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. Failure to thrive						
Space on right to provide other relevant details. Additional Information	Plea	Please provide the following clinical information regarding the individual being tested. This clinical information is crucial for an accurate				
Failure to thrive	inte	pretation of results. Check all that apply. If a feature is selected, please provide an additional descr	ription of the finding. Use blank			
Failure to thrive Microcephaly (OFC <3" centile) Macrocephaly (OFC <3" centile) Short stature (IH <3" centile) Obesity/Overgrowth Obesity/Overgr	spac	e on right to provide other relevant details.				
Microcephaly (OFC -93th centile)	Grov	vth	Additional Information			
Macrocephaly (OFC > 97th centile)	_					
Macrocephaly (OFC > 97th centile)		Microcephaly (OFC <3 rd centile)				
Short stature (Ht <3 rd centile) Tall stature (Ht <3 rd centile) Obesity/Overgrowth Neurological/Muscular Spasticity/Hyperreflexia		Macrocephaly (OFC >97 th centile)				
Tall stature (Ht > 57th Centile) Obesity/Overgrowth Neurological/Muscular		Short stature (Ht <3 rd centile)				
Obesity/Overgrowth Neurological/Muscular Spasticity/Hyperreflexia		Tall stature (Ht >97 th centile)				
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Autism/Autism Spectrum Disorders		Intrauterine growth restriction				
☐ Metabolic abnormalities		Autism/Autism Spectrum Disorders				
☐ Mitochondrial abnormalities	l	Metabolic abnormalities				
☐ Pigmentary abnormalities		Mitochondrial abnormalities				
□ Other skin findings □ Organomegaly						
☐ Organomegaly		Other skin findings				
		Organomegaly				
		Cancer/tumor formation				



Exome Family Member Consent Form

106 Gregor Mendel Circle • Greenwood, SC 29646 Toll Free: (800) 473-9411 • Fax: (864) 941-8141

Website: www.ggc.org

LAB USE	ONLY
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Last Name	First	MI	DOB	Numeric Identifier (MRN or SSN)

ABOUT THE WHOLE EXOME SEQUENCING TEST

Signature:

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 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.

Date:

Physician/Counselor Statement: I have provided a	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 fir	ndings, and limitations as outlined in the patient's consent
document. I have answered his/her/their questions a	bout this testing.
Physician/Counselor Signature:	Date: