

STOP! DO NOT USE THE CONTENTS OF THIS PACKET IF THE SAMPLE IS FOR CHROMOSOME ANALYSIS ONLY. All that is required is a physician's order.

Fetal Examination/Genetic Evaluation Information and Instructions

Contents: Fetal Examination Protocol
Explanatory letter to family
Authorization for Fetal Examination
Authorization to Obtain Medical Records
Authorization to Release Medical Records
Courier instructions
Lab Request Forms for Parental DNA Isolation

Checklist: **_____ Referral call to the Greenwood Genetic Center to discuss indication for genetic evaluation. During regular business hours call the Fetal Examination office at 864-388-1700 or after business hours contact the Clinical Geneticist on call through GGC's call service at (1-866-744-3934). This call should be made by referring physician or nurse. A fetus received without notification, and without indication for examination at the GGC, will be transported back to the referring hospital (at referring hospital's expense).**

- _____ Explanatory letter given to family
- _____ **White copy of signed Authorization for Fetal Examination***
- _____ **White copy of signed Authorization to Obtain Medical Records***
- _____ **Authorization to Release Medical Records**
- _____ Specimen with identification
- _____ (If possible) Wedge of placenta including amnion with identification
- _____ Purple top tube (for DNA isolation) from each parent
- _____ Prenatal and delivery records

***PLEASE NOTE: The Fetal Examination will not be completed without these signed authorizations.**

Protocol for Fetal Examination for Birth Defects or Suspected Genetic Disorder

The Greenwood Genetic Center offers Fetal Examination and Genetic Consultation in cases of known or suspected birth defects, genetic disorders and potential teratogenic exposures.

IMPORTANT NOTICE

The Greenwood Genetic Center cannot provide a complete autopsy including histologic examination. Please call the Greenwood Genetic Center Fetal Examination office at 864-388-1700 during regular business hours or the Clinical Geneticist on call after business hours through GGC's call service at (1-866-744-3934) prior to transport to discuss the indications for examination and genetic evaluation.

Cases in which it is expected that histologic examination of tissues will be necessary (e.g. suspected infection, intrauterine fetal demise of unknown cause) should be autopsied at the referring hospital. We are available to consult with the pathologist if questions arise. Tissue samples (i.e. skin, umbilical cord, gonad) can be submitted to the Greenwood Genetic Center Cytogenetic Laboratory for chromosome analysis/karyotype but must be obtained prior to formalin fixation.

If a complete placental examination is indicated, it should be performed at the referring hospital (not possible at Greenwood Genetic Center). A full thickness wedge of placental tissue (including amnion) should be sent to Greenwood Genetic Center in saline (not formalin). This is particularly important with a macerated fetal specimen, to optimize the chance of successful cell culture and karyotype.

If the fetal specimen is appropriate for examination at the Greenwood Genetic Center, the following protocol should be followed:

I. If not previously discussed with mother/family, discuss fetal examination and give explanatory letter included in this packet. Note that although either parent can consent to fetal examination and disposition of the specimen, **only the mother can sign for the release of her medical records.**

II. Obtain signed Authorization for Fetal Examination, included in this packet, and include Instruction for Disposal of Body. **Keep yellow copy** with original record. **Send white copy** with the specimen. Obtain both signed Authorization to Release Medical Records and Authorization to Obtain Medical Records, and send with specimen. If family has not decided disposition or not yet chosen a funeral home, write accordingly and contact the Greenwood Genetic Center as soon as possible when a decision is made.

III. Obtain cord blood (when possible) if the fetus is born alive; it is desirable that a 5 ml green (sodium heparin) tube be submitted to Greenwood Genetic Center for chromosome analysis. **If the delivering physician requests other studies** (CBC, hemoglobin electrophoresis, serology, TORCH titers, IgM, studies for suspected sepsis chorioamnionitis) these must be obtained and processed at the referring institution.

**Greenwood Genetic Center
Fetal Examination Protocol
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IV. Measure fetal length, weight and head circumference, and record. **Include this information with specimen.**

V. **Fetal specimen should be wrapped in towels lightly moistened with STERILE SALINE and placed in a sealed plastic bag. Do not “float” the specimen in saline.** Place identifying information on inside and outside of bag. Include mother’s full name, date of mother’s birth, date and time of delivery, and referring physician’s name (Resident and Attending physician). **Keep specimen refrigerated. DO NOT PLACE IN FORMALIN. Do not freeze.**

- A. Place cold pack/s in the bottom of a leak proof container**
- B. Place towels or gauze on top of the cold packs to form a barrier**
- C. Place the sealed plastic bag containing the specimen on top of the barrier. Do not allow the specimen to come in contact with or be in close contact with the cold packs. The specimen must not freeze**
- D. Place copies of prenatal and delivery records on top of the sealed plastic bag or secured to the outside of the leak proof container**
- E. If specimen meets DHEC specifications, attach Burial/Removal/Transit Permit form (BRT) in a visible location on the leak proof container**

VI. **Purple top tube (for DNA isolation) on both parents.**

VII. Sending the specimen:

- A. During regular business hours (Monday-Friday 8:00 a.m.-5:00 p.m.) call:
(864) 388-1700 Anatomic Studies Office or
1-888-GGC-GENE Laboratory (Toll Free)
Send specimen to: Greenwood Genetic Center – Anatomic Studies Laboratory
 106 Gregor Mendel Circle
 Greenwood, SC 29646
- B. Evenings, weekends, and holidays call
(866) 744-3934 Ask for the Clinical Geneticist on call
Send specimen to: Self Regional Healthcare Emergency Department
 1325 Spring Street
 Greenwood, SC 29646

VII. A courier service does pick up specimens in a variety of locations. A list of courier contacts is included in this packet. Call the Laboratory for details or questions.

Dear Parent:

Please accept our expression of sympathy; we wish that our introduction to you could have been at a more pleasant event than this one. Your physician has requested that examination and genetic studies be performed on your fetus/infant to better understand the suspected or actual abnormalities that led to the unexpected and unhappy conclusion of your pregnancy.

The genetic evaluation will include a thorough physical examination, including external measurements, study of the internal organs, and X-rays to look at the bone structures. It also often includes laboratory tests, including analysis of chromosomes and other studies. Note that we request a blood sample on both parents as this may be necessary to properly interpret lab results on the infant.

A complete study also requires that we review medical records (prenatal, delivery, previous pregnancies). We hope that this effort will be helpful to you and your physician, to explain the recent events and to help in the planning and management of future pregnancies in your family.

The examination will be performed by staff members of the Greenwood Genetic Center (GGC). There is no charge for the examination, but your insurance will be charged for any laboratory services. We will submit a brief preliminary report to your physician within a few weeks, and a final report in about eight weeks. You will receive a letter advising you that each has been submitted. We encourage you to discuss the results with your physician. Physicians and genetic counselors at the GGC will also be available for consultation and counseling if this is desired.

If you wish to have funeral or memorial services for your baby, the autopsy is usually completed within 4 days, but it is sometimes longer. If you prefer, you may donate the body to the GGC. Final disposition will then be at our discretion. Usually, the remains are buried. A memorial marker is placed at the burial site in honor of all the families we have served. If you would like further information about these options, please call us at (864)388-1700.

Sincerely wishing you well at this time of grief and loss,

Sincerely,



Roger E. Stevenson, M.D.
Fetal Examination Service

**GREENWOOD GENETIC CENTER
FETAL EXAMINATION SERVICE**

AUTHORIZATION FOR FETAL EXAMINATION

Addressograph

I hereby give the Greenwood Genetic Center and their staff permission to perform an examination of the body of

Name	Date of Birth	Date of Death
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This examination includes examination, removal and retention of such organs and parts of such organs and tissues as may be deemed proper by the examining physician, photography and x-rays for documentation of abnormalities, in the interest of determining the cause of the anomalies and advancing medical knowledge regarding the cause of birth defects and genetic disorders.

Printed name of Mother of deceased fetus/infant	Mother's DOB
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Signature of parent/legal guardian/next of kin	Relationship to deceased	Date
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Signature of Witness	Printed name of Witness	Date
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Permit for DONATION of Body and Tissues

A body may be donated (via the South Carolina State Anatomical Gift Act) for scientific study. Final arrangements regarding the remains of the deceased will be made by the Greenwood Genetic Center. Such donations may be made by completing the following authorization.

I hereby authorize the donation of the body of _____, deceased, to the Greenwood Genetic Center. I understand that the disposition of the remains will be at the discretion of the Greenwood Genetic Center and its agents at no monetary cost to the family or heirs of the deceased.

Signature of parent/legal guardian/next of kin	Relationship to deceased	Date
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Signature of Witness	Printed name of Witness	Date
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Permit for Release of Body to Funeral Home

I hereby authorize and request the Greenwood Genetic Center to release the body of _____, deceased, to the _____

Funeral home located in _____
City State

Signature of parent/legal guardian/next of kin	Relationship to deceased	Date
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Signature of Witness	Printed name of Witness	Date
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Please use ball point pen and write firmly. WHITE COPY- to accompany specimen. YELLOW COPY- Original record.

**Greenwood Genetic Center
Fetal Examination Service**

AUTHORIZATION TO RELEASE MEDICAL RECORDS

A copy of the Genetic Evaluation report will be sent to the referring physician, which is either the mother's obstetrician or the infant's pediatrician or neonatologist. Many times, it is important that other physicians or genetic counselors who have been involved in the care of the mother or the baby also receive a copy of this report. Please sign and note below which health care providers, other than the referring physician, you want to receive a copy of this report.

Mother's Name: _____ DOB: _____

Infant's Name (if applicable) _____ DOB: _____

By my signature below, I hereby authorize the Greenwood Genetic Center to release medical information to the following physicians/counselors/facilities:

Mother's Signature _____ Date _____

Physicians:

Name Location (Street or Office name, City, Phone No.)

Name Location (Street or Office name, City, Phone No.)

Name Location (Street or Office name, City, Phone No.)

Genetic counselor:

Name Location (Street or Office name, City, Phone No.)

Other:

Name Location (Street or Office name, City, Phone No.)

Courier Services

Fetal Courier Information

Courier service Monday-Friday

Beavex

Greenville 1-864-801-2720

Beavex

Columbia 1-803-359-0088

SPECIAL PICKUPS WILL BE THE RESPONSIBILITY OF THE REFERRING HOSPITAL OR OFFICE. THIS INCLUDES SCHEDULING A COURIER SERVICE AND CHARGES.

Courier contacts:

<u>HOSPITAL or OFFICE</u>	<u>CONTACT</u>	<u>TELEPHONE</u>	<u>REGULAR PICK UP SCH.</u>
GREENVILLE			
Greenville Memorial Lab.	Sandra Bruce	864-455-3833	10:00 AM
St. Francis Hospital	Debbie Ellenburg	864-255-1670	Call Beavex For Pick Up
St. Francis Eastside Hospital Lab.	Jennifer Clayton or Mira Vadhar	864-675-4036	Call Beavex For Pick Up
Pathology Consultants (Greenville)	Joanne Taylor Lynn Ziegler or Curt Nauss	864-675-4033 864-295-3482	Call Beavex For Pick Up
SPARTANBURG			
Mary Black Hospital	Michael Russell	864-573-3784	Call Beavex For Pick Up
Spartanburg Reg. Med. Ctr. Clinical Lab	Claudia Tirpak	864-560-6450	8:30 AM
Pathology	Claudia Tirpak	864-560-6450	Call Beavex For Pick Up
COLUMBIA			
Palmetto Health Richland Histology	Kristen Rose	803-434-6710	10:00-10:30 AM Call Beavex by 8:00 A.M. F/Fetal Pick Up
FLORENCE			
McLeod OB/GYN	Denise Dover	843-669-1264	Call Beavex For Pick Up
McLeod Reg. Med. Ctr. Lab	Rebecca Early, M.T.	843-777-2978	3:00 PM Call Beavex F/Fetal Pick Up
PeeDee Pathology (FETAL PICK UP ONLY, NOT TISSUE)	Cheryl Atkinson or Clarissa White	843-664-4314	3:05 PM Call Beavex by Noon F/Fetal P/U

- The specimen must be available for immediate transfer to courier—they will not wait. Please check the time the courier arrives at the office or laboratory and have the specimen ready and available.**
- Some hospitals use their own courier service.
- The specimen can be transported by a family member or friend if the mother gives signed consent specifying the individuals name. The family member/friend will be required to show proper identification. The specimen must be appropriately packaged and there must be a burial transit permit, which is generated at the referring institution.
- No back-transport to funeral homes or hospitals is available.** Funeral homes must make arrangements with the Greenwood Genetic Center for pick up of specimen.



Molecular Diagnostic Request Form

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	<input type="checkbox"/> B <input type="checkbox"/> W <input type="checkbox"/> Other:	Sex	<input type="checkbox"/> M <input type="checkbox"/> F	DOB	MM / DD / YYYY
Specimen Collection Date	Type of specimen	ICD9 Code	Numeric Identifier (MR # or SS #)		Home telephone
MM / DD / YYYY					

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		

Genetic Counselor/Care coordinator:

Name		Address	
Telephone	Fax	City, State, Zip	

Billing: For in-state insurance billing, include copy of card and insured's name, DOB, and relationship to patient.

We DO NOT bill out of state patients or insurance companies. We accept institutional billing or check/Visa/MasterCard.

Institution/Organization	Telephone	Fax	
Address		City, State, Zip	
MasterCard # Visa # (circle one)	Exp. Date	Signature	Auth/Precert #

Indication For Study:

<input type="checkbox"/> Unknown mutation(s) Please list clinical features _____ _____ <input type="checkbox"/> Family History _____ <input type="checkbox"/> Known mutation(s) _____ <input type="checkbox"/> Population Screening/ Other _____ Is the patient currently pregnant? <input type="checkbox"/> Yes <input type="checkbox"/> No If Yes, LMP date: _____ OR EDC: _____	Pedigree
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Comments:

Attach clinical information and/or family history. A brief pedigree can be drawn above or attached separately

If multiple tests are requested, please indicate the order the testing should be completed or if all tests should be performed simultaneously.

Last Name	First	MI	DOB	SS#
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**Maternal cell contamination analysis is required with all prenatal studies.
Please submit separate request forms for prenatal and maternal samples.**

- 3-Methylcrotonylglycinuria (*MCCC1* and *MCCC2*) Sequencing
- 3-Methylcrotonylglycinuria (*MCCC1* and *MCCC2*) Del/Dup ♣
- Aarskog syndrome (*FGD1*) Sequencing
- Aarskog syndrome (*FGD1*) Del/Dup ♣
- ACSL4*-related X-linked intellectual disability - Seq
- ACSL4*-related X-linked intellectual disability - Del/Dup ♣
- Adrenoleukodystrophy, X-linked (*ABCD1*) Sequencing
- Adrenoleukodystrophy, X-linked (*ABCD1*) Del/Dup ♣
- Aminoglycoside-induced hearing loss (*MTRNR1*)
- Allan Herndon Dudley syndrome (*MCT8*) Sequencing
- Allan Herndon Dudley syndrome (*MCT8*) Del/Dup ♣
- Alpha-Mannosidosis (*MAN2B1*) Sequencing
- Alpha-Mannosidosis (*MAN2B1*) Del/Dup ♣
- Angelman syndrome (check all that apply)
 - Methylation analysis
 - UBE3A* Sequencing
 - UBE3A* Deletion/Duplication ♣
- ARX*-related spectrum (*ARX*) Sequencing
- ARX*-related spectrum (*ARX*) Del/Dup ♣
- Aspartylglycosaminuria (*AGA*) Sequencing
- Aspartylglycosaminuria (*AGA*) Del/Dup ♣
- ATRX* syndrome (*XNP*) Sequencing
- ATRX* syndrome (*XNP*) Del/Dup ♣
- Beckwith-Wiedemann syndrome Methylation/MLPA
- Beta-mannosidosis (*MANBA*) Sequencing
- Beta-mannosidosis (*MANBA*) Del/Dup ♣
- Biotinidase deficiency (*BTD*) Sequencing
- Biotinidase deficiency (*BTD*) Del/Dup ♣
- Borjeson-Forsman-Lehmann Syndrome (*PHF6*) Sequencing
- Borjeson-Forsman-Lehmann Syndrome (*PHF6*) Del/Dup ♣
- Cardio-Facio-Cutaneous (CFC) syndrome (check all that apply)
 - Tier 1 (*BRAF*) Sequencing
 - Tier 2 (*MAP2K1* and *MAP2K2*) Sequencing
 - Tier 3 (*KRAS*) Sequencing
 - Full CFC syndrome panel (Tiers 1, 2 and 3) - Sequencing
- Cardio-Facio-Cutaneous (CFC) syndrome - Del/Dup (all tiers) ♣
- Carnitine Palmitoyltransferase Deficiency 1A (*CPT1A*) Seq
- Carnitine Palmitoyltransferase Deficiency 1A (*CPT1A*) Del/Dup ♣
- Carnitine Palmitoyltransferase II Deficiency (*CPT2*) Seq
- Carnitine Palmitoyltransferase II Deficiency (*CPT2*) Del/Dup ♣
- CASK*-related X-linked intellectual disability - Sequencing
- CASK*-related X-linked intellectual disability - Del/Dup ♣
- CDKL5* - Atypical Rett syndrome - Sequencing
- CDKL5* - Atypical Rett syndrome - Del/Dup ♣
- CHD7*-related disorders: CHARGE or Kallmann syndrome 5 - Seq
- CHD7*-related disorders: CHARGE or Kallmann syndrome 5 - Del/Dup ♣
- Christianson syndrome /X-linked Angelman (*SLC9A6*) Seq
- Christianson syndrome /X-linked Angelman (*SLC9A6*) Del/Dup ♣
- Citrullinemia, Type 1 (*ASS1*) Sequencing
- Citrullinemia, Type 1 (*ASS1*) Del/Dup ♣
- Coffin-Lowry syndrome (*RPS6KA3*) Sequencing
- Coffin-Lowry syndrome (*RPS6KA3*) Del/Dup ♣
- Congenital Disorders of Glycosylation type 1a (*PMM2*) Seq
- Congenital Disorders of Glycosylation type 1a (*PMM2*) Del/Dup ♣
- Congenital Disorders of Glycosylation type 1b (*MPI*) Seq
- Congenital Disorders of Glycosylation type 1b (*MPI*) Del/Dup ♣
- Congenital Disorders of Glycosylation type 1c (*ALG6*) Seq
- Congenital Disorders of Glycosylation type 1c (*ALG6*) Del/Dup ♣
- Connexin 26 (*GJB2*) Sequencing
- Connexin 26 (*GJB2*) Del/Dup ♣
- Copper Transport Disorders (*ATP7A*) Sequencing
- Copper Transport Disorders (*ATP7A*) Del/Dup ♣
- Costello syndrome (check one) - Sequencing
 - Tier 1 (*HRAS*, first coding exon sequencing)
 - Tier 2 (*HRAS*, remaining exons sequencing)
 - Full Costello syndrome panel (Tiers 1 and 2)
- Costello syndrome - Del/Dup (all tiers included) ♣
- Creatine Transporter Deficiency syndrome (*SLC6A8*) Seq * PAX
- Creatine Transporter Deficiency syndrome (*SLC6A8*) Del/Dup ♣
- Cystic Fibrosis (*CFTR*) includes ACMG/ACOG panel
- DCX*-related lissencephaly & subcortical band heterotopia - Seq
- DCX*-related Disorders - Del/Dup ♣
- DMD/BMD deletion/duplication detection (MLPA)
- Early Infantile Epileptic Encephalopathy 4 (*STXBP1*) Seq
- Early Infantile Epileptic Encephalopathy 4 (*STXBP1*) Del/Dup ♣
- FGFR2*-related disorders (check all that apply)
 - Apert syndrome
 - Beare-Stevenson with cutis gyrata
 - Crouzon syndrome
 - Jackson-Weiss syndrome
 - Pfeiffer syndrome
 - Other _____
- FGFR2*-related disorders - Del/Dup ♣
- FGFR3*-related disorders (must select the phenotype(s) below)
 - Achondroplasia
 - Crouzon with acanthosis nigricans
 - Hypochondroplasia
 - Non-syndromic craniosynostosis
 - Thanatophoric dysplasia type I
 - Thanatophoric dysplasia type II
 - Other _____
- FGFR3*-related disorders - Del/Dup ♣
- FLNA*-related disorders - Sequencing
 - Otopalatodigital Spectrum Disorders
 - X-linked Periventricular Heterotopia
 - X-linked Periventricular Heterotopia, Ehlers-Danlos type
 - X-linked Cardiac Valvular Dysplasia
 - X-linked Chronic Idiopathic Neuronal Intestinal Pseudoobstruction
- FLNA*-related disorders - Del/Dup ♣
- Fragile X syndrome (*FMR1*) triplet repeat analysis
- FRAXE syndrome (*FMR2*) triplet repeat analysis
- Fucosidosis (*FUCA1*) Sequencing
- Fucosidosis (*FUCA1*) Del/Dup ♣
- Galactosemia, Classic (*GALT*) Sequencing
- Galactosemia, Classic (*GALT*) Del/Dup ♣
- Galactosialidosis (*CTSA*) Sequencing
- Galactosialidosis (*CTSA*) Del/Dup ♣
- Gaucher Disease (*GBA*) Sequencing
- Gaucher Disease (*GBA*) Del/Dup ♣
- Glutaric acidemia, type 1 (*GCDH*) Sequencing
- Glutaric acidemia, type 1 (*GCDH*) Del/Dup ♣
- *GLI3*-related Disorders (check one) Sequencing
 - Greig cephalopolysyndactyly
 - Pallister-Hall syndrome
 - Isolated postaxial polysyndactyly
- GLI3*-related Disorders - Del/Dup ♣
- GM1 gangliosidosis (*GLB1*) Sequencing
- GM1 gangliosidosis (*GLB1*) Del/Dup ♣
- Hemochromatosis (*HFE*) mutation analysis
- Hemochromatosis (*HFE*) Del/Dup ♣
- Hunter syndrome (*IDS*) Sequencing
- Hunter syndrome (*IDS*) Del/Dup (MLPA)
- Hurler Syndrome (*IDUA*) Sequencing
- Hurler Syndrome (*IDUA*) Del/Dup ♣
- Kabuki syndrome (*MLL2*) Sequencing
- Kabuki syndrome (*MLL2*) Del/Dup ♣

All assays require a purple top (EDTA) tube unless otherwise specified
 * Requires Qiagen PAXGENE tube (available upon request) and purple top (EDTA) tube
 ♣ Single gene del/dup analysis via custom array



Molecular Diagnostic Request Form

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

Last Name	First	MI	DOB	SS#
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- Krabbe Disease (*GALC*) Sequencing
- Krabbe Disease (*GALC*) Del/Dup ♣
- Leopard syndrome (check one)
 - Tier 1 (*PTPN11*) - sequencing
 - Tier 2 (*RAF1* exons 7, 14 and 17) - sequencing
 - Tier 3 (*BRAF*) - sequencing
 - Full Leopard syndrome panel (Tiers 1, 2, and 3) - sequencing
- Leopard syndrome Del/Dup (all tiers included)
- Marfan syndrome (*FBN1*) Sequencing
- Marfan syndrome (*FBN1*) Del/Dup ♣
- Maroteaux-Lamy Syndrome (*ARSB*) Sequencing
- Maroteaux-Lamy Syndrome (*ARSB*) Del/Dup ♣
- Maternal Cell Contamination
- MCAD (*ACADM*) Sequencing
- MCAD (*ACADM*) Del/Dup ♣
- MED12 related disorders** (exons 4, 5, 20, 21, 22, 28, 36) Seq
 - Circle one : Lujan-Fryns syndrome FG syndrome
- MED12 related disorders - Del/Dup ♣**
 - Circle one : Lujan-Fryns syndrome FG syndrome
- Morquio Syndrome A, MPS IVA (*GALNS*) Sequencing
- Morquio Syndrome A, MPS IVA (*GALNS*) Del/Dup ♣
- Morquio Syndrome B, MPS IVB (*GLB1*) Sequencing
- Morquio Syndrome B, MPS IVB (*GLB1*) Del/Dup ♣
- Mucopolipidosis II & III Alpha/Beta (*GNPTAB*) Sequencing
- Mucopolipidosis II & III Alpha/Beta (*GNPTAB*) Del/Dup ♣
- Mucopolipidosis III Gamma (*GNPTG*) Sequencing
- Mucopolipidosis III Gamma (*GNPTG*) Del/Dup ♣
- Myotonic dystrophy (*DM1*) triplet repeat analysis
- Myotubular Myopathy, X-linked (*MTM1*) Sequencing
- Myotubular Myopathy, X-linked (*MTM1*) Del/Dup ♣
- Noonan syndrome (check one)
 - Tier 1 (*PTPN11*)
 - Tier 2 (*SOS1*)
 - Tier 3 (*RAF1* & *KRAS* and *SHOC2* – p.S2G mutation only)
 - Tier 4 (*BRAF*, *MAP2K1* and *NRAS* sequencing)
 - Full Noonan syndrome panel (Tiers 1, 2, 3, & 4)
- Noonan syndrome – Del/Dup (all tiers included) ♣
- OPHN1*-related X-linked intellectual disability - Sequencing
- OPHN1*-related X-linked intellectual disability - Del/Dup ♣
- Ornithine transcarbamylase deficiency (*OTC*) Sequencing
- Ornithine transcarbamylase deficiency (*OTC*) Del/Dup ♣
- P63-related disorders (check one)
 - EEC syndrome
 - Isolated slit-hand/foot malformation
 - Hay-Wells syndrome
 - Other _____
- Pelizaeus-Merzbacher Disease, Spastic paraplegia 2 (*PLP1*) Seq
- PLP1* deletion/duplication (MLPA)
- Phenylketonuria (*PAH*) Sequencing
- Phenylketonuria (*PAH*) Del/Dup ♣
- Pitt-Hopkins syndrome (*TCF4*) Sequencing
- Pitt-Hopkins syndrome (*TCF4*) Del/Dup ♣
- POLG1*-related disorders - Sequencing
- POLG1*-related disorders - Del/Dup ♣
- Prader Willi syndrome, Methylation analysis
- Primary Carnitine Deficiency, systemic (*SLC22A5*) Sequencing
- Primary Carnitine Deficiency, systemic (*SLC22A5*) Del/Dup ♣
- PTEN related disorders** (check one) Sequencing
 - Autism with macrocephaly
 - Bannayan-Riley-Ruvalcaba syndrome
 - Cowden syndrome
 - Proteus-like syndrome
- PTEN deletion/duplication (MLPA)**
- Renpenning Syndrome (*PQBP1*) Sequencing
- Renpenning Syndrome (*PQBP1*) Del/Dup ♣
- Rett syndrome (check one)
 - MECP2* sequencing
 - MECP2* Deletion/duplication detection (MLPA)
 - Atypical Rett syndrome - *CDKL5* (*STK9*) Sequencing
 - Atypical Rett syndrome - *CDKL5* (*STK9*) Del/Dup ♣
 - Congenital Rett variant (*FOXP1*) Sequencing
 - Congenital Rett variant (*FOXP1*) Del/Dup ♣
- Russell-Silver syndrome (11p15.5 related) Methylation/MLPA
- Saethre-Chotzen (*TWIST*) Sequencing
- Saethre-Chotzen (*TWIST*) Del/Dup (MLPA)
- Sanfilippo A (*SGSH*) Sequencing
- Sanfilippo A (*SGSH*) Del/Dup ♣
- Sanfilippo B (*NAGLU*) Sequencing
- Sanfilippo B (*NAGLU*) Del/Dup ♣
- Sanfilippo C (*HGSNAT*) Sequencing
- Sanfilippo C (*HGSNAT*) Del/Dup ♣
- Sanfilippo D (*GNS*) Sequencing
- Sanfilippo D (*GNS*) Del/Dup ♣
- Sialidosis (*NEU1*) Sequencing
- Sialidosis (*NEU1*) Del/Dup ♣
- Simpson-Golabi-Behmel Syndrome Type 1 (*GPC3*) Seq
- Simpson-Golabi-Behmel Syndrome Type 1 (*GPC3*) Del/Dup ♣
- Sotos syndrome *NSD1* Full sequencing
- Sotos syndrome *NSD1* Del/Dup (MLPA)
- Sly syndrome, MPS VII (*GUSB*) Sequencing
- Sly syndrome, MPS VII (*GUSB*) Del/Dup ♣
- Thrombosis Panel
 - Factor V Leiden
 - Prothrombin c.G20210A
- Uniparental Disomy–parental samples required -check one
 - Chromosome 7 (Russell Silver syndrome UPD 7)
 - Chromosome 14 (UPD 14)
 - Chromosome 15 (Angelman/Prader-Willi syndrome UPD 15)
- VLCAD deficiency (*ACADVL*) Sequencing
- VLCAD deficiency (*ACADVL*) Del/Dup ♣
- X-inactivation analysis
- X-linked Hydrocephalus (*L1CAM*) Sequencing
- X-linked Hydrocephalus (*L1CAM*) Del/Dup ♣
- X-linked, Female Limited Epilepsy w/ ID (*PCDH19*) Seq
- X-linked, Female Limited Epilepsy w/ ID (*PCDH19*) Del/Dup ♣
- X-linked Opitz G/BBB Syndrome (*MID1*) Sequencing
- X-linked Opitz G/BBB Syndrome (*MID1*) Del/Dup ♣
- Other: _____
- DNA Banking

♣ Single gene del/dup analysis via custom array

Next Generation Sequencing Panels

All NGS panels require a separate requisition form. These forms can be found on the website.

Connective Tissue Disorders (31 genes)
 Epilepsy/Seizures (103 genes)
 Lysosomal Storage Disorders (74 genes)
 Skeletal Dysplasias (10 genes)

Syndromic Autism (62 genes)
 2nd Tier Rett/Angelman (19 genes)
 X-linked Intellectual Disability (90 genes)



Molecular Diagnostic Request Form

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	<input type="checkbox"/> B <input type="checkbox"/> W <input type="checkbox"/> Other:	Sex	<input type="checkbox"/> M <input type="checkbox"/> F	DOB	MM / DD / YYYY
Specimen Collection Date	Type of specimen	ICD9 Code	Numeric Identifier (MR # or SS #)		Home telephone
MM / DD / YYYY					

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		

Genetic Counselor/Care coordinator:

Name		Address	
Telephone	Fax	City, State, Zip	

Billing: For in-state insurance billing, include copy of card and insured's name, DOB, and relationship to patient.

We DO NOT bill out of state patients or insurance companies. We accept institutional billing or check/Visa/MasterCard.

Institution/Organization	Telephone	Fax	
Address		City, State, Zip	
MasterCard # Visa # (circle one)	Exp. Date	Signature	Auth/Precert #

Indication For Study:

<input type="checkbox"/> Unknown mutation(s) Please list clinical features _____ _____ <input type="checkbox"/> Family History _____ <input type="checkbox"/> Known mutation(s) _____ <input type="checkbox"/> Population Screening/ Other _____ Is the patient currently pregnant? <input type="checkbox"/> Yes <input type="checkbox"/> No If Yes, LMP date: _____ OR EDC: _____	Pedigree
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Comments:

_____ _____ <p style="text-align: center;">Attach clinical information and/or family history. A brief pedigree can be drawn above or attached separately</p>

If multiple tests are requested, please indicate the order the testing should be completed or if all tests should be performed simultaneously.

Last Name	First	MI	DOB	SS#
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**Maternal cell contamination analysis is required with all prenatal studies.
Please submit separate request forms for prenatal and maternal samples.**

- 3-Methylcrotonylglycinuria (MCCC1 and MCCC2) Sequencing
- 3-Methylcrotonylglycinuria (MCCC1 and MCCC2) Del/Dup ♣
- Aarskog syndrome (FGD1) Sequencing
- Aarskog syndrome (FGD1) Del/Dup ♣
- ACSL4-related X-linked intellectual disability - Seq
- ACSL4-related X-linked intellectual disability - Del/Dup ♣
- Adrenoleukodystrophy, X-linked (ABCD1) Sequencing
- Adrenoleukodystrophy, X-linked (ABCD1) Del/Dup ♣
- Aminoglycoside-induced hearing loss (MTRNR1)
- Allan Herndon Dudley syndrome (MCT8) Sequencing
- Allan Herndon Dudley syndrome (MCT8) Del/Dup ♣
- Alpha-Mannosidosis (MAN2B1) Sequencing
- Alpha-Mannosidosis (MAN2B1) Del/Dup ♣
- Angelman syndrome (check all that apply)
 - Methylation analysis
 - UBE3A Sequencing
 - UBE3A Deletion/Duplication ♣
- ARX-related spectrum (ARX) Sequencing
- ARX-related spectrum (ARX) Del/Dup ♣
- Aspartylglycosaminuria (AGA) Sequencing
- Aspartylglycosaminuria (AGA) Del/Dup ♣
- ATRX syndrome (XNP) Sequencing
- ATRX syndrome (XNP) Del/Dup ♣
- Beckwith-Wiedemann syndrome Methylation/MLPA
- Beta-mannosidosis (MANBA) Sequencing
- Beta-mannosidosis (MANBA) Del/Dup ♣
- Biotinidase deficiency (BTD) Sequencing
- Biotinidase deficiency (BTD) Del/Dup ♣
- Borjeson-Forsman-Lehmann Syndrome (PHF6) Sequencing
- Borjeson-Forsman-Lehmann Syndrome (PHF6) Del/Dup ♣
- Cardio-Facio-Cutaneous (CFC) syndrome (check all that apply)
 - Tier 1 (BRAF) Sequencing
 - Tier 2 (MAP2K1 and MAP2K2) Sequencing
 - Tier 3 (KRAS) Sequencing
 - Full CFC syndrome panel (Tiers 1, 2 and 3) - Sequencing
- Cardio-Facio-Cutaneous (CFC) syndrome - Del/Dup (all tiers) ♣
- Carnitine Palmitoyltransferase Deficiency 1A (CPT1A) Seq
- Carnitine Palmitoyltransferase Deficiency 1A (CPT1A) Del/Dup ♣
- Carnitine Palmitoyltransferase II Deficiency (CPT2) Seq
- Carnitine Palmitoyltransferase II Deficiency (CPT2) Del/Dup ♣
- CASK-related X-linked intellectual disability - Sequencing
- CASK-related X-linked intellectual disability - Del/Dup ♣
- CDKL5 - Atypical Rett syndrome - Sequencing
- CDKL5 - Atypical Rett syndrome - Del/Dup ♣
- CHD7-related disorders: CHARGE or Kallmann syndrome 5 - Seq
- CHD7-related disorders: CHARGE or Kallmann syndrome 5 - Del/Dup ♣
- Christianson syndrome /X-linked Angelman (SLC9A6) Seq
- Christianson syndrome /X-linked Angelman (SLC9A6) Del/Dup ♣
- Citrullinemia, Type 1 (ASS1) Sequencing
- Citrullinemia, Type 1 (ASS1) Del/Dup ♣
- Coffin-Lowry syndrome (RPS6KA3) Sequencing
- Coffin-Lowry syndrome (RPS6KA3) Del/Dup ♣
- Congenital Disorders of Glycosylation type 1a (PMM2) Seq
- Congenital Disorders of Glycosylation type 1a (PMM2) Del/Dup ♣
- Congenital Disorders of Glycosylation type 1b (MPI) Seq
- Congenital Disorders of Glycosylation type 1b (MPI) Del/Dup ♣
- Congenital Disorders of Glycosylation type 1c (ALG6) Seq
- Congenital Disorders of Glycosylation type 1c (ALG6) Del/Dup ♣
- Connexin 26 (GJB2) Sequencing
- Connexin 26 (GJB2) Del/Dup ♣
- Copper Transport Disorders (ATP7A) Sequencing
- Copper Transport Disorders (ATP7A) Del/Dup ♣
- Costello syndrome (check one) - Sequencing
 - Tier 1 (HRAS, first coding exon sequencing)
 - Tier 2 (HRAS, remaining exons sequencing)
 - Full Costello syndrome panel (Tiers 1 and 2)
- Costello syndrome - Del/Dup (all tiers included) ♣
- Creatine Transporter Deficiency syndrome (SLC6A8) Seq * PAX
- Creatine Transporter Deficiency syndrome (SLC6A8) Del/Dup ♣
- Cystic Fibrosis (CFTR) includes ACMG/ACOG panel
- DCX-related lissencephaly & subcortical band heterotopia - Seq
- DCX-related Disorders - Del/Dup ♣
- DMD/BMD deletion/duplication detection (MLPA)
- Early Infantile Epileptic Encephalopathy 4 (STXBP1) Seq
- Early Infantile Epileptic Encephalopathy 4 (STXBP1) Del/Dup ♣
- FGFR2-related disorders (check all that apply)
 - Apert syndrome
 - Beare-Stevenson with cutis gyrata
 - Crouzon syndrome
 - Jackson-Weiss syndrome
 - Pfeiffer syndrome
 - Other _____
- FGFR2-related disorders - Del/Dup ♣
- FGFR3-related disorders (must select the phenotype(s) below)
 - Achondroplasia
 - Crouzon with acanthosis nigricans
 - Hypochondroplasia
 - Non-syndromic craniosynostosis
 - Thanatophoric dysplasia type I
 - Thanatophoric dysplasia type II
 - Other _____
- FGFR3-related disorders - Del/Dup ♣
- FLNA-related disorders - Sequencing
 - Otopalatodigital Spectrum Disorders
 - X-linked Periventricular Heterotopia
 - X-linked Periventricular Heterotopia, Ehlers-Danlos type
 - X-linked Cardiac Valvular Dysplasia
 - X-linked Chronic Idiopathic Neuronal Intestinal Pseudoobstruction
- FLNA-related disorders - Del/Dup ♣
- Fragile X syndrome (FMR1) triplet repeat analysis
- FRAXE syndrome (FMR2) triplet repeat analysis
- Fucosidosis (FUCA1) Sequencing
- Fucosidosis (FUCA1) Del/Dup ♣
- Galactosemia, Classic (GALT) Sequencing
- Galactosemia, Classic (GALT) Del/Dup ♣
- Galactosialidosis (CTSA) Sequencing
- Galactosialidosis (CTSA) Del/Dup ♣
- Gaucher Disease (GBA) Sequencing
- Gaucher Disease (GBA) Del/Dup ♣
- Glutaric acidemia, type 1 (GCDH) Sequencing
- Glutaric acidemia, type 1 (GCDH) Del/Dup ♣
- GLI3-related Disorders (check one) Sequencing
 - Greig cephalopolysyndactyly
 - Pallister-Hall syndrome
 - Isolated postaxial polysyndactyly
- GLI3-related Disorders - Del/Dup ♣
- GM1 gangliosidosis (GLB1) Sequencing
- GM1 gangliosidosis (GLB1) Del/Dup ♣
- Hemochromatosis (HFE) mutation analysis
- Hemochromatosis (HFE) Del/Dup ♣
- Hunter syndrome (IDS) Sequencing
- Hunter syndrome (IDS) Del/Dup (MLPA)
- Hurler Syndrome (IDUA) Sequencing
- Hurler Syndrome (IDUA) Del/Dup ♣
- Kabuki syndrome (MLL2) Sequencing
- Kabuki syndrome (MLL2) Del/Dup ♣

All assays require a purple top (EDTA) tube unless otherwise specified
 * Requires Qiagen PAXGENE tube (available upon request) and purple top (EDTA) tube
 ♣ Single gene del/dup analysis via custom array



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Last Name	First	MI	DOB	SS#
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- Krabbe Disease (*GALC*) Sequencing
- Krabbe Disease (*GALC*) Del/Dup ♣
 - Leopard syndrome (check one)
 - Tier 1 (*PTPN11*) - sequencing
 - Tier 2 (*RAF1* exons 7, 14 and 17) - sequencing
 - Tier 3 (*BRAF*) - sequencing
 - Full Leopard syndrome panel (Tiers 1, 2, and 3) - sequencing
 - Leopard syndrome Del/Dup (all tiers included)
 - Marfan syndrome (*FBN1*) Sequencing
 - Marfan syndrome (*FBN1*) Del/Dup ♣
 - Maroteaux-Lamy Syndrome (*ARSB*) Sequencing
 - Maroteaux-Lamy Syndrome (*ARSB*) Del/Dup ♣
 - Maternal Cell Contamination
 - MCAD (*ACADM*) Sequencing
 - MCAD (*ACADM*) Del/Dup ♣
 - MED12* related disorders (exons 4, 5, 20, 21, 22, 28, 36) Seq
 - Circle one : Lujan-Fryns syndrome FG syndrome
 - MED12* related disorders - Del/Dup ♣
 - Circle one : Lujan-Fryns syndrome FG syndrome
 - Morquio Syndrome A, MPS IVA (*GALNS*) Sequencing
 - Morquio Syndrome A, MPS IVA (*GALNS*) Del/Dup ♣
 - Morquio Syndrome B, MPS IVB (*GLB1*) Sequencing
 - Morquio Syndrome B, MPS IVB (*GLB1*) Del/Dup ♣
 - Mucopolipidosis II & III Alpha/Beta (*GNPTAB*) Sequencing
 - Mucopolipidosis II & III Alpha/Beta (*GNPTAB*) Del/Dup ♣
 - Mucopolipidosis III Gamma (*GNPTG*) Sequencing
 - Mucopolipidosis III Gamma (*GNPTG*) Del/Dup ♣
 - Myotonic dystrophy (*DM1*) triplet repeat analysis
 - Myotubular Myopathy, X-linked (*MTM1*) Sequencing
 - Myotubular Myopathy, X-linked (*MTM1*) Del/Dup ♣
 - Noonan syndrome (check one)
 - Tier 1 (*PTPN11*)
 - Tier 2 (*SOS1*)
 - Tier 3 (*RAF1* & *KRAS* and *SHOC2* – p.S2G mutation only)
 - Tier 4 (*BRAF*, *MAP2K1* and *NRAS* sequencing)
 - Full Noonan syndrome panel (Tiers 1, 2, 3, & 4)
 - Noonan syndrome – Del/Dup (all tiers included) ♣
 - OPHN1*-related X-linked intellectual disability - Sequencing
 - OPHN1*-related X-linked intellectual disability - Del/Dup ♣
 - Ornithine transcarbamylase deficiency (*OTC*) Sequencing
 - Ornithine transcarbamylase deficiency (*OTC*) Del/Dup ♣
 - *P63*-related disorders (check one)
 - EEC syndrome
 - Isolated slit-hand/foot malformation
 - Hay-Wells syndrome
 - Other _____
 - Pelizaeus-Merzbacher Disease, Spastic paraplegia 2 (*PLP1*) Seq
 - PLP1* deletion/duplication (MLPA)
 - Phenylketonuria (*PAH*) Sequencing
 - Phenylketonuria (*PAH*) Del/Dup ♣
 - Pitt-Hopkins syndrome (*TCF4*) Sequencing
 - Pitt-Hopkins syndrome (*TCF4*) Del/Dup ♣
 - POLG1*-related disorders - Sequencing
 - POLG1*-related disorders - Del/Dup ♣
 - Prader Willi syndrome, Methylation analysis
- Primary Carnitine Deficiency, systemic (*SLC22A5*) Sequencing
- Primary Carnitine Deficiency, systemic (*SLC22A5*) Del/Dup ♣
- PTEN* related disorders (check one) Sequencing
 - Autism with macrocephaly
 - Bannayan-Riley-Ruvalcaba syndrome
 - Cowden syndrome
 - Proteus-like syndrome
- PTEN* deletion/duplication (MLPA)
- Renpenning Syndrome (*PQBP1*) Sequencing
- Renpenning Syndrome (*PQBP1*) Del/Dup ♣
- Rett syndrome (check one)
 - MECP2* sequencing
 - MECP2* Deletion/duplication detection (MLPA)
 - Atypical Rett syndrome - *CDKL5* (*STK9*) Sequencing
 - Atypical Rett syndrome - *CDKL5* (*STK9*) Del/Dup ♣
 - Congenital Rett variant (*FOXP1*) Sequencing
 - Congenital Rett variant (*FOXP1*) Del/Dup ♣
- Russell-Silver syndrome (11p15.5 related) Methylation/MLPA
- Saethre-Chotzen (*TWIST*) Sequencing
- Saethre-Chotzen (*TWIST*) Del/Dup (MLPA)
- Sanfilippo A (*SGSH*) Sequencing
- Sanfilippo A (*SGSH*) Del/Dup ♣
- Sanfilippo B (*NAGLU*) Sequencing
- Sanfilippo B (*NAGLU*) Del/Dup ♣
- Sanfilippo C (*HGSNAT*) Sequencing
- Sanfilippo C (*HGSNAT*) Del/Dup ♣
- Sanfilippo D (*GNS*) Sequencing
- Sanfilippo D (*GNS*) Del/Dup ♣
- Sialidosis (*NEU1*) Sequencing
- Sialidosis (*NEU1*) Del/Dup ♣
- Simpson-Golabi-Behmel Syndrome Type 1 (*GPC3*) Seq
- Simpson-Golabi-Behmel Syndrome Type 1 (*GPC3*) Del/Dup ♣
- Sotos syndrome *NSD1* Full sequencing
- Sotos syndrome *NSD1* Del/Dup (MLPA)
- Sly syndrome, MPS VII (*GUSB*) Sequencing
- Sly syndrome, MPS VII (*GUSB*) Del/Dup ♣
- Thrombosis Panel
 - Factor V Leiden
 - Prothrombin c.G20210A
- Uniparental Disomy–parental samples required -check one
 - Chromosome 7 (Russell Silver syndrome UPD 7)
 - Chromosome 14 (UPD 14)
 - Chromosome 15 (Angelman/Prader-Willi syndrome UPD 15)
- VLCAD deficiency (*ACADVL*) Sequencing
- VLCAD deficiency (*ACADVL*) Del/Dup ♣
- X-inactivation analysis
- X-linked Hydrocephalus (*L1CAM*) Sequencing
- X-linked Hydrocephalus (*L1CAM*) Del/Dup ♣
- X-linked, Female Limited Epilepsy w/ ID (*PCDH19*) Seq
- X-linked, Female Limited Epilepsy w/ ID (*PCDH19*) Del/Dup ♣
- X-linked Opitz G/BBB Syndrome (*MID1*) Sequencing
- X-linked Opitz G/BBB Syndrome (*MID1*) Del/Dup ♣

♣ Single gene del/dup analysis via custom array

Next Generation Sequencing Panels

All NGS panels require a separate requisition form. These forms can be found on the website.

Connective Tissue Disorders (31 genes)
 Epilepsy/Seizures (103 genes)
 Lysosomal Storage Disorders (74 genes)
 Skeletal Dysplasias (10 genes)

Syndromic Autism (62 genes)
 2nd Tier Rett/Angelman (19 genes)
 X-linked Intellectual Disability (90 genes)