Greenwood Genetic Center
Where Compassion Inspires Progress

Greenwood Genetic Center
106 Gregor Mendel Circle ● Greenwood, South Carolina 29646
(864) 941-8100 ● 1-888-GGC-GENE (Toll Free)
FAX (864) 941-8114

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.**

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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. **Please call the GGC Laboratory for details or questions at 1-800-473-9411.**

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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.

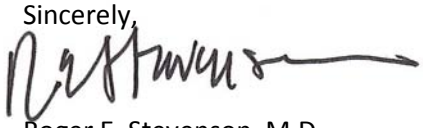
A complete study also requires that we review medical records (prenatal, delivery, previous pregnancies). We hope that this effort will be helpful to you and your physician, to explain the recent events and to help in the planning and management of future pregnancies in your family.

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

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
Signature of parent/legal guardian/next of kin	Relationship to deceased	Date
Signature of Witness	Printed name of Witness	Date

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
Signature of Witness	Printed name of Witness	Date

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
Signature of Witness	Printed name of Witness	Date

Please use ball point pen and write firmly. WHITE COPY- to accompany specimen. YELLOW COPY- Original record.

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GREENWOOD GENETIC CENTER
FETAL EXAMINATION SERVICE

AUTHORIZATION TO OBTAIN MEDICAL RECORDS

Addressograph

The fetal examination offered by the Greenwood Genetic Center is comprehensive evaluation that includes the mother's medical history, pregnancy history, information surrounding the labor and delivery, and the neonatal history if the infant is live born. These medical records are important and necessary to complete the consultation.

If you do not choose to consent to release the medical records listed below, the fetal examination cannot be performed at the Greenwood Genetic Center, but can be performed at the referring hospital.

Mother's name_____
First Middle Last
DOB _____ SSN_____

Baby's name (if applicable) _____
First Middle Last
Date of Delivery _____ Date of Death _____

This release is for pregnancy and delivery and neonatal records pertaining to the delivery date noted above. These records include:

1. Prenatal records including flow sheets and progress notes

Doctor's name or practice_____
City State

2. Ultrasound records, reports

Doctor's name or hospital_____
City State

3. Delivery records (includes Admission history and physical, laboratory studies, consultation reports, ultrasound reports, Labor and Delivery Summary, operative note, Discharge Summary)

4. Neonatal records (if applicable). This includes Admission history and physical, laboratory studies, radiology reports, consultation reports, and Death Summary.

Purpose of release: **Genetic consultation including fetal examination**

Records to be released to: Greenwood Genetic Center
Fetal Examination Service/Center for Anatomic Studies
101 Gregor Mendel Circle
Greenwood, SC 29646

This authorization may not be revoked once the examination is completed. The Greenwood Genetic Center does not redisclose medical records received from other physicians or institutions. If I have questions regarding the disclosure of medical records or the use and disclosure of my protected health information, I may contact the Greenwood Genetic Center Privacy Officer (864-941-8100 or 1-888-GGC-GENE).

Signature (Mother) Printed name (Mother) Date

Signature (Witness) Printed name (Witness) Date

Please use ball point pen and write firmly. WHITE COPY-to accompany specimen. YELLOW COPY-original record.

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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.)

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Courier Services

Please contact the Greenwood Genetic Center Laboratory at 1-800-473-9411 for information regarding courier services.

Please note the following...

1. **The specimen must be available for immediate transfer to courier—they will not wait. Please have the specimen ready and available when the courier arrives.**
2. **The specimen must be placed into a Styrofoam leak-proof container with cool packs.**
3. Some hospitals use their own courier service.
4. 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.
5. **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

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

LAB USE ONLY

Patient Information (Please Print):

Last Name First MI				Address
Race ☐ B ☐ W ☐ Other:		Sex ☐ M ☐ F	DOB MM / DD / YYYY	City, State, Zip
Specimen Collection Date MM / DD / YYYY	Type of specimen Blood/Maternal	ICD9 Code	**Numeric Identifier (MR # or SS #)**	Home telephone

Referring Physician:

Name Fetal Exam / Roger E. Stevenson		Address	
Institution		City, State, Zip	
NPI#		Telephone	Fax
Email Address:		Preferred Method to Receive Results: ☐ Secure Email ☐ Fax ☐ 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 Fetal Exam Case		Telephone	Fax
Address		City, State, Zip	
MasterCard # Visa # (circle one)	Exp. Date	Signature	Auth/Precert #

Indication For Study:

☐ Unknown mutation(s)

Please list clinical features_____

_____

☐ Family History _____

☐ Known mutation(s) _____

☐ Population Screening/ Other _____

Is the patient currently pregnant?

☐ Yes ☐ No

If Yes, LMP date: _____ OR EDC:_____

Pedigree

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.

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LAB USE ONLY

Last Name	First	MI	DOB	SS#

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-Forssman-Lehmann Syndrome (*PHF6*) Sequencing**
- ☐ **Borjeson-Forssman-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

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Website: www.ggc.org Highlighted boxes are required

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Last Name First MI	DOB	SS#

- ☐ **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 ♦**
- ☐ **Mucolipidosis II & III Alpha/Beta *(GNPTAB)* Sequencing**
- ☐ **Mucolipidosis II & III Alpha/Beta *(GNPTAB)* Del/Dup ♦**
- ☐ **Mucolipidosis III Gamma (*GNPTG*) Sequencing**
- ☐ **Mucolipidosis 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 *(FOXG1)* Sequencing
  - ☐ Congenital Rett variant *(FOXG1)* 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

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

LAB USE ONLY

Patient Information (Please Print):

Last Name First MI			Address
Race ☐ B ☐ W ☐ Other:	Sex ☐ M ☐ F	DOB MM / DD / YYYY	City, State, Zip
Specimen Collection Date MM / DD / YYYY	Type of specimen: Blood/Paternal	ICD9 Code	**Numeric Identifier (MR # or SS #)**	Home telephone

Referring Physician:

Name: Fetal Exam / Roger E. Stevenson		Address
Institution		City, State, Zip
NPI#	Telephone	Fax
Email Address:	Preferred Method to Receive Results: ☐ Secure Email ☐ Fax ☐ 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: Fetal Exam Case		Telephone	Fax
Address		City, State, Zip	
MasterCard # Visa # (circle one)	Exp. Date	Signature	Auth/Precert #

Indication For Study:

☐ Unknown mutation(s)

Please list clinical features_____

_____

☐ Family History _____

☐ Known mutation(s) _____

☐ Population Screening/ Other _____

Is the patient currently pregnant?

☐ Yes ☐ No

If Yes, LMP date: _____ OR EDC:_____

Pedigree

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.

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LAB USE ONLY

Last Name	First	MI	DOB	SS#

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-Forssman-Lehmann Syndrome (*PHF6*) Sequencing**
- ☐ **Borjeson-Forssman-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#

- ☐ **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 ♦**
- ☐ **Mucolipidosis II & III Alpha/Beta *(GNPTAB)* Sequencing**
- ☐ **Mucolipidosis II & III Alpha/Beta *(GNPTAB)* Del/Dup ♦**
- ☐ **Mucolipidosis III Gamma (*GNPTG*) Sequencing**
- ☐ **Mucolipidosis 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 *(FOXG1)* Sequencing
  - ☐ Congenital Rett variant *(FOXG1)* 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

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Connective Tissue Disorders (31 genes)
Epilepsy/Seizures (103 genes)
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