



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	City, State, Zip	
Specimen Collection Date	Type of specimen	ICD9 Code	Medical record #	Home telephone	SS #	

Referring Physician:

Name		Address	
Institution		City, State, Zip	
NPI#	Telephone	Fax	

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"/> Symptomatic _____ <input type="checkbox"/> Family History _____ <input type="checkbox"/> Unknown mutation(s) <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 so, provide LMP date _____	Pedigree
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Comments:

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



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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*)
- Aarskog syndrome (*FGD1*)
- Aminoglycoside-induced hearing loss
- Allan Herndon Dudley syndrome (*MCT8*)
- **Angelman syndrome** (check one)
 - Methylation analysis
 - UBE3A
 - Rett/Angelman syndrome 2nd tier panel
(Includes *UBE3A*, *CDKL5*, *FOXG1*, *TCF4* and *SLC9A6*)
- ARX-related spectrum (ARX)**
- ATRX syndrome (XNP) ***
- Biotinidase deficiency (BTD)**
- **Cardio-Facio-Cutaneous (CFC) syndrome** (check one)
 - Tier 1 (*BRAF*)
 - Tier 2 (*MAP2K1* and *MAP2K2*)
 - Tier 3 (*KRAS*)
 - Full CFC syndrome panel (Tiers 1, 2 and 3)
- CDKL5 - Atypical Rett syndrome (STK9)**
- CHARGE (CHD7)**
- Christianson syndrome /X-linked Angelman syndrome (SLC9A6)**
- Coffin-Lowry syndrome (RSK2) ***
- Congenital Disorders of Glycosylation type 1a (PMM2)**
- Congenital Disorders of Glycosylation type 1b (MPI)**
- Congenital Disorders of Glycosylation type 1c (ALG6)**
- Connexin 26 (GJB2)**
- **Costello syndrome** (check one)
 - Tier 1 (*HRAS*, first coding exon)
 - Tier 2 (*HRAS*, remaining exons)
 - Full Costello syndrome panel (Tiers 1 and 2)
- Creatine Transporter Deficiency syndrome (SLC6A8) ***
- Cystic Fibrosis (CFTR) includes ACMG/ACOG panel**
- DMD/BMD deletion/duplication detection**
- DNA Banking**
- **FGFR2-related disorders** (check one)
 - Apert syndrome
 - Beare-Stevenson with cutis gyrate
 - Crouzon syndrome
 - Jackson-Weiss syndrome
 - Pfeiffer syndrome
 - Other _____
- **FGFR3-related disorders** (check one)
 - Achondroplasia
 - Hypochondroplasia
 - Non-syndromic craniosynostosis
 - Thanatophoric dysplasia type I
 - Thanatophoric dysplasia type II
 - Other _____
- Fragile X syndrome (FMR1)**
- FRAXE syndrome (FMR2)**
- Galactosemia, Classic (GALT)**
- Glutaric acidemia, type 1 (GCDH)**
- Hemochromatosis (HFE)**
- Hunter syndrome (IDS)**
- **Leopard syndrome** (check one)
 - Tier 1 (*PTPN11*)
 - Tier 2 (*RAF1* exons 7, 14 and 17)
 - Full Leopard syndrome panel (Tiers 1 and 2)
- Maternal Cell Contamination**
- MCAD (ACADM 985 G>A mutation only)**
- **MED 12 related disorders**
 - Circle one : Lujan-Fryns syndrome FG syndrome**
 - Tier 1 (sequence exons 4,5,20,21,22,28,36)
 - Tier 2 (full RNA sequencing) *
- Mucopolipidosis II/IIIA (GNPTAB)**
- Myotonic dystrophy (DM1)**
- Non-syndromic X-linked Mental Retardation Panel**
- **Noonan syndrome** (check one)
 - Tier 1 (*PTPN11*)
 - Tier 2 (*SOS1*)
 - Tier 3 (*RAF1* and *KRAS*)
 - Full Noonan syndrome panel (Tiers 1, 2 and 3)
- **P63-related disorders** (check one)
 - EEC syndrome
 - Isolated slit-hand/foot malformation
 - Hay-Wells syndrome
 - Other _____
- Phenylketonuria (PAH)**
- Pitt-Hopkins syndrome (TCF4)**
- Prader Willi syndrome, Methylation analysis**
- **PTEN related disorders** (check one)
 - Autism with macrocephaly
 - Bannayan-Riley-Ruvalcaba syndrome
 - Cowden syndrome
 - Proteus syndrome
 - Proteus-like syndrome
- **Rett syndrome (MECP2)** (check one)
 - Full sequencing
 - Deletion/duplication detection (MLPA)
 - Atypical Rett syndrome - *CDKL5 (STK9)*
 - Congenital Rett variant (*FOXG1*)
 - Rett/Angelman syndrome 2nd tier panel
(Includes *UBE3A*, *CDKL5*, *FOXG1*, *TCF4* and *SLC9A6*)
- Saethre-Chotzen (TWIST)**
- Sanfilippo A (SGSH)**
- Sanfilippo B (NAGLU)**
- Sotos syndrome NSD1 Full sequencing**
- Sotos syndrome NSD1 deletion/duplication (MLPA)**
- Thrombosis Panel**
 - Factor V Leiden
 - Prothrombin 20210A
 - MTHFR C677T and A1298C
- **Uniparental Disomy-parental samples required** -check one
 - Chromosome 7 (Russell Silver syndrome)
 - Chromosome 14
 - Chromosome 15
- VLCAD deficiency (ACADVL)**
- X-inactivation analysis**
- X-linked Hydrocephalus (L1CAM)**

All assays require a purple top (EDTA) tube unless otherwise specified

*Requires Qiagen PAXGENE tube (available upon request) and purple top EDTA tube