# 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 ☐ B ☐ W ☐ Other: | | Sex ☐ M ☐ 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 | |
| | | | |
| Select Credit Card Type<br>Credit Card # | Exp. Date | Signature | Auth/Precert # |
| | | | |

**Indication For Study:**

☐ Symptomatic _____
☐ Family History _____
☐ Unknown mutation(s)
☐ Known mutation(s) _____
☐ Population Screening/ Other _____

Is the patient currently pregnant?

☐ Yes ☐ No If so, provide LMP date _____

Pedigree

Comments:

_____

_____

_____

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

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| 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)***
- ☐ **Aarskog syndrome *(FGD1)***
- ☐ ***ACSL4*-related X-linked intellectual disability**
- ☐ **Aminoglycoside-induced hearing loss**
- ☐ **Allan Herndon Dudley syndrome *(MCT8)***
- ☐ **Alpha-Mannosidosis (*MAN2B1)***
- ☐ **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*)**
- ☐ **Borjeson-Forssman-Lehmann Syndrome (*PHF6)***
- ☐ **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)
- ☐ ***CASK*-related X-linked intellectual disability**
- ☐ ***CDKL5* - Atypical Rett syndrome *(STK9)***
- ☐ ***CHD7*-related disorders**
  **Circle one: CHARGE Kallmann syndrome 5**
- ☐ **Christianson syndrome /X-linked Angelman syndrome (*SLC9A6*)**
- ☐ **Citrullinemia, Type 1 (*ASS1*)**
- ☐ **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
  - ☐ Thanantophoric dysplasia type II
  - ☐ Other _____
- ☐ **Fragile X syndrome *(FMR1)***
- ☐ **FRAXE syndrome *(FMR2)***
- ☐ **Galactosemia, Classic *(GALT)***
- ☐ **Glutaric acidemia, type 1 *(GCDH)***
- ☐ **Hemochromatosis *(HFE)***
- ☐ **Hunter syndrome *(IDS)***
- ☐ **Hurler Syndrome (*IDUA*)**
- ☐ **Kabuki syndrome (*MLL2*)**
- ☐ **Leopard syndrome** (check one)
  - ☐ Tier 1 (*PTPN11*)
  - ☐ Tier 2 (*RAF1* exons 7, 14 and 17)
  - ☐ Tier 3 (*BRAF*)
  - ☐ Full Leopard syndrome panel (Tiers 1, 2, and 3)
- ☐ **Marfan syndrome (*FBN1*)**
- ☐ **Maroteaux-Lamy Syndrome (*ARSB*)**
- ☐ **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)*
- ☐ **Mucolipidosis II/IIIA *(GNPTAB)***
- ☐ **Myotonic dystrophy *(DM1)***
- ☐ **Noonan syndrome** (check one)
  - ☐ Tier 1 (*PTPN11*)
  - ☐ Tier 2 (*SOS1*)
  - ☐ Tier 3 (*RAF1 & KRAS* and *SHOC2* - S2G mutation only)
  - ☐ Tier 4 (*BRAF* and *MAP2K1* sequencing)
  - ☐ Full Noonan syndrome panel (Tiers 1, 2, 3, and 4)
- ☐ ***P63*-related disorders** (check one)
  - ☐ EEC syndrome
  - ☐ Isolated slit-hand/foot malformation
  - ☐ Hay-Wells syndrome
  - ☐ Other _____
- ☐ **Phenylketonuria *(PAH)***
- ☐ **Pitt-Hopkins syndrome *(TCF4)***
- ☐ ***POLG1*-related disorders**
- ☐ **Prader Willi syndrome, Methylation analysis**
- ☐ ***PTEN* related disorders** (check one) **full sequencing**
  - ☐ Autism with macrocephaly
  - ☐ Bannayan-Riley-Ruvalcaba syndrome
  - ☐ Cowden syndrome
  - ☐ Proteus-like syndrome
- ☐ ***PTEN* deletion/duplication (MLPA)**
- ☐ **Rett syndrome** (check one)
  - ☐ *MECP2* Full sequencing
  - ☐ *MECP2* 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)* full sequencing**
- ☐ **Saethre-Chotzen *(TWIST)* deletion/duplication (MLPA)**
- ☐ **Sanfilippo A *(SGSH)***
- ☐ **Sanfilippo B *(NAGLU)***
- ☐ **Sanfilippo C (*HGSNAT*)**
- ☐ **Sanfilippo D (*GNS*)**
- ☐ **Sotos syndrome *NSD1* Full sequencing**
- ☐ **Sotos syndrome *NSD1* deletion/duplicat ion (MLPA)**
- ☐ **Thrombosis Panel**
  - ☐ Factor V Leiden
  - ☐ Prothrombin 20210A
- **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)***
- ☐ **Other:**_____

>> **>> Please note that the XLID 92 Gene Panel requires a separate requisition form that is available on our website http://www.ggc.org/images/TestPDFs/xlid-request-form.pdf**

**All assays require a purple top (EDTA) tube unless otherwise specified**
***Requires Qiagen PAXGENE tube (available upon request) and purple top (EDTA) tube**