

## MOLECULAR GENETICS REQUISITION

Highlighted fields are required

### PATIENT INFORMATION

Patient Name: (Last, First) \_\_\_\_\_  
Date of Birth: MM \_\_\_\_\_ /DD \_\_\_\_\_ /YY \_\_\_\_\_  
Gender:  Male  Female  Unknown  
Ethnic Background (Select all that apply):  
 African American  Hispanic  
 Asian  Native American Indian  
 Ashkenazi Jewish  Other Jewish  
 European Caucasian  Other (please specify): \_\_\_\_\_

### REFERRING PHYSICIAN

Physician: \_\_\_\_\_  
Phone: \_\_\_\_\_  
Fax: \_\_\_\_\_  
Email: \_\_\_\_\_

### ADDITIONAL REPORT RECIPIENTS

Physician: \_\_\_\_\_  
Phone: \_\_\_\_\_  
Fax: \_\_\_\_\_  
Email: \_\_\_\_\_

### SAMPLE INFORMATION

Date of Collection: MM \_\_\_\_\_ /DD \_\_\_\_\_ /YY \_\_\_\_\_  
Hospital: \_\_\_\_\_  
Accession #: \_\_\_\_\_  
Sample Type: (Please select one)  
 Blood  Bone Marrow  Other: \_\_\_\_\_  
 Cord Blood  Amniotic Fluid  
 Tissue (specify source): \_\_\_\_\_  
 DNA (specify source): \_\_\_\_\_  
(DNA concentration): \_\_\_\_\_ ug/ul  
 Tumor section: \_\_\_\_\_

### INDICATION FOR STUDY

Autism spectrum disorder  Cognitive impairment  
 Developmental delay  Dysmorphic features  
 Failure to thrive  Short stature  
 Family history of cognitive impairment  
 Suspected thrombophilia  
 Carrier screening for \_\_\_\_\_  
 Congenital malformation (specify) \_\_\_\_\_  
 Other \_\_\_\_\_

### TARGETED ANALYSIS

(please provide proband's test results)

Gene Name: \_\_\_\_\_ Mutation: \_\_\_\_\_  
Name of Proband: \_\_\_\_\_ GC Lab #: \_\_\_\_\_

- Alport Syndrome (*COL4A3, COL4A4, COL4A5*)
- Angelman Syndrome Methylation Analysis
- Angelman Syndrome Methylation w/ reflex to FISH for 15q11 microdeletion  
Array CGH (Microarray):
  - 180K Oligonucleotide/SNP Array
  - 60K Oligonucleotide/SNP Array
  - Prenatal Targeted Oligo/SNP Array
  - Targeted Parental Array (please include child's results)
  - Chromosome Analysis with reflex to Array CGH
- Ataxia Telangiectasia (*ATM*)
- Autosomal Dominant Polycystic Kidney Disease (*PKD1, PKD2*)
- Bannayan-Riley-Ruvalcaba Syndrome (*PTEN*)
- Birt-Hogg-Dube Syndrome (*FLCN*)
- CFTR-related Disorders:**
  - Cystic Fibrosis Targeted Mutation Panel
  - Cystic Fibrosis *CFTR* gene sequence analysis
  - Cystic Fibrosis *CFTR* gene deletion & duplication analysis

- Congenital Central Hypoventilation Syndrome (*PHOX2B*)
- Costello Syndrome (*HRAS*)
- Craniosynostosis Syndromes:**
  - Craniosynostosis Panel (*FGFR1, FGFR2, FGFR3, TWIST1*)
  - Apert Syndrome
  - Crouzon Syndrome
  - Crouzon Syndrome with Acanthosis Nigricans
  - Muenke Syndrome
  - Non-Syndromic Craniosynostosis
  - Pfeiffer Syndrome
  - Saethre-Chotzen Syndrome
- Denys-Drash Syndrome (*WT1*)
- DNA Extraction

Last: \_\_\_\_\_ First: \_\_\_\_\_ DOB: \_\_\_\_\_

## MOLECULAR GENETICS REQUISITION (page 2)

### Duchenne Muscular Dystrophy:

- DMD Deletion/duplication analysis
- DMD Sequencing
- Early On-Set Familial Alzheimer Disease Panel (*APP, PSEN1, PSEN2*)
- Familial Adenomatous Polyposis (FAP) -related disorder (*APC*)
- Familial Mediterranean Fever (*MEFV*)
- Fragile X DNA Analysis

### Hearing Loss:

- Hearing Loss Panel (Connexin 26 and 30, *mt-RNR-1* and *mt-TS1*)
- Connexin 26 and 30 Targeted Mutation Analysis
- Connexin 26 Targeted Mutation Analysis
- Connexin 26 - *GJB2* Full Gene Sequence Analysis
- Connexin 30 Targeted Mutation Analysis
- Mitochondrial DNA Hearing Loss Panel (*mt-RNR1, mtTS1*)
- HFE-related Hemochromatosis
- Hereditary Hemorrhagic Telangiectasia (*ACVRL1, ENG, GDF2, RASAI, SMAD4*)
- Hereditary Mismatch Repair Deficiency Syndrome (*MLH1, MSH2, PMS2, MSH6*)
- Huntington Disease (*HTT*)
- JAK2 V617F Mutation Analysis
- Legius/NF1-like Syndrome (*SPRED1*)
- Li-Fraumeni Syndrome (*TP53*)
- Marfan Syndrome (*FBN1*)
- MECP2 Sequencing (Rett Syndrome)
- Multiple Endocrine Neoplasia, Type 1 (*MEN1*)
- Multiple Endocrine Neoplasia, Types 2A & 2B (*RET*)
- Myotonic Dystrophy DNA Analysis
- Neurofibromatosis, Type 1 (*NF1*)
- Neurofibromatosis, Type 2 (*NF2*)
- Nevoid Basal Cell Carcinoma Syndrome (Gorlin Syndrome) (*PTCH1, SUFU*)
- Noonan Panel (*BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1, SOS2*)
- Osteogenesis Imperfecta (*COL1A1, COL1A2*)
- Pallister-Hall Syndrome (*GLI3*)
- Pancreatitis Panel (*CASR, CFTR, CPA1, CTSC, PRSS1, SPINK1*)
- Paternity/Identity Testing
- Peutz-Jeghers Syndrome (*STK11*)

- Prader-Willi Methylation Analysis
- Prader-Willi Methylation Analysis with reflex to FISH for 15q11 microdeletion
- PTEN-related disorder (*PTEN*)
- RASA1-related disorder (*RASA1*)
- Rasopathy Panel (*BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASAI, RIT1, SHOC2, SOS1, SOS2, SPRED1*)
- Retinoblastoma (*RBI*) (peripheral blood only)
- Schwannomatosis (*SMARCB1*)
- Simpson-Golabi-Behmel Syndrome (*GPC3*)

### Skeletal Dysplasias:

- Achondroplasia
  - Hypochondroplasia
  - Achondroplasia/Hypochondroplasia Panel
  - Thanatophoric Dysplasia (types I and II)
  - Sotos Syndrome (*NSD1*)
- ### Spinal Muscular Atrophy (SMA):
- Carrier Testing (*SMN1* with intron 7 c.\*3+80T>G SNP)
  - Diagnostic Testing (*SMN1* & *SMN2* with intron 7 c.\*3+80T>G SNP)

- Stickler Syndrome Panel (*COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3*)

### Thrombophilia/Obstetric Complication Panel:

- Thrombophilia Panel (Factor II, Factor V, and MTHFR)
- Prothrombin (Factor II) Mutation Analysis
- Factor V Leiden Mutation Analysis
- MTHFR Mutation Analysis (MTHFR A1298C)
- MTHFR Mutation Analysis (MTHFR C677T)
- TPMT Mutation Analysis
- Tuberous Sclerosis (*TSC1* & *TSC2*)
- Twin Zygosity
- Von Hippel-Lindau (*VHL*)
- Wilms Tumor (*WT1*)
- Other (please specify):