

Highlighted fields are required

MOLECULAR GENETICS REQUISITION

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

Gene Name: _____ Mutation: _____
Name of Proband: _____ GC Lab #: _____

- Angelman Syndrome Methylation Analysis**
- Angelman Syndrome Methylation w/ reflex to FISH for 15q11 microdeletion**

Array CGH (Microarray):

- 180K Oligonucleotide/SNP Array
- Prenatal Targeted Oligo/SNP Array
- Targeted Parental Array (please include child's results)
- Chromosome Analysis with reflex to Array CGH
- Ashkenazi Jewish Mutation Panel: (Canavan, Gaucher, Cystic Fibrosis, Tay-Sachs, Familial Dysautonomia)**

Ataxia Telangiectasia (ATM)

- ATM Comprehensive (Seq & Del/Dup Analysis)
- ATM Sequence Analysis
- ATM Deletion/Duplication Analysis

Bannayan-Riley-Ruvalcaba Syndrome (PTEN)

- PTEN Comprehensive (Seq & Del/Dup Analysis)
- PTEN Sequence Analysis
- PTEN Deletion/Duplication Analysis

Bloom Syndrome (BLM)

- BLM Comprehensive (Seq & Del/Dup Analysis)
- BLM Sequence Analysis
- BLM Deletion/Duplication Analysis

Canavan Disease

CFTR-related Disorders:

- Cystic Fibrosis Targeted Mutation Panel
- Cystic Fibrosis CFTR gene sequence analysis
- Cystic Fibrosis Targeted with reflex to CFTR full gene sequencing
- Cystic Fibrosis CFTR gene deletion & duplication analysis

Congenital Central Hypoventilation Syndrome (PHOX2B)

- PHOX2B Comprehensive (Seq & Del/Dup Analysis)
- PHOX2B Sequence Analysis
- PHOX2B Deletion/Duplication Analysis

Last: _____ First: _____ DOB: _____

MOLECULAR GENETICS REQUISITION (page 2)

Costello Syndrome (*HRAS*)

- HRAS* Comprehensive (Seq & Del/Dup Analysis)
- HRAS* Sequence Analysis
- HRAS* Deletion/Duplication Analysis

Craniosynostosis Syndromes:

- Craniosynostosis Comprehensive Panel (*FGFR1*, *FGFR2*, *FGFR3*, *TWIST*)
- Apert Syndrome
- Crouzon Syndrome
- Crouzon Syndrome Acanthosis Nigricans
- FGFR1* gene sequencing
- FGFR2* gene sequencing
- FGFR3* gene sequencing
- Muenke Syndrome
- Non-Syndromic Craniosynostosis
- Pfeiffer Syndrome
- Saethre-Chotzen Syndrome

Denys-Drash Syndrome (*WT1*)

- WT1* Comprehensive (Seq & Del/Dup Analysis)
- WT1* Sequence Analysis
- WT1* Deletion/Duplication Analysis

DNA Extraction

Duchenne Muscular Dystrophy

- DMD Deletion analysis
- DMD Deletion analysis with reflex to duplication analysis
- DMD Deletion & duplication analysis
- DMD Gene sequence analysis

Exome Sequencing (please use our separate Exome Sequencing Requisition Form)

Familial Dysautonomia

Fragile X DNA Analysis

Gaucher Disease

Hearing Loss:

- Comprehensive Hearing Loss Panel (Connexin 26 and 30, *mt-RNR1* 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*, *mt-TS1*)

Hereditary Hemochromatosis

Huntington Disease

JAK2 V617F Mutation Analysis

MECP2 gene sequencing (Rett syndrome)

Mitochondrial Disorders

- Mitochondrial DNA Targeted Mutation Analysis
- Mitochondrial Whole Genome Sequencing
- Mitochondrial DNA Hearing Loss Panel (*mt-RNR1*, *mt-TS1*)

Myotonic Dystrophy DNA Analysis

Osteogenesis Imperfecta (*COL1A1*, *COL1A2*)

Paternity/Identity Testing

Prader-Willi Methylation Analysis

Prader-Willi Methylation Analysis with reflex to FISH for 15q11 microdeletion

Rett Syndrome

- Rett Comprehensive Panel (MECP2, CDKL5, FOXG1)
- MECP2 gene sequencing
- CDKL5 gene sequencing
- FOXG1 gene sequencing

Skeletal Dysplasias:

- Achondroplasia
- Hypochondroplasia
- Comprehensive Achondroplasia/Hypochondroplasia Panel
- Thanatophoric Dysplasia (types I and II)

Shwachman-Diamond Syndrome (*SBDS*)

- SBDS* Comprehensive (Seq & Del/Dup Analysis)
- SBDS* Sequence Analysis
- SBDS* Deletion/Duplication Analysis

Simpson-Golabi-Behmel Syndrome (*GPC3*)

- GPC3* Comprehensive (Seq & Del/Dup Analysis)
- GPC3* Sequence Analysis
- GPC3* Deletion/Duplication Analysis

Sotos Syndrome (*NSD1*)

- NSD1* Comprehensive (Seq & Del/Dup Analysis)
- NSD1* Sequence Analysis
- NSD1* Deletion/Duplication Analysis

Spinal Muscular Atrophy (SMA):

- Carrier Testing
- Diagnostic Testing

Tay-Sachs Disease

Thrombophilia/Obstetric Complication Panel:

- Comprehensive Thrombophilia Panel (Factor II, Factor V and MTHFR)
- Prothrombin (Factor II) Mutation Analysis
- Factor V Leiden Mutation Analysis
- MTHFR Mutation Analysis

Tuberous Sclerosis (*TSC1* & *TSC2*)

- TSC1* & *TSC2* Comprehensive (Seq & Del/Dup Analysis)

Tumor Molecular Testing (please provide H&E slide):

- BRAF Mutation Analysis
- KRAS Mutation Analysis
- Combined BRAF and KRAS Mutation Analysis
- Colorectal Cancer Mutation Screening

Twin Zygosity

- Other (please specify): _____