

PATIENT INFORMATION
Name: _____
Last Name, First Name
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: _____

ADDITIONAL PROFESSIONAL REPORT RECIPIENTS:

Physician: _____

Phone: _____

Fax: _____

SAMPLE INFORMATION:
Date of Collection: ____/____/____

Hospital: _____

Accession #: _____

Sample Type (Please select one):

-
- Blood
-
-
- Bone Marrow
-
-
- Cord Blood
-
-
- Amniotic Fluid
-
-
- Tissue (specify source): _____
-
-
- DNA (specify source): _____
-
- (DNA concentration): _____ ug/ul
-
-
- Other (specify): _____

INDICATION FOR STUDY:

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

Targeted Analysis: Gene Name: _____
Name of Proband: _____

Mutation: _____
GC Lab # _____

-
- Angelman Syndrome Methylation Analysis**
-
-
- Ashkenazi Mutation Panel:**
- (Canavan, Gaucher, Cystic Fibrosis, Tay-Sachs, Familial Dysautonomia)
-
-
- BCR/ABL Diagnostic Testing**
-
-
- BCR/ABL Quantitative Testing**
- (specify
-
- Major or
-
- Minor)
-
-
- Canavan Disease**
-
- CFTR-related Disorders:**
-
-
- Cystic Fibrosis Targeted Mutation Panel
-
-
- Cystic Fibrosis
- CFTR*
- sequence analysis
-
- Craniosynostosis Syndromes:**
-
-
- Craniosynostosis Comprehensive Panel
-
-
- Apert Syndrome
-
-
- Crouzon Syndrome
-
-
- Crouzon Syndrome Acanthosis Nigricans
-
-
- FGFR1*
-
-
- FGFR2*
-
-
- FGFR3*
-
-
- Muenke Syndrome
-
-
- Non-Syndromic Craniosynostosis
-
-
- Pfeiffer Syndrome
-
-
- Saethre-Chotzen Syndrome
-
-
- Duchenne Muscular Dystrophy**
-
-
- 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*
- Sequence Analysis
-
-
- Connexin 30 Targeted Mutation Analysis
-
-
- mtDNA Hearing Loss Panel (
- mt-RNR1*
- ,
- mt-TS1*
-)
-
-
- Hemochromatosis**
-
-
- JAK2 V617F Mutation Analysis**
-
- Microarray (Array CGH):**
-
-
- 180K Oligonucleotide Array
-
-
- Prenatal Targeted Array
-
-
- Targeted Parental Array (please include child's results)
-
-
- Mitochondrial DNA Targeted Mutation Analysis**
-
-
- Myotonic Dystrophy DNA Analysis**
-
-
- Paternity/Identity Testing**
-
-
- Prader-Willi Methylation Analysis**
-
- Thrombophilia / Obstetric Complication Panel:**
-
-
- Comprehensive Thrombophilia Panel (Factor II, Factor V and MTHFR)
-
-
- Prothrombin (Factor II)
-
-
- Factor V Leiden
-
-
- MTHFR
-
- Spinal Muscular Atrophy:**
-
-
- Carrier Testing
-
-
- Diagnostic Testing
-
- Skeletal Dysplasias**
-
-
- Achondroplasia
-
-
- Hypochondroplasia
-
-
- Comprehensive Achondroplasia / Hypochondroplasia Panel
-
-
- Thanatophoric Dysplasia (types I and II)
-
-
- Tay-Sachs Disease**
-
-
- Other**
- (please specify): _____