CENTER FOR HUMAN GENETICS LABORATORY

University Hospitals Laboratory Services Foundation
W.O. Walker Center, 6th Floor
10524 Euclid Avenue
Cleveland, OH 44106 Tel: (216) 983-1134 Fax: (216) 983-1144
www.chglab.org

University Hospitals

Prenatal and Postnatal Cytogenetic and Molecular Specimens

PATIENT INFORMATION (Label)
Name (Last, First)_____ Phone (H) (___)_____ DOB ____/____/_____
Address_____ (W) (___)_____ SS# ____-____-_____
City/State/Zip_____ **Sex: ☐ Male ☐ Female**

Referring Institution_____ **MRN**_____

Sample: ☐ Peripheral Blood ☐ Cord Blood / Liveborn ☐ CVS ☐ Amniotic Fluid _____cc's Gest Age_____
☐ DNA ☐ Products of Conception (specify)_____ **☐ Tissue (specify)**_____ **☐ Other (specify)**_____

Date/Time of specimen collection:_____ Person Collecting_____ Location (institution): _____

Ethnicity: ☐ Caucasian ☐ African American ☐ Ashkenazi Jewish ☐ Hispanic ☐ Asian ☐ Native American
☐ Other _____

REFERRING PHYSICIAN/PROVIDER
Name_____ Signature_____
Phone:_____ Fax:_____ Pager:_____

BILLING INFORMATION Results also sent to_____
Bill: ☐ Referring Institution ☐ Insurance
(Attach Full Patient Demographics and Copy of Insurance card; Medicare - most genetic tests require current date signed ABN)

INDICATIONS FOR TESTING / ICD-code: (Required) **☐ Symptomatic ☐ Non-symptomatic ☐ Patient/couple pregnant**

TEST REQUESTED

Prenatal Cytogenetic and Molecular Tests

___ **PRENATAL CHROMOSOME ANALYSIS/KARYOTYPE**
(Amniotic Fluid AFP done automatically unless otherwise specified)
___ **AneuVysion FISH (prenatal screen for X, Y, 13, 18, 21)**
___ **Save Cells for future testing - extra charges apply**
Reason _____
Additional tests on Amnio/CVS
(check all that apply:)
☐ CMV ^ ☐ Toxoplasmosis ^ ☐ Parvovirus ^ ☐ Herpes I/II ^
___ **PRENATAL CYTOGENOMIC MICROARRAY (CMA)^ - SNP**
___ **Maternal Cell Contamination Studies (Purple EDTA Tube)**
___ **Prenatal Other** _____

Pregnancy Screening
___ Cystic Fibrosis Carrier Screen (41 Mutations)
___ Ashkenazi Jewish Common Mutation Carrier Screening Panel
(for individual test, see back)

Maternal Serum Screening

(Red or Yellow Gel Separator Tube Required)
Completed Information Required:
Patient Current Weight _____
Insulin-Dependent Diabetic: ____ Yes ____ No
Twin Pregnancy: ____ Yes ___ No ___Unknown
Previous Child with ONTD: ____ Yes ____ No
Family History of ONTD: ____ Yes ____ No
Gestational Age Dating, complete one:
EDC by Ultrasound _____
Last Menstrual Period _____
By Physical Exam: _____ weeks
Date of Exam _____
Test Requested
___ Quad Check (AFP/UE3/hCG/Inhibin A)
___ Triple Check (AFP/UE3/hCG)
___ AFP Only **(order as GAFP1)*
___ Repeat Test at This Laboratory

Commonly Ordered Tests

___ CYTOGENOMIC MICROARRAY (CMA)^ - SNP (NaHep Tube)
___ CHROMOSOME ANALYSIS, HIGH RESOLUTION (NaHep Tube)
(also known as karyotype or cytogenetics)
___ Fragile X Syndrome PCR Analysis (Purple EDTA Tube)
(includes reflex to Methylation status for positive cases)
^ = *send-out tests*

___ Thrombosis Panel (Purple EDTA Tube)
Panel includes the following, or mark off the individual tests
__ Factor V Leiden, R506Q mutation
__ Reflex to Factor V HR2 Haplotype for positive R506Q
__ Prothrombin (Factor II) mutation, G20210A mutation
__ MTHFR, C677T and A1298C mutations

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Postnatal Cytogenetic and Molecular Specimens

Patient Name_____**MRN**_____

Cytogenetic Tests (Requires Dark Green NaHep Tube)

___ **CHROMOSOME ANALYSIS, HIGH RESOLUTION**
(also known as karyotype or cytogenetics)
___ **STAT chromosomes for newborns under 14 days**
- includes 5 cell preliminary result called within 48-72 hours
- (extra charge, for newborns only - contact lab with questions)
___ **Mosaicism Study - extra 10 cells counted**
(automatically performed for Q. 45,X or short stature in a female)
___ **CYTOGENOMIC MICROARRAY (CMA)^ - SNP**
___ - FISH for CMA Confirmation - contact lab for more information
- Other FISH analysis - Contact lab to arrange
___ Parental Cytogenomic Microarray (CMA) sample
___ Parental CMA FISH only
Child's name_____
___ Sibling CMA FISH only: proband name_____

___ **Other Cytogenetic Test**_____
^ = *send-out tests*

Molecular: (Requires Purple EDTA Tube)

___ **Fragile X Syndrome PCR Analysis**
(includes reflex to Methylation for positive cases)
___ **Fragile X Syndrome - Methylation Analysis Only**
(contact lab to arrange)
___ **Prader-Willi Syndrome Methylation Analysis**
___ **Angelman Syndrome Methylation Analysis**
___ **Reflex to FISH for positive PWS/AS specimens**
(needs Dark Green top Na-Hep + Purple Top EDTA Tubes)
___ **Uniparental Disomy (6, 7, 14, 15 available) chr#** _____
___ *- requires sample from at least one parent, prefer both*
___ **Hereditary Hemochromatosis**
___ **SLCO2A1 Targeted Sequencing**
___ **Y Deletion Analysis**
___ **Zygosity Testing;** parental samples should be submitted

___ **DNA Extract and Store: purpose** _____
___ **Other Molecular Test**_____

Molecular: (Requires Purple EDTA Tube)

Ashkenazi Jewish Common Mutation Screening Panel

Mark off the individual tests
__ Bloom Syndrome
__ Canavan Disease
__ Familial Dysautonomia
__ Fanconi Anemia, group C
__ Gaucher Disease
__ Mucolipidosis, type IV
__ Niemann Pick Disease Types A/B
__ Tay Sachs Disease

Cystic Fibrosis

___ Cystic Fibrosis Carrier Screen (41 Mutations)
___ Cystic Fibrosis Gene Sequencing CFTR
___ Cystic Fibrosis - Newborn screening confirmation
Mutation(s) detected_____
___ CFTR Known Familial Mutation Testing
Familial Mutation(s)_____

Old Order Amish

___ MYBPC3 Gene Targeted Mutation Analysis
___ SPG21 Gene Targeted Mutation Analysis
___ PCCB (Propionyl-CoA Carboxylase) Targeted Mutation Analysis

___ Hearing Loss Panel (in order listed)

Performed sequentially in the following order, or individually:
__ Connexin 26 (sequencing)
__ Connexin 30 (deletion testing)
__ Connexin 30 (sequencing)
__ MTRNR1 and MTTS1 (sequencing)
___ Known Familial Mutation Testing, Gene_____
Familial Mutation(s)_____

Prion: PRNP Gene Analysis: Please Call 216-368-0587
All testing MUST be submitted through the
National Prion Disease Pathology Surveillance Center

Metabolic Disorders

___ Fumarate Hydratase (FH) Gene Sequencing
___ Pyruvate Carboxylase (PC) Gene Sequencing
___ Pyruvate Dehydrogenase Complex (PDC) panel
Performed sequentially in the following order, or individually
__ PDHA1 Gene Sequencing
__ PDHB Gene Sequencing
__ PDHX Gene Sequencing
__ DLD Gene Sequencing
__ DLAT Gene Sequencing
___ PDC Next Generation Sequencing panel (23 gene panel)
___ Known Familial Mutation Testing, Gene_____
Familial Mutation(s)_____

___ Thrombosis Panel

Panel includes the following, or mark off the individual tests
__ Factor V Leiden, R506Q mutation
__ Reflex to Factor V HR2 Haplotype for positive R506Q
__ Prothrombin (Factor II) mutation, G20210A mutation
__ MTHFR, C677T and A1298C mutations

Familial Cancer syndromes

___ Fumarate Hydratase (FH) Gene Sequencing
___ Lynch Syndrome Panel
Panel includes the following, or mark off the individual tests
__ MLH1 Sequencing and Deletion/ Duplication
__ MSH2 Sequencing and Deletion/ Duplication
__ MSH6 Sequencing and Deletion/ Duplication
__ PMS2 Sequencing and Deletion/ Duplication
___ Known Familial Mutation Testing, Gene_____
Familial Mutation(s)_____