all children's hospital

JOHNS HOPKINS MEDICINE

GENETICS LABORATORIES TEST REQUEST

CYTOGENETICS
T: 727-767-8559
F: 727-767-8367

MOLECULAR GENETICS
T: 727-767-8985
F: 727-767-8367

BIOCHEMICAL GENETICS
T: 727-767-8689
F: 727-767-8514

Patient (last, first):	Sex: ☐ M ☐ F	Client Name:
Address:	Ethnic origin:	Address:
City/State/Zip:	Pregnant? ☐ Y ☐ N	City/State/Zip:
Home Phone:	Transfused? ☐ Y ☐ N	Phone: Fax:
Date of Birth (MM/DD/YY):	Med. Rec.#	Referring Physician:

Clinical Indications / Family History (attach pedigree, if appropriate): DIAGNOSTIC CODE(S):

CONSTITUTIONAL CHROMOSOME MICROARRAY ANALYSIS (CMA)

BLOOD: 4cc sod hep (green) + 4cc EDTA (lav)

CMA ☐ Microarray **Constitutional**

CONSTITUTIONAL CYTOGENETICS

KARYOTYPE / CHROMOSOME ANALYSIS

BLOOD: 3cc sodium heparin (green)

CS ☐ Routine Chromosomes
CS ☐ High Resolution (specify chromosome)
CS ☐ Neonatal Chromosomes ☐ STAT
CS ☐ Mosaic Chromosome Study
CS ☐ Skin ☐ Autopsy ☐ Other
3x3 mm² in Sterile Media
☐ Chromosomes ☐ Cryopreservation
☐ Send out testing for:_____

ONCOLOGY CHROMOSOME MICROARRAY ANALYSIS (CMAO)

BM/BLOOD: 2cc sod hep (green) + 2cc EDTA (lav)

CMAO ☐ Microarray **Oncology**

ONCOLOGY CYTOGENETICS

KARYOTYPE / CHROMOSOME ANALYSIS

BM/BLOOD: 3cc sod hep (green)

CSO☐ Bone Marrow Chromosomes
CSO☐ Leukemic Blood Chromosomes
CSO☐ Solid Tumor ***3x3 mm² in Sterile Media***
CSO☐ Lymphoma ***3x3 mm² in Sterile Media***

CYTOGENETICS & FISH

KARYOTYPE / CHROMOSOME & FISH ANALYSIS

BLOOD: 3cc sodium heparin (green)

CS ☐ 1p36 deletion Syndrome
CS ☐ Ambiguous genitalia Sex XX / XY
CS ☐ Angelman Syndrome 15q
CS ☐ Cri du Chat 5p
CS ☐ DiGeorge/VCF Syndrome 22q
CS ☐ Kallman Xp
CS ☐ Miller Dieker 17p
CS ☐ Neurofibromatosis (NF1) 17q
CS ☐ Prader-Willi
CS ☐ Smith Magenis 17p
CS ☐ Sotos 5q
CS ☐ SRY Yp Male determination
CS ☐ Steroid Sulfatase Xp
CS ☐ Trisomy 13 ☐ Trisomy 18 ☐ Trisomy 21
CS ☐ Williams 7q
CS ☐ Wolf-Hirschhorn 4p
CS ☐ Other FISH: **(inquire)**

BONE MARROW TRANSPLANTATION MONITORING

FISH: ***BM/BLOOD: 3cc sodium heparin (green)***

CS ☐ Post BMT XX/XY by FISH

STR ANALYSIS

BM/BLOOD: 0.5cc EDTA (lavender)

BMD ☐ Donor Specimen
STRPRE ☐ Pre Transplant Recipient
STRPOST ☐ Post Transplant Recipient

LINEAGE SPECIFIC STR ANALYSIS:

BLOOD: 8.5 cc ACD (yellow), BM: 1cc ACD (yellow)

STRPOST1 ☐ Post-Trans, Lineage-Specific
☐T-cell ☐ B-cell ☐ Myeloid ☐ NK ☐ Lymphoid
☐RBC

MOLECULAR GENETICS

BLOOD: 2cc EDTA (lav) (0.5cc min - infants)
AMNIO / BM / BUCCAL / TISSUE (Inquire)
**** = include parents***

FGFR3 ☐ Achondroplasia/Hypochondroplasia
TBX19 ☐ Adrenal Insufficiency
NR0B1 ☐ Adrenal hypoplasia (aka DAX1)
AR ☐ Androgen Insensitivity Syndrome
UBE3A ☐ Angelman Syndrome (sequencing)
ANG ☐ Angelman Syn (methylation)*
PTEN ☐ Autism/Microcephaly/PHTS
FOXE1 ☐ Bamforth-Lazarus Syndrome
BWS ☐ Beckwith-wiedemann (methylation)
BTK ☐ Bruton's Agammaglobulinemia
PPT1 ☐ Batten Disease,Ceroid Lipofucinosis
TPP1 ☐ Batten Disease,Ceroid Lipofucinosis
CHSP ☐ Cancer Hotspot Panel (50 genes)
CX26 ☐ Deafness, Autosomal Recessive 1A
CX30 ☐ Deafness (del/dup)
DMD ☐ Dytrophinopathies (del/dup)
DBANK ☐ DNA Preparation/Banking
DYT1 ☐ Dystonia (hotspots)
FAC5M3 ☐ Factor V Leiden (R506Q mut only)
FX ☐ Fragile X Syndrome (no FISH)
ACTHR ☐ Glucocorticoid Def (aka MC2R)
RET ☐ Hirschprung Disease (entire)
HUNT ☐ Huntington Disease
HFE ☐ Hemochromatosis (entire)
TACR3 ☐ Hypogonadotropic Hypogonadism
PAX8 ☐ Hypothyroidism, Nongoiterous
LHCGR ☐ Leydig Cell Hypoplasia (aka LHR)
MBL ☐ Mannan Binding Ligand Genotype

MOLECULAR GENETICS (continued)

MCC ☐ Maternal Cell Contamination
SLC19A2 ☐ Megaloblastic anemia, Rogers
RET ☐ MEN IIA, IIB, FMTC (hotspots)
MTM1 ☐ Myotubular Myopathy, X-linked
PPT1 ☐ Neuronal Ceroid Lipofuscinosis
MC4R ☐ Obesity, Inherited
PAR ☐ Paternity / Zygosity*
GHR ☐ Pituitary Dwarfism II, Laron Synd.
LHX3 ☐ Pituitary Dwarfism III (ex 1,2,3)
LHX4 ☐ Pituitary Short Stature
PROP1 ☐ Pituitary Hormone Deficiency
PIT1 ☐ Pituitary Horm Def (aka POU1F1)
PWD ☐ Prader-Willi (methylation)*
PGM ☐ Prothrombin (FII, G20210A mut)
MECP2 ☐ Rett Syndrome (sequencing)
MECP2 ☐ Rett Syndrome (del/dup)
PITX2 ☐ Rieger Syndrome
CYP19A1 ☐ Pseudohermaphroditism
HSD17B3 ☐ Pseudohermaphroditism
HESX1 ☐ Septo-optic dysplasia
SRY ☐ Sex reversal, Gonadal Dys.
SMA ☐ Spinal Muscular Atrophy (del/dup)
SRD5A2 ☐ Steroid 5-alpha Reductase Def.
MTHFR ☐ Thrombosis (C677T, A1298C)
MTHFR ☐ Thromb., Homocystinuria (entire)
THRB ☐ Thyroid Hormone Resistance
UPD ☐ UPD (3,6,7,14,15,18)*
VWF28 ☐ von Willebrand Disease (ex 28)
Other ☐ _____

BIOCHEMICAL GENETICS

BLOOD: 3cc red or green
URINE: 2cc urine (random or 24 hour)
FILTER CARD: 4 spots / CSF: 1cc
Store and Ship All Fluids Refrigerated (Cold Pack)

AAQCSF ☐ Amino Acids, Quant *CSF*
AAQS ☐ Amino Acids, Quant *BLOOD*
AAQU ☐ Amino Acids, Quant *URINE*
AASCSF ☐ Amino Acid Screen *CSF*
AASS ☐ Amino Acid Screen *BLOOD*
AASU ☐ Amino Acid Screen *URINE*
CARNS ☐ Carnitine, Free & Total *BLOOD*
CARNU ☐ Carnitine, Free & Total *URINE*
MPS ☐ Mucopolysaccharide Scn *URINE*
OASU ☐ Organic Acid Screen *URINE*
PKU ☐ PKU *BLOOD or CARD*
PKUTYR ☐ PKU w/ Tyrosine *BLOOD or CARD*

SPECIMEN COLLECTION DATE/TIME:_____ DRAWN BY:_____ FORM COMPLETED BY:_____

CLIA ID: 10D0700790, FLA LIC: L800000135, CAP: 153609-01

Patient ID

UNLESS OTHERWISE SPECIFICED - SHIP AT ROOM TEMP TO:
All Children's Hospital, 601 Fifth Street South, St. Petersburg Fl, 33701
ATTN: Pathology and Laboratory Medicine, Clinical Genetic Laboratories, Dept.7020
PHYSICIAN SIGNATURE:_____ DATE:_____

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