MEDICAL GENETICS CYTOGENETICS REQUISITION

Attending Physician

Requesting Physician

Specimen Type: Blood Skin biopsy POC Placenta Tissue (specify) __________________________

See specimen requirements below - Deliver to HL423 University of Kentucky Hospital

CLINICAL DIAGNOSIS (REQUIRED)

Angelman syndrome Family history of chromosome abnormality* Partner of woman with multiple miscarriages #

Ambiguous genitalia* Fetal demise* Prader-Willi syndrome

Autism spectrum disorder Infertility Sex reversal*

Craniofacial abnormalities* Klinefelter syndrome Short stature

Craniofacial abnormalities* Mental retardation Stillbirth*

Developmental delay Miscarriage Trisomy 13

DiGeorge/Velo-Cardio-Facial syndromes Multiple miscarriages #______ (not pregnant) Trisomy 18

Down syndrome Multiple miscarriages #______ (pregnant) Turner syndrome

Dysmorphism* Multiple congenital anomalies* Other:

*List suspected diagnosis and/or relevant physical findings, medical history and family history:

TESTING REQUESTED (check all that apply)

Chromosome analysis
Peripheral blood in sodium heparin: Neonates: 1-2 mL, Children and adults: 3-5 mL
Skin biopsy, 4 mm x 4 mm punch biopsy (sterile)
POC, placenta, fetal and autopsy specimens – see web site http://www.hosp.uky.edu/clinlab

FISH Testing: select specific test below
FISH testing may be ordered separately or in addition to Chromosome analysis.
FISH Specimen Requirement: Peripheral blood in sodium heparin- All ages: 1-2mL

Angelman syndrome (D15S10)
22q11.2 deletion TUPLE1 (HIRA) (DiGeorge/Velo-Cardio-Facial syndromes)
Prader-Willi syndrome (SNRPN)
X and Y centromere probes for sex determination (DXZ1/DYZ3) (STAT for newborns < 8 days old only)
SRY (sex determining region of Y)

Mitomycin C breakage studies (Fanconi anemia)
A control specimen from a normal, non-related individual is required.
Specimen Requirement: Peripheral blood in sodium heparin- Neonates: 2mL, Children/adults: 3-5mL, Control: 3-5mL

Establish fibroblast cell line for biochemical/DNA testing/Reestablish fibroblast cell line from frozen cells
Skin punch biopsy, 4 mm x 4 mm (sterile)
Appropriate requisition and consent forms for biochemical/DNA testing must accompany specimen.

Freeze fibroblast cells viably after establishing cell line

Reestablish fibroblast cell line from frozen cells and grow