

Screening the Human Genome for Structural and Copy Number Changes in Individuals with Congenital Malformations with or without Developmental Delay

Clinical Checklist

Pedigree #: _____

Mother: _____

Father: _____

PLACE GENETICS STICKER HERE
(Name, DOB, HSC, SEX)

Height: ____ cm (____ %) Weight: ____ kg (____ %) HC: ____ cm (____ %)

Referring Physician/Genetic Counsellor: _____

Open Consent Closed Consent

Consent to Submit Data to DECIPHER Database Yes No

Inclusion Criteria:

*One major malformation (example- cleft lip, renal or cardiac defects) +/- developmental delay

OR

*Three or more minor anomalies (example-epicanthal folds, flat supraorbital ridges) +/- developmental delay

*Individuals with known chromosomal deletions, duplications or translocations may be included.

Exclusion Criteria:

*Non-English speaking without the presence of an interpreter

*Known single gene disorder

Blood Sample:

*If < 5.5 kg 5 cc in ACD tube

*If > 5.5 kg 10 cc in ACD tube

Karyotype Results and location where performed: _____

Molecular Studies and

Results: _____

FEATURES:

Facial Dysmorphism

Ears

- lowset
 - simple
 - posteriorly rotated
 - Other _____
-
-

Eyes

- Thick eyebrows
 - Sparse eyelashes
 - Thick eyelashes
 - Curly eyelashes
 - Hypertelorism
 - Hypotelorism
 - Other _____
- Downslanting PF
 - Upslanting PF
 - Short PF
 - Wide PF
 - Epicanthal folds
-
-

Forehead/Hair

- Frontal bossing
 - High hairline
 - Low hairline
 -
 - Other _____
- Temporal narrowing
 - Hirsutism
-
-

Mouth/Nose

- Cleft lip
 - Cleft palate
 - Thin lips
 - Full lips
 - Long philtrum
 - Short philtrum
 - Other _____
- Depressed nasal bridge
 - Wide nasal bridge
 - Anteverted nares
 - High palate
 - Arched Palate
-
-

Other Dysmorphic Facial Features:

Developmental Delay Mild Moderate Severe Profound Unknown
Specify _____

Respiratory Involvement
Specify _____

Cardiac Involvement
Specify _____

GI/GU Involvement (Include renal)
Specify _____

Hematology/Endocrine
Specify _____

MSK Involvement (Include limbs and spine)
Specify _____

Skin Involvement
Specify _____

Neurological Involvement
Specify _____

Neurological Imaging MRI CT Ultrasound Other
Results _____

Other _____

PLEASE RETURN