

**Bench to Bassinet**  
**Pediatric Cardiac Genomics Consortium: CHD GENES**  
 Form 105: Congenital Extracardiac Findings  
 Version: B - 11/01/2010

**SECTION A: ADMINISTRATIVE INFORMATION**

A1. Study Identification Number:

A2. Study Visit:

A3. Date Form Completed:  MM/DD/YYYY

A4. Exam Date:  MM/DD/YYYY

**SECTION B: SUBJECT HEIGHT AND WEIGHT**

B1. Subject's Height

**Feet/inches   Centimeters   Unknown   Source Pending**

a. Height Unit:

Feet/Inches: b.  ft                      c.  in

d. Centimeters:  cm

e. Date Obtained:  MM/DD/YYYY

B2. Subject's Weight

**pounds/ounces   kilograms   Unknown   Source Pending**

a. Weight Unit:

Pounds/ounces: b.  lbs                      c.  oz

d. Kilograms:  kgs

e. Date Obtained:  MM/DD/YYYY

**SECTION C: CONGENITAL EXTRACARDIAC FINDINGS**

C1. Was there evidence of any abnormal extracardiac findings in the subject's medical record?  **Yes**    **No**    **Source Pending**

C2. Was there evidence of dysmorphic facies in the subject's medical record?  **Yes**    **No**    **Source Pending**

C3. Were any abnormalities of the head noted in the medical record?  **Yes**    **No**    **Source Pending**

a. Craniosynostosis

    i. Specify:

b. Cutis Aplasia

c. Macrocephaly

d. Microcephaly

e. Other

    i. Specify:

C4. Were any abnormalities of the eyes noted in the medical record?  **Yes**    **No**    **Source Pending**

a. Anophthalmia

b. Cataracts

c. Coloboma

d. Congenital Blindness

e. Duane Anomaly

f. Epicanthal Folds

g. Esotropia

h. Nystagmus

- i. Posterior Embryotoxin
- j. Ptosis
- k. Strabismus
- l. Tortous Retinal Vessels
- m. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C5. Were any abnormalities of the ears noted in the medical record?  Yes  No  Source Pending

- a. Absent/ Abnormal Ear Canal
- b. Ear Pit
- c. Ear Tag
- d. Hearing Loss
- e. Low Set
- f. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C6. Were any abnormalities of the nose noted in the medical record?  Yes  No  Source Pending

C7. Were any abnormalities of the neck noted in the medical record?  Yes  No  Source Pending

- a. Branchial Cleft/Cyst
- b. Excess Nuchal Skin
- c. Webbed Neck
- d. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C8. Were any abnormalities of the mouth noted in the medical record?  Yes  No  Source Pending

- a. Macroglossia
- b. Micrognathia
- c. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C9. Were any abnormalities of the palate noted in the medical record?  Yes  No  Source Pending

- a. Bifid Uvula
- b. Cleft Lip
- c. Cleft Palate
- d. Submucosal Cleft Palate
- e. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C10. Were any abnormalities of the airway noted in the medical record?  Yes  No  Source Pending

- a. Choanal Atresia
- b. Choanal Stenosis
- c. Esophageal Atresia
- d. Laryngo-bronchio-tracheomalacia
- e. Tracheoesophageal (TE) Fistula
- f. Subglottic Stenosis
- g. Tracheal Stenosis
- h. Tracheal Web

i. Other

i. Specify:

**Yes** **No** **Source Pending**

C11. Were any abnormalities of the chest noted in the medical record?

- a. Pectus Carinatum
- b. Pectus Excavatum
- c. Supernumerary Nipples
- d. Wide Spaced Nipples
- e. Other

i. Specify:

**Yes** **No** **Source Pending**

C12. Were any pulmonary abnormalities noted in the medical record?

- a. Bilateral Right-sidedness
- b. Bilateral Left-sidedness
- c. Congenital Cystic Adenomatoid Malformation (CCAM)
- d. Diaphragmatic Hernia
- e. Lung Hypoplasia
- f. Other

i. Specify:

**Yes** **No** **Source Pending**

C13. Were any hepatic abnormalities noted in the medical record?

- a. Bile Duct Paucity
- b. Biliary Atresia
- c. Left-sided or Midline Liver
- d. Other

i. Specify:

**Yes** **No** **Source Pending**

C14. Were any abdominal abnormalities noted in the medical record?

- a. Annular Pancreas
- b. Asplenia
- c. Duodenal Atresia
- d. Gastroschisis
- e. Hirshsprung's
- f. Inguinal Hernia
- g. Malrotation
- h. Omphalocele
- i. Polysplenia
- j. Pyloric Stenosis
- k. Umbilical Hernia
- l. Other

i. Specify:

**Yes** **No** **Source Pending**

C15. Were any renal abnormalities noted in the medical record?

- a. Cystic or Polycystic Kidney
- b. Dysplastic Kidney
- c. Horseshoe Kidney

- d. Hydronephrosis
- e. Pelvic Kidney
- f. Single Kidney
- g. Ureteral Abnormalities
- h. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C16. Were any genitourinary abnormalities noted in the medical record?

- a. Ambiguous Genitalia
- b. Anteriorly Placed Anus
- c. Bicornuate Uterus
- d. Bifid Scrotum
- e. Hypospadias
- f. Imperforate/ Atretic Anus
- g. Micropenis
- h. Undescended Testis
- i. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C17. Were any general skeletal abnormalities noted in the medical record?

- a. Cervical Instability
- b. Congenital Hip Dysplasia
- c. Absent Radius
- d. Extra Rib
- e. Missing Rib
- f. Sacral Dimple
- g. Scoliosis (congenital)
- h. Tethered Cord
- i. Vertebral, butterfly
- j. Vertebral, fused
- k. Vertebral, other

i. Specify:

l. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C18. Were any abnormalities of the hands noted in the medical record?

- a. Abnormal Fingers (not thumb)
- b. Abnormal Thumb
- c. Polydactyly
- d. Syndactyly
- e. Other

i. Specify:

**Yes**      **No**      **Source Pending**

C19. Were any abnormalities of the feet noted in the medical record?

- a. Abnormal Toe
- b. Club Foot
- c. Polydactyly

- d. Syndactyly
- e. Wide-spaced Big Toe
- f. Other

i. Specify:

**Yes                  No                  Source  
Pending**

C20. Were any neurological abnormalities noted in the medical record?

- a. Absent Corpus Callosum
- b. Chiari Malformation
- c. Dandy-Walker
- d. Holoprosencephaly
- e. Hydrocephalus
- f. Neural Tube Defect
- g. Other

i. Specify:

**Yes                  No                  Source  
Pending**

C21. Were any dermatological abnormalities noted in the medical record?

- a. Cafe Au Lait Spots
- b. Hemangioma
- c. Hyperpigmented Lesions
- d. Hypopigmented Lesions
- e. Port Wine Spots
- f. Telangiectasia
- g. Other

i. Specify:

**Yes                  No                  Source  
Pending**

C22. Were any endocrinologic abnormalities noted in the medical record?

a. Specify:

**Yes                  No                  Source  
Pending**

C23. Were any abnormalities of the thymus noted in the medical record?

- a. Specify Type:
- Absent
  - Ectopic
  - Hypoplastic
  - Other
  - Unknown
  - Source Pending

i. Other, specify:

**Yes                  No                  Source  
Pending**

C24. Were any hematologic abnormalities noted in the medical record?

- a. Rh/ABO Incompatible
- b. Other

i. Specify:

**Yes                  No                  Source  
Pending**

C25. Were any genetic syndromes noted in the medical record?

- a. Alagille
- b. Cardiofacial Cutaneous
- c. Cat Eye
- d. CHARGE

- e. Cri-du-chat
- f. DGS/ VCFS/ CTAF
- g. Ehlers Danlos
- h. Ellis-Van Creveld
- i. Goldenhar
- j. Holt Oram
- k. Jacobsen
- l. Kabuki
- m. Marfan
- n. Microdeletion Syndrome
- i. Specify:
- o. Microduplication
- i. Specify:
- p. Noonan
- q. Translocation
- i. Specify:
- r. Trisomy 13
- s. Trisomy 18
- t. Trisomy 21 (Down)
- u. Turner
- v. VATER
- w. VACTERL
- x. Williams
- y. Multiple congenital anomalies, NOS
- z. Other
- i. Specify:

**Yes          No          Source  
Pending**

C26. Were any immunologic abnormalities noted in the medical record?  Yes  No  Source Pending

a. Specify: