Bench to Bassinet
Pediatric Cardiac Genomics Consortium: CHD GENES
Form 111: Genetic Testing
Version: D - 11/18/2011

SECTION A: ADMINISTRATIVE INFORMATION

A1. Study Identification Number:

A2. Study Visit: **Proband Subject Baseline Visit**

A3. Date Form Completed: *MM/DD/YYYY*

SECTION B: KARYOTYPE

	Yes	No	Unknown	Source Pending
B1. Karyotype Obtained:	○	○	○	○

B2. Date Karyotype Obtained: *MM/DD/YYYY*

B3. Source of Karyotype Data:
- ○ Actual report from lab
- ○ Family member
- ○ Mentioned in medical record, but no lab report
- ○ Genetic counselor verbal or written report
- ○ Nurse verbal or written report
- ○ Physician's verbal or written report
- ○ Other

a. Specify:

B4. Tissue Studied:
- ○ Amniotic fluid
- ○ Blood
- ○ Chorionic villi
- ○ Fibroblasts
- ○ Saliva
- ○ Other
- ○ Unknown
- ○ Source Pending

a. Specify:

	Normal	Abnormal	Unknown	Source Pending
B5. Karyotype Result:	○	○	○	○

If abnormal, please specify the abnormality: (check all that apply)

a. Balanced reciprocal translocation	☐	k. 47, XXX	☐
b. Balanced Robertsonian translocation	☐	l. 47, XXY	☐
c. Deletion	☐	m. 47, XYY	☐
d. Duplication	☐	n. Paracentric inversion	☐
e. Extra marker chromosome	☐	o. Pericentric inversion	☐
f. Trisomy 13	☐	p. Structural X chromosome abnormality	☐
g. Trisomy 18	☐	q. Structural Y chromosome abnormality	☐
h. Trisomy 21	☐	r. Unbalanced reciprocal translocation	☐
i. 22q11 deletion	☐	s. Unbalanced Robertsonian translocation	☐
j. 45, X	☐	t. Other	☐

i. If other, specify:

	Normal	Abnormal	Unknown	Source Pending
B6. Is the mother's karyotype known?	○	○	○	○

If abnormal, please specify the abnormality: (check all that apply)

- a. Balanced reciprocal translocation ☐
- b. Balanced Robertsonian translocation ☐
- c. Deletion ☐
- d. Duplication ☐
- e. Extra marker chromosome ☐
- f. Trisomy 13 ☐
- g. Trisomy 18 ☐
- h. Trisomy 21 ☐
- i. 22q11 deletion ☐
- j. 45, X ☐
- k. 47, XXX ☐
- l. Paracentric inversion ☐
- m. Pericentric inversion ☐
- n. Structural X chromosome abnormality ☐
- o. Structural Y chromosome abnormality ☐
- p. Unbalanced reciprocal translocation ☐
- q. Unbalanced Robertsonian translocation ☐
- r. Other ☐
- i. If other, specify: _____

	Normal	Abnormal	Unknown	Source Pending
B7. Is the father's karyotype known?	○	○	○	○

If abnormal, please specify the abnormality: (check all that apply)

- a. Balanced reciprocal translocation ☐
- b. Balanced Robertsonian translocation ☐
- c. Deletion ☐
- d. Duplication ☐
- e. Extra marker chromosome ☐
- f. Trisomy 13 ☐
- g. Trisomy 18 ☐
- h. Trisomy 21 ☐
- i. 22q11 deletion ☐
- j. 45, X ☐
- k. 47, XXY ☐
- l. 47, XYY ☐
- m. Paracentric inversion ☐
- n. Pericentric inversion ☐
- o. Structural X chromosome abnormality ☐
- p. Structural Y chromosome abnormality ☐
- q. Unbalanced reciprocal translocation ☐
- r. Unbalanced Robertsonian translocation ☐
- s. Other ☐
- i. If other, specify: _____

SECTION C: KARYOTYPE FORMULA

	Yes	No	Unknown	Source Pending
C1. Mosaic:	○	○	○	○

Cell Line 1

C2. Number of chromosomes:

C3. Sex chromosomes:
- ○ XX
- ○ XY
- ○ X
- ○ XXY
- ○ XXX
- ○ Unknown
- ○ Source Pending

C4. Abnormal chromosome:

- a. Band 1:
- b. Band 2:
- c. Band 3:

Cell Line 2 (if applicable)

	Yes	No
C5. Do you have data on another cell line?	○	○

C6. Number of chromosomes:

C7. Sex chromosomes:
- ○ XX
- ○ XY

- X
- XXY
- XXX
- Unknown
- Source Pending

C8. Abnormal chromosome:

a. Band 1:

b. Band 2:

c. Band 3:

Cell Line 3 (if applicable)

	Yes	No
C9. Do you have data on another cell line?		

C10. Number of chromosomes:

C11. Sex chromosomes:

- XX
- XY
- X
- XXY
- XXX
- Unknown
- Source Pending

C12. Abnormal chromosome:

a. Band 1:

b. Band 2:

c. Band 3:

C13. Karyotype Formula:

SECTION D: MICROARRAY TESTING

	Yes	No	Unknown	Source Pending
D1. Microarray Testing Performed:				

D2. Date test performed: *MM/DD/YYYY*

D3. Microarray Type:

- Oligonucleotide by aCGH
- SNP
- Targeted BAC
- Whole genomic BAC
- Unknown
- Source Pending

D4. Microarray Source:

- Affymetrix
- Agilent
- Illumina
- Nimblegen
- Other
- Unknown

a. Specify:

D5. Tissue Studied:

- Amniotic fluid
- Blood
- Chorionic villi
- Fibroblasts
- Saliva
- Other
- Unknown
- Source Pending

a. Specify:

D6. Number of probes on the array:

SECTION E: MICROARRAY RESULTS

	Yes	No	Unknown	Source Pending
E1. Variant present?	○	○	○	○

E2. Type of Variant:
- ○ Deletion
- ○ Duplication
- ○ Run of homozygosity
- ○ Unknown
- ○ Source Pending

E3. Chromosome:

E4. Starting band:

E5. Ending band:

E6. Starting base pair:

E7. Ending base pair:

E8. Size (in kb):

E9. Genome version:

E10. Type of microarray testing:
- ○ Oligonucleotide by aCGH
- ○ SNP
- ○ Targeted BAC
- ○ Whole genomic BAC

E11. Confirmatory testing performed:
- ○ FISH
- ○ qPCR
- ○ FISH and qPCR
- ○ Sequencing
- ○ Not Performed
- ○ Other
- ○ Unknown
- ○ Source Pending

a. Specify:

E12. Variant Confirmed:
- ○ Yes
- ○ Mosaic
- ○ Partially
- ○ No
- ○ Unknown
- ○ Source Pending

	Yes	No	Unknown	Source Pending
E13. Was the mother tested for this abnormality?	○	○	○	○
a. Was the abnormality present?	○	○	○	○
E14. Was the father tested for this abnormality?	○	○	○	○
a. Was the abnormality present?	○	○	○	○

E15. Final Interpretation:
- ○ Normal
- ○ De novo loss (other than 22q11 deletion)
- ○ De novo gain
- ○ Gain, unknown if inherited
- ○ Loss, unknown if inherited
- ○ Inherited Loss, maternal non-mosaic
- ○ Inherited Loss, paternal non-mosaic
- ○ Inherited Loss, maternal mosaic
- ○ Inherited Loss, paternal mosaic
- ○ Inherited Gain, maternal non-mosaic
- ○ Inherited Gain, paternal non-mosaic
- ○ Inherited Gain, maternal mosaic
- ○ Inherited Gain, paternal mosaic
- ○ 22q11 deletion, de novo
- ○ 22q11 deletion, inherited maternal
- ○ 22q11 deletion, inherited paternal
- ○ 22q11 deletion, unknown if inherited
- ○ Other
- ○ Unknown
- ○ Source Pending

a. Specify:

SECTION F: OTHER GENETIC TESTING

	Yes	No	Unknown	Source

				Pending
F1. Was other genetic testing done?	○	○	○	○

SECTION G: FISH

	Yes	No	Unknown	Source Pending
G1. FISH Analysis:	○	○	○	○

G2. Probes Analyzed:

- ○ Alagille
- ○ Alpha-satellite
- ○ Angelman
- ○ Chromosome paints
- ○ Prader-Willi
- ○ Smith-Magenis
- ○ Subtelomeric
- ○ Williams
- ○ 22q11
- ○ Other
- ○ Unknown
- ○ Source Pending

a. Specify:

G3. FISH Results:

- ○ Normal
- ○ Deletion
- ○ Duplication
- ○ Translocation
- ○ Unknown
- ○ Source Pending

a. Specify:

	Yes	No	Unknown	Source Pending
G4. Was the mother tested for this abnormality?	○	○	○	○
a. Was the abnormality present?	○	○	○	○
G5. Was the father tested for this abnormality?	○	○	○	○
a. Was the abnormality present?	○	○	○	○

SECTION H: TARGETED COPY NUMBER TESTING

	Yes	No	Unknown	Source Pending
H1. Targeted Copy Number Testing:	○	○	○	○

H2. Type of Test:

- ○ Exon array
- ○ MLPA
- ○ qPCR
- ○ Other
- ○ Unknown
- ○ Source Pending

a. Specify:

	Yes	No	Unknown	Source Pending
H3. Copy Number Variant Identified?	○	○	○	○

H4. Type of Variant:

- ○ Deletion
- ○ Duplication
- ○ Unknown
- ○ Source Pending

H5. Specify gene(s) and/or chromosome locus:

	Yes	No	Unknown	Source Pending
H6. Was the mother tested for this abnormality?	○	○	○	○
a. Was the abnormality present?	○	○	○	○
H7. Was the father tested for this abnormality?	○	○	○	○
a. Was the abnormality present?	○	○	○	○

SECTION I: MUTATION TESTING

	Yes	No	Unknown	Source Pending
I1. Mutation Testing:	○	○	○	○

I2. Date of test: _____ *MM/DD/YYYY*

I3. Source of information:
- ○ Actual report from lab
- ○ Family member
- ○ Mentioned in medical record, but no lab report
- ○ Genetic counselor verbal or written report
- ○ Nurse verbal or written report
- ○ Physician's verbal or written report
- ○ Other

a. Specify: _____

I4. Gene analyzed:

○ BRAF	○ NKX2.5
○ CHD7	○ NRAS
○ ELN	○ PTPN11
○ EVC	○ RAF1
○ EVC2	○ SHOC2
○ HRAS	○ SOS1
○ JAG1	○ TBX5
○ KRAS	○ ZIC3
○ MEK1	○ Other
○ MEK2	○ Unknown
○ MID1	○ Source Pending

a. Specify: _____

	Yes	No	Unknown	Source Pending
I5. Variant found?	○	○	○	○

a. Is this a pathogenic mutation or a variant of unknown significance?
- ○ Pathogenic mutation
- ○ Variant of unknown significance
- ○ Unknown
- ○ Source Pending

b. What kind of variant is it?
- ○ Deletion
- ○ Insertion
- ○ Missense
- ○ Nonsense
- ○ Silent substitution
- ○ Unknown
- ○ Source Pending

c. Zygosity:
- ○ Hemizygous
- ○ Heterozygous
- ○ Homozygous
- ○ Unknown
- ○ Source Pending

d. Specify Amino Acid change: _____

	Yes	No	Unknown	Source Pending
e. Was the mother tested for this abnormality?	○	○	○	○
i. Was the abnormality present?	○	○	○	○
f. Was the father tested for this abnormality?	○	○	○	○
i. Was the abnormality present?	○	○	○	○

SECTION J: OTHER TESTING

	Yes	No	Unknown	Source Pending
J1. Other test method:	○	○	○	○

J2. Specify:

J3. Source of information:
- Actual report from lab
- Family member
- Mentioned in medical record, but no lab report
- Genetic counselor verbal or written report
- Nurse verbal or written report
- Physician's verbal or written report
- Other

a. Specify:

J4. Date of test: *MM/DD/YYYY*

J5. Test result:
- Negative
- Confirmatory
- Other
- Unknown
- Source Pending

a. Specify:

	Yes	No	Unknown	Source Pending
J6. Was the mother tested for this abnormality?				
a. Was the abnormality present?				
J7. Was the father tested for this abnormality?				
a. Was the abnormality present?				