

Congenital Heart Disease

Selection Criteria – Congenital Heart Disease

CHD = In this context, "CHD" includes patients/relatives who required surgical or catheter-based intervention, so as not to capture individuals with trivial valvular disease, minor septal defects, small PDA's, etc.

Familial CHD = Non-syndromic CHD at any age AND ≥ 1 first degree relative with documented CHD ± additional affected family members

Extra Cardiac Anomalies (ECA) = major structural non-cardiac anomalies, for example diaphragmatic hernia, cleft lip/palate, intestinal atresia, midline defects, structural brain abnormalities, functional neurological impairment (when diagnosed by a neurologist/geneticist/paediatrician and fitting criteria for moderate neurodevelopmental disability on the basis of formal testing e.g. Bayley Scales in children or neurocognitive assessments in older children and adults).

Exclusion criteria:

- Deceased individuals
- NGS-based test performed from 2013 onwards
- Individuals from families with a known disease-causing variant
- Unascertained sudden death

Diagnosis:

- Familial CHD (go to i.)
 CHD + extra cardiac anomalies (go to ii.)

i. Familial CHD

Diagnostic criteria:

(MUST meet diagnostic criteria for dx)

- Non-syndromic CHD at any age **AND** ≥ 1 first and second degree relative with documented CHD
 ± Additional* affected family members

*If there is FHx of same condition in more distant relatives, forward case to adjudication committee

ii. CHD + extra cardiac anomalies (ECA)

Diagnostic criteria:

(MUST meet diagnostic criteria for dx)

- CHD at any age **AND** ≥ 1 documented major structural non-cardiac anomaly **AND** normal chromosomal microarray at any age*

*If previous CMA results are abnormal, but do not fully explain or are inconsistent with the phenotype, patient can be considered for inclusion.

Clinical History

Height _____ (cm)	Weight _____ (kg)
Calculated total Body Surface Area (m2)	_____
Date of diagnosis: _____ (If unsure of the exact date please set to January 1 of the year the event took place and set the "Exact date" option to "No".)	Is this an exact date? <input type="checkbox"/> Yes <input type="checkbox"/> No
Maternal diabetes?	<input type="checkbox"/> No <input type="checkbox"/> Gestational <input type="checkbox"/> Type I <input type="checkbox"/> Type II

Cardiac transplant:	<input type="checkbox"/> Yes <input type="checkbox"/> Listed <input type="checkbox"/> No
Date inserted:	_____ (If unsure of the exact date please set to January 1 of the year the event took place and set the "Exact date" option to "No".)
Is this an exact date?	<input type="checkbox"/> Yes <input type="checkbox"/> No
Date listed:	_____ (If unsure of the exact date please set to January 1 of the year the event took place and set the "Exact date" option to "No".)
Is this an exact date?	<input type="checkbox"/> Yes <input type="checkbox"/> No
LVAD:	<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> N/A
Date inserted:	_____ (If unsure of the exact date please set to January 1 of the year the event took place and set the "Exact date" option to "No".)
Is this an exact date?	<input type="checkbox"/> Yes <input type="checkbox"/> No
Procedures relevant for CHD:	<input type="checkbox"/> Open heart surgery <input type="checkbox"/> Cardiac catheterization
Number of procedures relevant for CHD:	<input type="checkbox"/> 1 <input type="checkbox"/> 4 <input type="checkbox"/> 2 <input type="checkbox"/> 5 <input type="checkbox"/> 3
First procedure: <i>*SNOMED CT Capture Field</i>	_____
First procedure description (if code not found):	_____
Second procedure: <i>*SNOMED CT Capture Field</i>	_____
Second procedure description (if code not found):	_____
Third procedure: <i>*SNOMED CT Capture Field</i>	_____
Third procedure description (if code not found):	_____
Fourth procedure: <i>*SNOMED CT Capture Field</i>	_____
Fourth procedure description (if code not found):	_____
Fifth procedure: <i>*SNOMED CT Capture Field</i>	_____
Fifth procedure description (if code not found):	_____
Uploads	
Echo at diagnosis or representative echo	
Number of ECGs at diagnosis or representative ECGs	
First ECG	
First ECG lead placement	<input type="checkbox"/> Standard <input type="checkbox"/> Elevated
Second ECG	

Second ECG lead placement	<input type="checkbox"/> Standard <input type="checkbox"/> Elevated
Third ECG	
Third ECG lead placement	<input type="checkbox"/> Standard <input type="checkbox"/> Elevated
Fourth ECG	
Fourth ECG lead placement	<input type="checkbox"/> Standard <input type="checkbox"/> Elevated
Fifth ECG	
Fifth ECG lead placement	<input type="checkbox"/> Standard <input type="checkbox"/> Elevated
CMRI at diagnosis or representative CMRI	
Ajmaline challenge report	
Flecainide challenge report	
Adrenaline challenge report	
Operation report	