Version 1, 24 September 2019



#### **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  $\geq$  1 first degree relative with documented CHD  $\pm$  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)		
<ul> <li>Non-syndromic CHD at any age AND ≥ 1 first and second degree relative with documented CHD</li> <li>± Additional* affected family members</li> <li>*If there is FHx of same condition in more distant relatives, forward case to adjudication committee</li> </ul>		
ii. CHD + extra cardiac anomalies (ECA)		
Diagnostic criteria: (MUST meet diagnostic criteria for dx)		
<ul> <li>CHD at any age AND ≥ 1 documented major structural non-cardiac anomaly AND normal chromosomal microarray at any age*</li> <li>*If previous CMA results are abnormal, but do not fully explain or are inconsistent with the phenotype, patient can be considered for inclusion.</li> </ul>		
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? Yes No	
Maternal diabetes?	☐ No ☐ Gestational ☐ Type I ☐ Type II	



Insulin-dependent   Oral hypoglycemic agents   Unknown
Born prematurely?    No   Yes   Unknown
Born prematurely?    No   Yes   Unknown
Gestation:  Other Conditions  Morphologic diagnostic group:  Abnormalities of position and connection of heart  Tetralogy of Fallot and variants  Abnormalities of great veins  Abnormalities of atriums and atrial septum  Abnormalities of AV valves and AV septal defect  Abnormalities of ventricles and ventricular septum  Abnormalities of VA valves and great arteries  Abnormalities of coronary arteries, arterial duct and pericardium; AV fistulae
Gestation:  Other Conditions  Morphologic diagnostic group:  Abnormalities of position and connection of heart  Tetralogy of Fallot and variants  Abnormalities of great veins  Abnormalities of atriums and atrial septum  Abnormalities of AV valves and AV septal defect  Abnormalities of ventricles and ventricular septum  Abnormalities of VA valves and great arteries  Abnormalities of coronary arteries, arterial duct and pericardium; AV fistulae
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Other
Number of CHD diagnosis:
$\begin{array}{ c c c c c c c c c c c c c c c c c c c$
First CHD diagnosis:
*SNOMED CT Capture Field
First CHD diagnosis description (if code not found):
Second CHD diagnosis:
*SNOMED CT Capture Field
Second CHD diagnosis description (if code not found):
Third CHD diagnosis:
*SNOMED CT Capture Field
Third CHD diagnosis description (if code not found):
Fourth CHD diagnosis:
*SNOMED CT Capture Field
Fourth CHD diagnosis description (if code not found):
Fifth CHD diagnosis:
*SNOMED CT Capture Field
Fifth CHD diagnosis description (if code not found):
Non-cardiac anomalies:



Hernias	Auditory abnormalities
Asplenia	Cleft lip/palate
Liver/renal abnormalities	Cleft lip/palate
Intestinal abnormalities	Dysmorphisms
Trachea and oesophageal abnormalities	Structural brain disorders
Spinal abnormalities	NDD
Skeletal abnormalities	Other
Optical and visual abnormalities	
Number of non-cardiac anomalies:	1 4
	□ 2   □ 3
First non-cardiac anomaly:	
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*SNOMED CT Capture Field	
First non-cardiac anomaly description (if code not	
found):	
Second non-cardiac anomaly:	
*SNOMED CT Capture Field	
Second non-cardiac anomaly description (if code not	
found):	
Third non-cardiac anomaly:	
*SNOMED CT Capture Field	
Third non-cardiac anomaly description (if code not	
found):	
Fourth non-cardiac anomaly:	
*SNOMED CT Capture Field	
Fourth non-cardiac anomaly description (if code not	
found):	
Fifth non-cardiac anomaly:	
*SNOMED CT Capture Field	
Fifth non-cardiac anomaly description (if code not	
found):	
Intervention Procedures	
PPM:	Yes No
Date implanted:	(If we want of the count data places and to leave and of
	(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?	Yes No
ICD:	
	(If the patient has had multiple ICDs, this refers to the
	first one)
Date implanted:	
	(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?	Yes No
Primary or secondary prevention	Primary Secondary
1	<u> </u>



Cardiac transplant:	Yes Listed 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?	Yes No
Date listed:	(If we was a fit has a wast data places and to large and 1 of
	(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?	Yes No
LVAD:	Yes No 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?	YesNo
Procedures relevant for CHD:	Open heart surgery
N	Cardiac catheterization
Number of procedures relevant for CHD:	
First procedure:	
*SNOMED CT Capture Field	
First procedure description (if code not found):	
Second procedure:	
*SNOMED CT Capture Field	
Second procedure description (if code not found):	
Third procedure:	
*SNOMED CT Capture Field	
Third procedure description (if code not found):	
Fourth procedure:	
*SNOMED CT Capture Field	
Fourth procedure description (if code not found):	
Fifth procedure:	
*SNOMED CT Capture Field	
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	Standard
	Elevated
Second ECG	



Second ECG lead placement	Standard
	☐ Elevated
Third ECG	
Third ECG lead placement	Standard
	☐ Elevated
Fourth ECG	
Fourth ECG lead placement	Standard
	☐ Elevated
Fifth ECG	
Fifth ECG lead placement	Standard
	☐ Elevated
CMRI at diagnosis or representative CMRI	
Ajmaline challenge report	
Flecainide challenge report	
Adrenaline challenge report	
Operation report	