Australian Genomics

Cardiomyopathies

Selection Criteria - Cardiomyopathies

Exclusion criteria:
    - Deceased individuals
    - NGS-based test performed from 2013 onwards
    - Individuals from families with a known disease-causing variant
    - Unascertained sudden death
 MUST meet diagnostic criteria for dx AND must be able to tick at least 1 box from at least 2 of the following categories: Age criteria, FHx criteria, Clinical criteria. PLEASE NOTE: FHx can be considered standalone if confirmed. If diagnostic criteria AND family history criteria are met, the patient is eligible.

Diagnosis
☐ HCM (go to i.)   ☐ LVNC (go to iv.)   ☐ DCM (go to ii.)   ☐ RCM (go to v.)   ☐ ARVC (go to iii.)

i. HCM
Diagnostic criteria: (MUST meet diagnostic criteria for dx)
☐ Adults: a wall thickness ≥ 15mm in one or more LV myocardial segments (± 1-2mm, i.e. within error of the measurement technique)   ☐ Children: LV wall thickness more than two standard deviations greater than the predicted mean   (z-score 2, where a z-score is defined as the number of standard deviations from the population mean)
Age criteria:
☐ Dx ≤ 50 yo   (If less than 18 yo, no additional criteria is needed)
Family history criteria: (If family history AND diagnostic criteria are met, the patient is eligible)
☐ ≥ 1 first or second degree relative with documented CM   ☐ ≥ 1 first or second degree relative with sudden death before 50 (must attempt to obtain PM report, death certificate and/or medical records to exclude other causes of death)
Clinical criteria:
☐ Wall thickness > 25mm (± 1-2mm, i.e. within error of the measurement technique)   ☐ Previous OHCA   (OHCA = out-of-hospital cardiac arrest)
ii. DCM
Diagnostic criteria:
☐ LVEDD (% predicted) > 112% + LVEF < 45%   ☐ Metabolic, infective and syndromic causes excluded (MUST meet diagnostic criteria for dx)   N.B. Worst echo report can be used.   Patients suspected of left dominant ARVC to be forwarded to the adjudication committee.
Age criteria:
☐ Dx ≤ 60 yo   (If less than 18 yo, no additional criteria is needed)
Family history criteria: (If family history AND diagnostic criteria are met, the patient is eligible.)

☐ ≥ 1 first or second degree relative with documented CM
☐ ≥ 1 first or second degree relative with sudden death before 50 (must attempt to obtain PM report, death certificate and/or medical records to exclude other causes of death)

Clinical criteria:
☐ Conduction disease
☐ OHCA
(OHCA = out-of-hospital cardiac arrest)

Predicted (%) - Left ventricular end diastolic dysfunction (LVEDD):
_____
(Percentage predicted (%) - Auto Calculation (Req - Height, Weight & measured LVEDD on echo report))

Peripartum presentation:
☐ Yes ☐ No

Current New York Heart Association (NYHA) functional classification:	
☐ I	☐ IV
☐ II	☐ NA
☐ III	

Previous worst recorded New York Heart Association (NYHA) functional classification:	
☐ I	☐ IV
☐ II	☐ NA
☐ III	

iii. ARVC

Diagnostic criteria:
(MUST meet diagnostic criteria for dx)

☐ Modified taskforce criteria - at least borderline
If borderline, <u>must have</u> FHx of ≥ first degree relative with documented ARVC or sudden cardiac death
N.B. Please forward patients suspected of left dominant ARVC to the adjudication committee.

Age criteria:
☐ Dx ≤ 60yo
(If less than 18, no additional criteria is needed)

Family history criteria:
(If family history AND diagnostic criteria are met, the patient is eligible.)

☐ ≥ 1 first or second degree relative with documented CM
☐ ≥ 1 first or second degree relative with sudden death before 50 (must attempt to obtain PM report, death certificate and/or medical records to exclude other causes of death)

Clinical criteria:
☐ Definite taskforce criteria

iv. LVNC

Diagnostic criteria:
(MUST meet diagnostic criteria for dx)

☐ Multiple trabeculations, deep intratrabecular recesses seen on colour flow Doppler and a 2-layered structure of the myocardium with ratio of non-compacted to compacted myocardium of > 2:1 in systole. This can include entirely normal cardiac function.

Age criteria:
☐ Dx ≤ 50yo
(If less than 18, no additional criteria is needed)

Family history criteria:
(If family history AND diagnostic criteria are met, the patient is eligible.)
☐ ≥ 1 first or second degree relative with documented CM ☐ ≥ 1 first or second degree relative with sudden death before 50 (must attempt to obtain PM report, death certificate and/or medical records to exclude other causes of death)
Clinical criteria:
☐ Impaired function on imaging

v. RCM
Diagnostic criteria:
(MUST meet diagnostic criteria for dx)
☐ Evidence of primary myocardial disease comprising LV diastolic dysfunction with normal / near normal wall thickness and systolic function
Age criteria:
☐ Dx ≤ 50yo (If less than 18, no additional criteria is needed)
Family history criteria:
(If family history AND diagnostic criteria are met, the patient is eligible.)
☐ ≥ 1 first or second degree relative with documented CM ☐ ≥ 1 first or second degree relative with sudden death before 50 (must attempt to obtain PM report, death certificate and/or medical records to exclude other causes of death)

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

Symptoms at diagnosis: ☐ Yes ☐ No	
☐ Shortness of breath ☐ Chest pain ☐ Pre-syncope ☐ Syncope ☐ Palpitations	☐ Heart failure ☐ OHCA ☐ Seizure(s) ☐ Epilepsy ☐ Other
Number of other additional symptoms:	☐ 1 ☐ 2
First additional symptom: *SNOMED CT Capture Field*	_____ _____
First additional symptom description (if code not found):	_____ _____
Second additional symptom: *SNOMED CT Capture Field*	_____ _____
Second additional symptom description (if code not found):	_____ _____

Asymptomatic list	☐ Diagnosed on family screening ☐ Incidental diagnosis; details: _____ _____

Other Conditions	
☐ HTN	☐ AF
☐ Diabetes	☐ Previous VT
☐ CAD	☐ Conduction system abnormalities
☐ Cancer	☐ Ventricular arrhythmias
☐ Syndrome diagnosis	☐ OHCA (other than at presentation)
☐ Metabolic conditions	☐ Other
☐ Neuromuscular conditions	(Do not include the principal diagnosis)

HTN	
First HTN date of diagnosis	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

Diabetes	
Date Diabetes diagnosed	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

CAD	
Number of coronary artery disease (CAD) conditions:	☐ 1 ☐ 2
First CAD type: *SNOMED CT Capture Field*	_____ _____
First CAD type description (if code not found):	_____
First CAD date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known
Second CAD type: *SNOMED CT Capture Field*	_____ _____
Second CAD type description (if code not found):	_____
Second CAD date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

Cancer	
Number of cancers:	☐ 1 ☐ 2
First cancer type: *SNOMED CT Capture Field*	_____ _____
First cancer type description (if code not found):	_____
First cancer date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known
Second cancer type: *SNOMED CT Capture Field*	_____ _____
Second cancer type description (if code not found):	_____
Second cancer date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known
Chemotherapy drugs:	☐ Yes ☐ No

Syndrome Diagnosis

Syndrome diagnosis: **OMIM Capture Field*	_____ _____
Syndrome diagnosis (if code not found):	_____
Syndrome diagnosis date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

Metabolic Conditions

Metabolic conditions: **OMIM Capture Field*	_____ _____
Metabolic conditions (if code not found):	_____
Metabolic conditions date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

Neuromuscular Conditions

Neuromuscular conditions: **OMIM Capture Field*	_____ _____
Neuromuscular conditions (if code not found):	_____
Neuromuscular conditions date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

AF

AF condition **SNOMED CT Capture Field*	_____ _____
AF conditions (if code not found):	_____
AF date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

Previous VT

Previous VT: **SNOMED CT Capture Field*	_____ _____
Previous VT (if code not found):	_____
Previous VT date of diagnosis:	_____
Is than an exact date?	☐ Yes ☐ No ☐ Date not known

Conduction System Abnormalities

Conduction system abnormalities type: **SNOMED CT Capture Field*	_____ _____
Conduction system abnormality type description (if code not found):	_____ _____
Conduction system abnormalities diagnosis date	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known

Ventricular Arrhythmias

Ventricular arrhythmia type: **SNOMED CT Capture Field*	_____ _____
Ventricular arrhythmia type description (if code not found):	_____ _____

Ventricular arrhythmia date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known
Out of hospital cardiac arrest (OHCA)	
OHCA	☐ Yes ☐ No
OHCA date:	_____
Other	
Number of other additional conditions:	☐ 1 ☐ 2
First additional condition: *SNOMED CT Capture Field*	_____ _____
First additional condition description (if code not found):	_____ _____
First additional condition date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known
Second additional condition: *SNOMED CT Capture Field*	_____ _____
Second additional condition description (if code not found):	_____ _____
Second additional condition date of diagnosis:	_____
Is this an exact date?	☐ Yes ☐ No ☐ Date not known
Echo Parameters at Diagnosis	
Left ventricular end diastolic diameter (LVEDD) on the Echo Report:	_____ (mm) If the patient has had more than 1 echo, use the measured value that is the highest
Intraventricular septal thickness at diastole (IVSD):	_____ (mm)
Left ventricular posterior wall end diastole (LVPWd)	_____ (mm)
FS and / or LVEF:	☐ FS only ☐ LVEF only ☐ FS and LVEF
For HCM:	
LV hypertrophy type: ☐ Asymmetric ☐ Apical ☐ Concentric ☐ Other ☐ Unknown	
LV outflow obstruction: ☐ Yes ☐ No ☐ Unknown	
For LVNC:	
Pathophysiology: ☐ DCM ☐ HCM ☐ RCM ☐ Normal ☐ Mixed ☐ Unknown	

Please specify: _____	
Fractional shortening (FS):	_____ (percentage)
Left ventricular ejection fraction (LVEF):	_____ (percentage)
LVNC:	☐ Yes ☐ No ☐ Unknown
Valve disease:	☐ Yes ☐ No ☐ Unknown
Valve disease type: *SNOMED CT Capture Field*	_____ _____
Valve disease type description (if code not found):	
Congenital heart disease (CHD):	☐ Yes ☐ No ☐ Unknown
Congenital heart disease detail: *SNOMED CT Capture Field*	_____ _____
Congenital heart disease detail description (if code not found):	_____ _____
RV involvement:	_____
If diagnosis was ARVC:	
LV involvement	☐ Yes ☐ No ☐ Unknown
Taskforce Criteria (ARVC only):	
CATEGORY 1: GLOBAL OR REGIONAL DYSFUNCTION AND STRUCTURAL ALTERATION	
Major	
By 2D echo: - Regional RV akinesia, dyskinesia, or aneurysm - and 1 of the following (end diastole): - PLAX RVOT ≥ 32 mm (corrected for body size [PLAX/BSA] ≥ 19 mm/m2) - PSAX RVOT ≥ 36 mm (corrected for body size [PSAX/BSA] ≥ 21 mm/m2) - or fractional area change ≤ 33%	☐ Yes
By MRI: - Regional RV akinesia or dyskinesia or dyssynchronous RV contraction - and 1 of the following (end diastole): - Ratio of RV end-diastolic volume to BSA ≥ 110 mL/m2 (male) or - ≥ 100 mL/m2 (female) or - RV ejection fraction ≤ 40%	☐ Yes
By RV angiography: - Regional RV akinesia, dyskinesia, or aneurysm	☐ Yes
Minor	
By 2D echo: - Regional RV akinesia or dyskinesia - and 1 of the following (end diastole): - PLAX RVOT ≥ 29 to < 32 mm (corrected for body size [PLAX/BSA] ≥ 16 to < 19 mm/m2) - PSAX RVOT ≥ 32 to < 36 mm (corrected for body size [PSAX/BSA] ≥ 18 to < 21 mm/m2) - or fractional area change > 33% to ≤ 40%	☐ Yes

By MRI: - Regional RV akinesia or dyskinesia or dyssynchronous RV contraction - and 1 of the following: - Ratio of RV end-diastolic volume to BSA ≥ 100 to < 110 mL/m2 (male) or ≥ 90 to < 100 mL/m2 (female) - or RV ejection fraction > 40% to ≤ 45%	☐ Yes
CATEGORY 2: TISSUE CHARACTERIZATION OF WALL	
Major	
Residual myocytes < 60% by morphometric analysis (or < 50% if estimated), with fibrous replacement of the RV free wall myocardium in ≥ 1 sample, with or without fatty replacement of tissue on endomyocardial biopsy.	☐ Yes
Minor	
Residual myocytes 60% to 75% by morphometric analysis (or 50% to 65% if estimated), with fibrous replacement of the RV free wall myocardium in ≥ 1 sample, with or without fatty replacement of tissue on endomyocardial biopsy.	☐ Yes
CATEGORY 3: REPOLARIZATION ABNORMALITIES	
Major	
Inverted T waves in right precordial leads (V1, V2, and V3) or beyond in individuals >14 years of age (in the absence of complete right bundle-branch block QRS ≥ 120 ms).	☐ Yes
Minor	
Inverted T waves in leads V1 and V2 in individuals >14 years of age (in the absence of complete right bundle-branch block) or in V4, V5, or V6.	☐ Yes
Inverted T waves in leads V1, V2, V3, and V4 in individuals >14 years of age in the presence of complete right bundle-branch block.	☐ Yes
CATEGORY 4: DEPOLARIZATION/CONDUCTION ABNORMALITIES	
Major	
Epsilon wave (reproducible low-amplitude signals between end of QRS complex to onset of the T wave) in the right precordial leads (V1 to V3).	☐ Yes
Minor	
Late potentials by SAECG in ≥ 1 of 3 parameters in the absence of a QRS duration of ≥ 110 ms on the standard ECG.	☐ Yes
Filtered QRS duration (fQRS) ≥ 114 ms.	☐ Yes
Duration of terminal QRS < 40μV (low-amplitude signal duration) ≥ 38 ms.	☐ Yes
Root-mean-square voltage of terminal 40 ms ≤ 20μV.	☐ Yes
Terminal activation duration of QRS ≥ 55 ms measured from the nadir of the S wave to the end of the QRS, including R', in V1, V2, or V3, in the absence of complete right bundle-branch block.	☐ Yes

CATEGORY 5: ARRHYTHMIAS	
Major	
Nonsustained or sustained ventricular tachycardia of left bundle-branch morphology with superior axis (negative or indeterminate QRS in leads II, III, and aVF and positive in lead aVL).	☐ Yes
Minor	
Nonsustained or sustained ventricular tachycardia of RV outflow configuration, left bundle-branch block morphology with inferior axis (positive QRS in leads II, III, and aVF and negative in lead aVL) or of unknown axis.	☐ Yes
>500 ventricular extrasystoles per 24 hours (Holter).	☐ Yes
CATEGORY 6: FAMILY HISTORY	
Major	
ARVC/D confirmed in a first-degree relative who meets current Task Force criteria.	☐ Yes
ARVC/D confirmed pathologically at autopsy or surgery in a first-degree relative.	☐ Yes
Identification of a pathogenic mutation categorized as associated or probably associated with ARVC/D in the patient under evaluation.	☐ Yes
Minor	
History of ARVC/D in a first-degree relative in whom it is not possible or practical to determine whether the family member meets current Task Force criteria.	☐ Yes
Premature sudden death (< 35 years of age) due to suspected ARVC/D in a first-degree relative.	☐ Yes
ARVC/D confirmed pathologically or by current Task Force Criteria in second-degree relative.	☐ Yes
Electrophysiological Parameters	
Exercise stress test (EST) performed?	☐ Yes ☐ No ☐ Unknown
EST type	☐ Bruce ☐ Sprint
EST result	☐ Normal ☐ Abnormal QTc in recovery ☐ Blood pressure drop during exercise ☐ VT ☐ Ventricular ectopic beats ☐ Other arrhythmia
Longest QTc in recovery:	_____ (msec)
VT:	☐ Monomorphic ☐ Polymorphic ☐ Bidirectional
Ventricular ectopic beats morphology:	☐ Monomorphic ☐ Polymorphic

Ventricular ectopic beats distributional pattern:	☐ Single ☐ Couplets ☐ Triplets
Ventricular ectopic beats, when?	☐ At rest ☐ During exercise ☐ During max exercise ☐ In recovery
Other arrhythmia:	_____
Holter monitor:	

☐ Normal	☐ Ventricular ectopic beats - monomorphic
☐ Unknown	☐ Ventricular ectopic beats - polymorphic
☐ Atrial tachycardia	☐ Ventricular ectopic beats - couplets
☐ Afib	☐ Ventricular ectopic beats - triplets
☐ Torsades	☐ Sustained VT
☐ AV Block II - intermittent	☐ Non sustained VT
☐ AV Block II - persistent	☐ AVRT/AVNRT
☐ AV Block III - intermittent	☐ Abnormal pauses for age
☐ AV Block III - persistent	☐ Significant Bradycardia
☐ Brugada pattern	☐ Other; please specify _____

Intervention Procedures	
PPM:	☐ Yes ☐ No
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
ICD:	☐ Yes ☐ No (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
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 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
CRT:	☐ 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?	☐ 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?	☐ Yes ☐ No
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	