Referral and follow-up of paediatric patients with family history or diagnosis of inherited cardiomyopathy (ICC)

A) Referral criteria

The following patients should be referred for assessment at the specialist paediatric inherited cardiomyopathy clinic:

- Age less than 18 years AND:
    - First-degree relative (parent/ sibling) with diagnosed genetic/ familial cardiomyopathy
      OR
    - Positive cardiomyopathy cascade genetic screening
      OR
    - Diagnosis of cardiomyopathy (abnormal heart function/ myocardial structure on echocardiogram)

B) Predictive genetic testing in inherited cardiomyopathies

When there is a pathogenic or likely pathogenic variant in a gene known to cause inherited cardiomyopathy in a family, it **may be** appropriate for cascade predictive genetic testing to be offered to children. There is no clear consensus in the community regarding best timing for this testing to be offered in children at risk of an inherited cardiomyopathy. ESC 2023 Guidelines for the management of cardiomyopathies outline specific issues to consider and advocate for a patient-centered approach. In general, considering family history, timing of clinical screening, autonomy of the child, and parental anxiety are key factors. The familial phenotype (P) and genotype (G) should be confirmed before considering genetic testing in a child. **Predictive genetic testing can only be requested by clinical genetics under the National genomic test directory, and therefore families should be referred to have genetic counselling if they wish to undergo predictive testing**.[1,8,9]

NB. This guidance is different from *diagnostic* genetic testing. Testing and indications are outlined in the National Genomic Test directory.[8,9]

C) Follow-up guidelines according to diagnosis

Structured according to current international guidelines (ESC 2023, AHA/ACC 2024 for HCM, HRS 2019, AHA 2019 Paediatric Cardiomyopathy Statement).[1-4]

General Practical Framework (All Phenotypes)

• Infancy–early childhood: follow-up only if genotype positive or early-onset family disease.
• Puberty/ adolescence: increase to annual review (growth and hormones can unmask phenotype).
• Review immediately if new symptoms (syncope, chest pain, palpitations).
• Repeat Holter/ CMR earlier if symptoms or abnormal findings.
• Discontinue follow-up only when genotype negative for known family variant and normal phenotype.[1,4]

1. Hypertrophic Cardiomyopathy (HCM)

Group	Screening starting age	Clinic + ECG + Echo	Holter	Cardiac MRI (CMR)
G + / P –	10–12 yrs (earlier if early-onset in family)[1,2]	Every 12 months (child/ adolescent)[1,2]	Every 1–2 yrs and individualized if arrhythmic symptoms[1,2]	Baseline ≥10 yrs; repeat every 3–5 yrs or if echo changes[1,2]
G unknown / P –	10–12 yrs (or 5 yrs before earliest family onset)[1,2]	Every 2 years [1–3 yrs][1,2]	Every 2–3 yrs or if symptoms [1,2]	Baseline by mid-teens; repeat if abnormal[,2]

Paediatric SCD risk calculators (these are not validated in syndromic HCM – i.e: Noonan Syndrome):

- hcmriskkids.org [5,7]
- Primacy Calculator – Primacy Calculator [6,10]

2. Dilated Cardiomyopathy (DCM)

Group	Screening starting age	Clinic + ECG + Echo	Holter	Cardiac MRI (CMR)
G + / P –	10–12 yrs (earlier if early-onset in family)[1] Could be considered from genetic diagnosis	Every 12–24 months[1] Annual if high-risk genotype (*EMD, PLN, DSP, LMNA, FLNC, TMEM43, RBM20*)	Every 1–2 yrs and individualized if arrhythmic symptoms[1]	Baseline ≥10 yrs; repeat every 3–5 yrs or if echo changes[1]
G unknown / P –	10–12 yrs (earlier if early family onset)[1]	Every 2–3 yrs[1]	Every 2–3 yrs or if symptoms[1]	Baseline by mid-teens; repeat if abnormal [1]

3. Arrhythmogenic Cardiomyopathy (ACM / ARVC)

Group	Screening starting age	Clinic + ECG + Echo	Holter	Cardiac MRI (CMR)
G + / P –	10–12 yrs (or 5 yrs before earliest family case)[1,3]	Every 12 months (child/ adolescent)[1,3]	Annually (adolescents)[1,3]	Baseline ≥10 yrs; repeat every 3–5 yrs[1,3]
G unknown / P –	10–12 yrs[1,3]	Every 2–3 yrs[1,3]	Every 2–3 yrs[1,3]	Baseline by late teens[1,3]

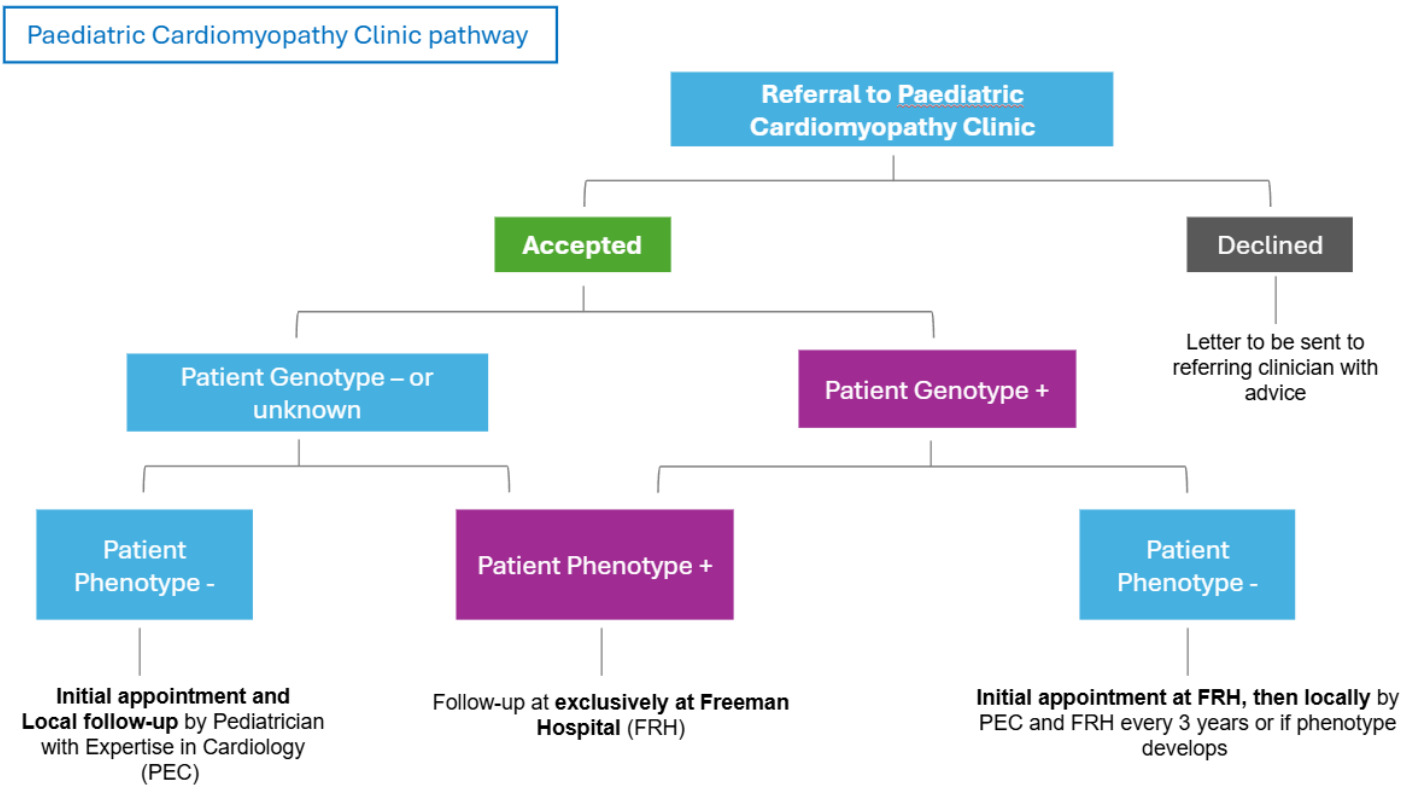
4. Restrictive Cardiomyopathy (RCM)

Group	Screening starting age	Clinic + ECG + Echo	Holter	Cardiac MRI (CMR)
G + / P –	From genetic diagnosis[1,4]	Annually[1,4]	Annually if conduction disease family history[1,4]	Baseline ≥10 yrs; repeat as indicated[1,4]
G unknown / P –	10–12 yrs[1,4]	Every 1–2 yrs[1,4]	Every 2 yrs[1,4]	Baseline once adolescent[1,4]

5. Left Ventricular Non-Compaction (LVNC)

Group	Screening starting age	Clinic + ECG + Echo	Holter	Cardiac MRI (CMR)
G + / P –	From genetic diagnosis[1,4]	Every 1–2 yrs[1,4]	Every 1–2 yrs if borderline LV function[1,4]	Baseline ≥10 yrs; repeat every 3–5 yrs[1,4]
G unknown / P –	From birth[1,4]	Every 2–3 yrs[1,4]	Every 2–3 yrs[1,4]	Baseline once; repeat if echo changes[1,4]

D) Proposed location of follow-up



E) References

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[5] Norrish G, Ding T, Field E, et al. Development of a Novel Risk Prediction Model for Sudden Cardiac Death in Childhood Hypertrophic Cardiomyopathy (HCM Risk-Kids). JAMA Cardiol. 2019;4(9):918-927. doi:10.1001/jamacardio.2019.2861.

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[8] NHS England. National genomic test directory (Rare and inherited disease). Version 8.1, published 10 July 2025; page updated 28 Aug 2025.

[9] NHS England. National genomic test directory: Testing criteria for rare and inherited disease. Version 7 (July 2024) eligibility criteria document.

[10] PRIMaCY calculator (primacycalculator.com). Accessed 27 Feb 2026.