

Genomic testing in Cardiology

Genomic testing in cardiology is outlined in the national genomic test directory (Rare and inherited disease eligibility criteria PDF). This can be accessed:

<https://www.england.nhs.uk/publication/national-genomic-test-directories/>.

Each genomic test is associated with a testing indication, which is coded using an R number. See below for the clinical indications and R numbers in cardiology.

Part II. Cardiology

R137	Congenital heart disease - microarray	15
R125	Thoracic aortic aneurysm or dissection.....	16
R127	Long QT syndrome	17
R128	Brugada syndrome and cardiac sodium channel disease	18
R129	Catecholaminergic polymorphic VT	19
R130	Short QT syndrome.....	20
R131	Hypertrophic cardiomyopathy.....	21
R132	Dilated and arrhythmogenic cardiomyopathy.....	22
R391	Barth syndrome	23
R133	Arrhythmogenic right ventricular cardiomyopathy	24
R135	Paediatric or syndromic cardiomyopathy.....	25
R136	Primary lymphoedema.....	27
R138	Sudden unexplained death or survivors of a cardiac event.....	28
R328	Progressive cardiac conduction disease	29
R384	Generalised arterial calcification in infancy.....	30
R140	Elastin-related phenotypes	31
R441	Unexplained death in infancy and sudden unexplained death in childhood.....	32
R454	Mavacamten for treating symptomatic obstructive hypertrophic cardiomyopathy	33

Please review the national genomic test directory for the testing criteria to see if your patient is eligible for testing.

Patients that do not fulfil these criteria may still warrant a genomic test, please discuss these cases at the regional ICC MDT meeting.

Consent for genomic testing

An example of a consent form is available, this can be completed when taking consent for genomic testing in cardiology. It is not essential you take written consent; however, the consent form included outlines the important discussion points that should be covered.

Samples

Please send an EDTA blood sample with a DNA request form to your local laboratory.

Please complete the 'NEY Genetic testing request form' with clinical information, including details of how the patient meets the National genomic test directory inclusion criteria and the test including R number.

If you anticipate a patient may need a genomic test in the future, you are able to send a EDTA blood sample for DNA storage only.

In a deceased patient, please send fresh frozen tissue, such as spleen.

Receiving the results and timeline

The genomic report will be sent to the email documented on the 'email address for the report' on the genomic test request form. The report will not routinely be linked to your patient's local record; therefore, it would be good practice to ensure you record their result in their local record.

Routine results will likely take a number of months. If you feel urgent testing is indicated, such as the patient is pregnant and results will influence management of pregnancy, please discuss this with clinical genetics. Urgent testing takes a number of weeks.

MDT Meetings

ICC MDT

There is a regional ICC MDT meeting held via teams on the second Tuesday of the month at 1300-1400. To refer your patient, please complete the [inherited cardiac conditions MDT case proforma](#) and send to nuth.heartteamadmin@nhs.net.

This meeting includes ICC cardiologists, cardiac family history nurses, genetic counsellors, clinical geneticists. Please use this meeting to discuss ICC cases, including cases where you have received genomic results, as outlined below in the outcomes of genomic testing and actions.

ICC Imaging MDT

There is a regional ICC imaging MDT meeting held via teams on the second Thursday of the month at 1300-1400.

This meeting includes ICC cardiologists, cardiologists with a specialist interest in imaging and clinical genetics. Please use this meeting if there is diagnostic uncertainty and MDT review of the images is required. To refer your patient, please complete the [ICC imaging MDT case proforma](#) and send to nuth.heartteamadmin@nhs.net.

General information

Minutes will be documented by NUTH heart admin team and distributed to the referring clinician and recorded on NUTH powerchart.

For calendar invites please contact nuth.heartteamadmin@nhs.net.

It is good practice to **present your own patient at the MDT**. Please try to ensure you make yourself available for this. If this is not feasible, cases can be discussed in a clinician's absence if there is clinical urgency for an outcome.

Outcomes of genomic testing and actions

There are three possible outcomes for the results:

1. Pathogenic / likely pathogenic variant

Finding a pathogenic / likely pathogenic variant which provides a genetic diagnosis for the clinical presentation. **Please inform your patient of the results** and onward plan for referral to clinical genetics.

Please refer your patient onto the Genetic counselling team via nuth.geneticsreferrals@nhs.net. The genetic counselling team will counsel regarding the the outcome, the inheritance, and the implications for the patient's family. The genetic counselling team will organise the family cascade testing of the likely pathogenic/pathogenic variant to those at risk of inheriting.

If you have clinical questions based on the findings of the molecular variant, e.g. specific genotype implications for patient management or risk stratification, then please refer and present your patient to the regional ICC MDT meeting via nuth.heartteamadmin@nhs.net, a case proforma is required.

2. Variant of uncertain significance

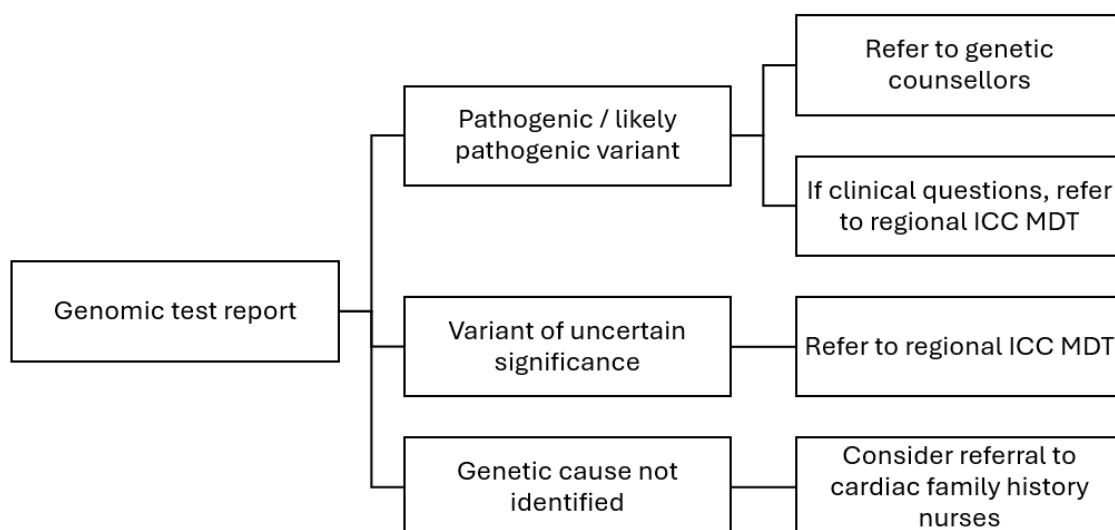
A variant of uncertain significance is a variant where there is insufficient evidence to determine if it is simply part of normal genetic variation (benign) or it is likely disease causing (pathogenic / likely pathogenic). This result should not be used for clinical decision making.


Please refer and present your patient to the regional ICC MDT meeting via nuth.heartteamadmin@nhs.net, a case proforma is required.

3. Genetic cause not identified

This result does not rule out a genetic cause for the patient's or the family history.

Please consider the likelihood of a genetic diagnosis and whether family first degree relative clinical screening is indicated. A referral to the Cardiac Family history nurses should be considered.



 North East and Yorkshire Genomic Laboratory Hub		Genetic Testing Request Form Rare Disease https://ney-genomics.org.uk/		Lab Use Only Lab No: Date received: dd/mm/yyyy	
Patient Information – use sticker if available				Requesting Consultant / Genetic Counsellor	
NHS No:		D.O.B:	dd/mm/yyyy	Full Name:	
Surname:		Sex:		Contact E-mail:	
Forename:		Ethnicity:		Hospital:	
Patient's Address:		Hospital No:		Ward /Clinic:	
Postcode:		Clinical Genetics No:		Address/Email for report:	
High risk of Infection: <input type="checkbox"/> If yes, please affix label to samples and form and specify likely infection:					
Test Required – please refer to National Genomic Test Directory (https://www.england.nhs.uk/publication/national-genomic-test-directories/). N.B. WGS requests require a WGS RD Trio Form and Records of Discussion Rare Disease samples will not be accepted without an R number and test name					
R Number:		Test:			
Clinical details Type of Test (please tick): Diagnostic <input type="checkbox"/> SNP array <input type="checkbox"/> Carrier <input type="checkbox"/> Karyotype <input type="checkbox"/> Carrier population risk <input type="checkbox"/> DNA storage ONLY <input type="checkbox"/> Pre-symptomatic/Predictive <input type="checkbox"/>			Please list how the patient meets the testing criteria and provide any additional pertinent clinical information. By requesting this test you are confirming that this patient meets the eligibility criteria as defined by the: National Genomic Test Directory . For familial tests include details of affected family members.		
Extracted DNA will be stored in the laboratory, please tick box if consent for storage has NOT been given <input type="checkbox"/>					
Known familial consanguinity? <input type="checkbox"/>					
Urgent? <input type="checkbox"/> Reason if Y:					
Telephone/Bleep for urgent results:					
Clinical Utility (Please provide additional information with other relevant clinical information above)			<input type="checkbox"/> Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme) <input type="checkbox"/> Patient, parents, or adult relative reproductive decision making <input type="checkbox"/> Unaffected relatives are seeking predictive testing		
Specimen details		Sample Type: <input type="checkbox"/> EDTA Blood (2- 5 ml) All genetic testing (except Karyotype) <input type="checkbox"/> Heparin Blood (2-5 ml) For Karyotype testing <u>only</u> <input type="checkbox"/> Other (please specify):			
Sample Date: dd/mm/yyyy					
Taken by:					
Once taken, samples should be sent to your local Genetics Laboratory Please ensure a minimum of 3 matching identifiers on tubes and form; Samples should be packed according to UN3373 / P650 and sent 1 st class post will normally be suitable for DNA extraction. Please store samples at 4°C if they cannot be transported the same day.					
Newcastle Genetics Laboratory	Newcastle Genetics Laboratory, Central Parkway, Newcastle upon Tyne, Tyne and Wear, NE1 3BZ	nuth.dna@nhs.net 0191 241 8787/8775/8754 www.newcastlelaboratories.com/lab_service/laboratory-rare-diseases-services/			
		scn-tr.sheffield.diagnosticgenetics@nhs.net 0114 271 7014 www.sheffieldchildrens.nhs.uk/SDGS.htm			
Sheffield Genetics Laboratory	Sheffield Diagnostic Genetics Service, Sheffield Children's NHS Foundation Trust, Western Bank, Sheffield, S10 2TH	leedsth-tr.genlabadmin@nhs.net 0113 206 5419/5205 https://www.leedsth.nhs.uk/services/pathology/the-leeds-genetics-laboratory/			
		https://www.leedsth.nhs.uk/services/pathology/the-leeds-genetics-laboratory/			
Leeds Genetics Laboratory	Leeds Genetics Laboratory, Genomic Specimen Reception, Bexley Wing (Level 5), St James's University Hospital, Beckett Street, Leeds, LS9 7TF				

411.027. Version 4.0. Author: Miranda Durkie, Sheffield Children's NHS Foundation Trust

An online genetic request form is available at <https://ney-genomics.org.uk/testing/referral-forms/>.

Affix patient label; or write
Name
Date of birth
NHS no.
Address

Consent form

Gene panel testing in cardiology and DNA storage

I have discussed genetic testing with my health professional and consent to:

1. Having a genetic test to look for important changes in known genes linked to

Insert name of gene panel

2. The Genetics laboratory storing any of my DNA left over after the test has been done. This might be used as a 'quality control' for other testing. Further genetic testing would only be done on my sample with my consent.
3. Results from my genetic test being stored as part of my health records.

I understand that:

1. Finding a significant gene change may give an explanation for the inherited heart condition in me / my family. Some of the genes being tested are also associated with a higher chance of other related medical problems.
2. The results of my test may reveal an 'uncertain' result. Genetic variation is common, and it is not always clear if a particular gene change is linked to an inherited heart condition. The interpretation of my results may change over time.
3. The results of my test may have implications for other members of my family. Results may be used to inform testing and counselling of other family members. This could be done in discussion with me, or in such a way that I am not directly identified in the process.
4. Sometimes a genetic test will result in feelings of anxiety, a burden of knowing about an increased risk of heart problems, and the need to share this information with family members.
5. A normal genetic test does not exclude an inherited link. Further advice and/or screening for me and my relatives might be suggested.
6. I can talk to a Clinical Genetics professional if I have further questions.

Note of any other specific issues discussed

Patient signature _____

Date _____

Discussion undertaken by _____ (clinician's name)

Inherited cardiac conditions MDT Case Proforma

Patient name	
Patient DOB	
MRN / NHS number	
Family ID / GC file number	
Responsible clinician	
MDT Attendees	
Case summary	
Results of genetic testing	
Question for MDT	
MDT Outcome	
Minutes written by	
Date of meeting	

Please refer your patient to the regional ICC MDT meeting via nuth.hearteamadmin@nhs.net.

ICC imaging MDT Case Proforma

Patient's name and DoB	
MRN / NHS number	
Family ID / GC file number if applicable	
Responsible clinician	
MDT Attendees	
Case summary	
Results of genetic testing if available	
MDT Outcome	
Minutes written by	
Date of meeting	

Please refer your patient to the regional ICC Imaging MDT meeting via nuth.heartteamadmin@nhs.net.