Delivery Tool for Core Training in Inherited Cardiovascular Conditions

Dr. E Wicks (Oxford), Dr. A Dimarco (Basildon), Dr S Page (Leeds), Prof. S Mohiddin (London). November 2022.

How to use this document:

This document describes all necessary curriculum requirements for basic training in inherited cardiac conditions (ICCs). The increasing incidence and awareness of inherited heart diseases necessitates acquisition of a minimum core of relevant knowledge and competencies for all cardiologists.

The term ICC embraces cardiovascular disease for which a genetic aetiology is confirmed or suspected, and includes families affected by sudden cardiac death. ICCs are often considered in three categories (heart muscle disease [cardiomyopathies], arrhythmia [channelopathies] and vasculopathies), and although familial hypercholesterolaemia (FH) and other monogenic disorders are often considered separately, many of the basic principles covered here are relevant to the management of a range of presentations where genetic causes should be considered.

This curriculum delivery tool details the training requirements needed to demonstrate core competence in ICC.

Methods of assessment:

This will be achieved through subspecialty MDTs, ward rounds and out-patient clinics, through on calls for general cardiology and general medicine, through existing workplace-based assessments (WBA), mini-Clinical Evaluation Exercise (mini-CEX), Case Based Discussion (CBD), a Patient Survey (PS), course certificates and the educational supervisors report.

Delivery of Core Knowledge Component

Trainees are expected to gain core knowledge of ICCs and heart muscle diseases through the following methods:

Either

i. Attendance at a training course on the basics of genetics and ICCs for formal training

Or

ii. Completion of online training and self-assessment for formal training

And

Supervised clinical exposure to an appropriate range of relevant clinical contexts and pathologies.

NOTE:

- Trainees may link with their regional specialist ICC centre and complete their placement within this centre under the supervision of a dedicated ICC specialist.
- Alternatively trainees may attend (remotely or in person) an appropriate range of clinical activity on an intermittent basis.
- If the trainee has difficulty in obtaining appropriate support for this training, they must inform either their TPD or ICC lead.
- The trainee may complete the recommended check list of component competencies and pathologies or provide an appropriate range of reflection and WBAs. If the check list is used it should be countersigned by the ICC supervisor.
- The trainees must also acquire and demonstrate a basic understanding of genetic counselling and the role counselling plays in family screening and in both predictive testing and diagnostic genetic testing.

- A trainee will be expected to understand indications for cascade and genetic family screening, and to demonstrate an ability to initiate screening, including an appreciation of appropriate genetic, social, moral and legal considerations.
- Typically, trainees should find that their involvement in at least 30 ICC cases, distributed across the spectrum of ICCs, should provide the clinical exposure needed to deliver competence for core training. Evidence of this should be available for review when discussing sign off of any section. Evidence should include an understanding of the key clinical features and of trainee participation in decision making for each case. Clinical exposure can be achieved through dedicated on-site clinical sessions and/or remote clinical attendance and/or contributions to MDTs.
- The theory component will be largely self-taught. In addition, on line training and guidance is available from the ESC myocardial and pericardial diseases working group, the Association of Inherited Cardiovascular Conditions (AICC), Health Education England's genomics resources, the American Heart association and from the National Centre for Biotechnology Information (NIH, USA). Portals to these rich educational resources and to other resources are included in the bibliography.

Components of competency in practice for the management of ICC patients include:

1. Understand the classification of ICCs (including chromosomal, single gene (Mendelian), polygenic and mitochondrial disorders) and have an appreciation of genetic heterogeneity and pleiotropy.
2. Diagnose the common ICCs and devise an initial management plan.
3. Take a family history and construct/interpret a pedigree, demonstrating an understanding of common inheritance patterns.
4. Understand indications for family screening and genetic evaluation and show competency in providing pre-screening counselling.
5. Demonstrate competency in the use of online genetic testing directories and other online reference material.
6. Understand how and when a genetic diagnosis in ICC patients is likely to be clinically useful.
7. Recognise features suggestive of inherited or inflammatory disease in patients with heart failure.
8. Understand mechanisms of symptom limitation in hypertrophic cardiomyopathy (HCM) and symptomatic indications for pharmacological and invasive therapy.
9. Demonstrate an understanding of avoidable/treatable risks posed by sepsis, general anaesthesia, planned/emergency surgery and pregnancy for patients with HCM and other forms of ICC.
10. Recognise the importance of sudden death and stroke prevention in HCM and other ICC conditions and the ability to complete risk assessments.
11. Safely prescribe cardiac and non-cardiac medications for patients with HCM, with inherited arrhythmia syndromes, and other ICCs, demonstrating an understanding of pharmacological risks in these conditions.
12. With supervision, recommend and provide counselling for prognostic therapies indicated for elevated risk.
13. Understand indications for pharmacological and invasive EP testing and interpret test results with supervision.
14. Appropriately refer for specialist cardio/genetics advice and present/discuss cases at cardio/genetics MDTs.
15. Demonstrate an understanding of the implications, for mother and baby, of ICCs during pregnancy and the roles of specialists in pre-pregnancy counselling, obstetric cardiology and genetics.

Checklist of Pathologies and Presentations to be Managed Under Supervision

1. **Heart Muscle Disorders**: Hypertrophic, dilated, restrictive, and arrhythmogenic cardiomyopathies.

Key differential diagnoses: Myocarditis, hypertensive heart disease, valvular heart disease, athletic cardiac remodelling, cardiac amyloid, Fabry disease. The key differential diagnoses should be actively discussed and considered; their management is not the focus of this curriculum.

2. **Ion Channel Disorders**: Long QT and Brugada syndromes, catecholaminergic polymorphic ventricular tachycardia and cardiac conduction disease.

Key differential diagnoses include other causes of syncope and arrhythmia in structurally normal hearts with or without repolarisation abnormalities: Drug-induced, athletic cardiac adaptation, autonomic dysfunction, idiopathic VT. The key differential diagnoses should be actively discussed and considered; their management is not addressed by this curriculum.

3. Vascular Disorders: Marfan, Loeys Deitz, Ehlers-Danlos syndromes.

4. Key Presentations:

a. Inpatient assessment and longer-term management following syncope, heart block, acute arrhythmia or acute heart failure in a young adult with an abnormal ECG and/or echocardiogram.
b. Inpatient assessment and longer-term management following an emergency admission with acute cardiac symptoms, elevated serum troponin, and an unobstructed coronary angiogram.
c. Outpatient referral for an unknown cause of sudden cardiac death in a young 1st-degree family member.

Bibliography

Websites

Genomics & genetics educational and reference resources

Genomics England:
https://www.genomicsengland.co.uk/genomic-medicine/understanding-genomics
https://www.genomicseducation.hee.nhs.uk/education/?swoof=1&product_cat=online-courses
www.genomicseducation.hee.nhs.uk/education/online-courses)
UK National genomic test directory: https://www.england.nhs.uk/publication/national-genomic-test-directories/
GeNotes: https://www.genomicseducation.hee.nhs.uk/genotes/

elearning for healthcare:
https://www.e-lfh.org.uk/programmes/genomics-in-the-nhs/
Includes foundation (pedigrees, patterns of inheritance etc.) and specialist courses
www.theaicc.org,

Uptodate:
https://www.uptodate.com/contents/search

NCBI:
Online Mendelian Inheritance in Man: https://www.ncbi.nlm.nih.gov/omim
ClinVar: https://www.ncbi.nlm.nih.gov/clinvar/
ClinGen: https://www.clinicalgenome.org

Specialist societies

The ESC Working Group on Myocardial and Pericardial Diseases
https://www.escardio.org/Working-groups/Working-Group-on-Myocardial-and-Pericardial-Diseases
Includes position papers and consensus documents on cardiac amyloidosis, cardiomyopathies and genetic testing
The Association of Inherited Cardiovascular Conditions
https://www.britishcardiovascularsociety.org/about/affiliates/association-for-inherited-cardiac-conditions-aicc
Includes links to recommended patient management pathways

Clinical Guidelines

The ECS Guidelines on Hypertrophic Cardiomyopathy
https://www.escardio.org/Guidelines/Clinical-Practice-Guidelines/Hypertrophic-Cardiomyopathy
The AHA/ACC Guidelines for the Diagnosis and Treatment of Patients with Hypertrophic Cardiomyopathy
The ESC Guidelines on Acute and Chronic Heart Failure
https://academic.oup.com/eurheartj/article/42/36/3599/6358045?login=false
The AHA/ACC/HFSA Guideline for the Management of Heart Failure
https://www.ahajournals.org/doi/10.1161/CIR.0000000000001063
The ECS Guidelines of Ventricular Arrhythmias and Prevention of Sudden Cardiac Death
https://www.escardio.org/Guidelines/Clinical-Practice-Guidelines/Ventricular-Arrhythmias-and-the-Prevention-of-Sudden-Cardiac-Death
The ESC Guidelines on cardiac pacing and cardiac resynchronisation therapy
https://www.escardio.org/Guidelines/Clinical-Practice-Guidelines/Cardiac-Pacing-and-Cardiac-Resynchronization-Therapy
ACC/AHA Guidelines for Implantation of Cardiac Pacemakers and Antiarrhythmia Devices
https://www.ahajournals.org/doi/10.1161/01.cir.97.13.1325
European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases
https://academic.oup.com/europace/article/24/8/1307/6562982?login=false

Healthcare Professional Medical Education

Annual conference and eLearning Courses - Cardiomyopathy UK Charity: https://www.cardiomyopathy.org/healthcare-professionals

Patient Support Groups

Cardiomyopathy UK https://www.cardiomyopathy.org/
Pumping Marvellous https://www.pumpingmarvellous.org/
British Heart Foundation https://www.bhf.org.uk/
Cardiac Risk in the Young https://www.c-r-y.org.uk/