The Royal College of Pathologists
Pathology: the science behind the cure

National Medical Examiner's Good Practice Series No. 18

Inherited cardiovascular conditions

January 2025

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About the National Medical Examiner's Good Practice Series

Medical examiners are senior doctors providing independent scrutiny of non-coronial deaths in England and Wales, with the role now a statutory requirement since 9 September 2024.

While there is extensive guidance available on a wide range of topics for NHS and public sector staff, the National Medical Examiner's Good Practice Series highlights how medical examiners and medical examiner officers can better meet the needs of local communities and work more effectively with colleagues and partners.

The Good Practice Series is a topical collection of focused summary documents, designed to be easily read and digested by busy front-line staff, with links to further reading, guidance and support.

Introduction

This paper explores good practice for medical examiners providing scrutiny of deaths where the deceased had inherited cardiovascular conditions (ICCs). While some people with ICCs may experience symptoms such as dizziness, fainting or shortness of breath, others may not realise they have an ICC as they do not notice any symptoms.

Sadly, in some cases, the first sign something was wrong is a death from sudden arrhythmic death syndrome (SADS).[1] Sudden unexpected deaths are always referred to the coroner and many other deaths caused by or involving ICCs will also need to be notified to the coroner.

According to the British Heart Foundation, the most common inherited heart conditions are cardiomyopathies and channelopathies.[2]

[1] British Heart Foundation. *Sudden arrhythmic death syndrome*. Available at: https://www.bhf.org.uk/informationsupport/conditions/sads
[2] British Heart Foundation. *Inherited heart conditions*. Available at: https://www.bhf.org.uk/informationsupport/conditions/inherited-heart-conditions

Recommendations for medical examiners and medical examiner officers

Medical examiners and officers should:

- consider factors that may increase the possibility that an undiagnosed heart condition caused or contributed to death and be aware of the full range of ICCs
- consider the possibility of an ICC when a patient has been resuscitated previously but did not make a full recovery and died from another cause
- consider that the family of the deceased and their GP may be able to provide insight into relevant information about family health and any ICCs
- be sensitive to the feelings of next of kin who may be alarmed by the prospect that they or other family members may have an ICC, and that the NHS may need to provide support or counselling, if needed.

If the cause of death is unexplained and, therefore, has been notified to the coroner, but it is suspected the patient had an ICC, medical examiners should, where appropriate, ensure that NHS clinical teams consider:

- alerting the laboratory to save any antemortem samples they may have in case of genetic testing
- making a referral or signposting the family to their local ICC clinic via the local ICC coordinator.

Where a death was notified to the coroner but the coroner decides not to investigate and issues a CN1A, the medical examiner should again ensure that NHS clinical teams recommend ICC testing and counselling to family members, where appropriate.

Context

Heart and circulatory diseases cause around a quarter (27%) of all deaths in the UK; over 170,000 deaths a year, of which sudden deaths account for 25–50%. Each week in the UK, at least 12 young people (aged under 35) die from an undiagnosed heart condition, approximately 624 per annum.[3] An estimated 340,000 people in the UK have an ICC.

ICC is used to describe diseases that affect the heart and great vessels and that are caused by genetic mutations. These can be classified as:

- cardiomyopathies:
  - hypertrophic cardiomyopathy (HCM): the heart muscle becomes thickened, affecting its ability to pump blood
  - dilated cardiomyopathy (DCM): the heart chambers enlarge, leading to weakened pumping function
  - arrhythmogenic cardiomyopathy, in which either right or left ventricle muscle is replaced by scar tissue, causing abnormal heart rhythms
  - idiopathic left ventricular hypertrophy
  - idiopathic left ventricular fibrosis.
- channelopathies (which can cause abnormal heart rhythms and in which the heart is morphologically normal):
  - long QT syndrome (LQTS): prolonged QT intervals in the heart's electrical cycle
  - Brugada syndrome: abnormal electrocardiogram patterns and increased risk of sudden cardiac arrest
  - catecholaminergic polymorphic ventricular tachycardia (CPVT): stress-induced ventricular arrhythmias
  - progressive cardiac conduction defect (PCCD).[4]

[3] British Heart Foundation. *UK factsheet*. Available at: https://www.bhf.org.uk/-/media/files/for-professionals/research/heart-statistics/bhf-cvd-statistics-uk-factsheet.pdf
[4] British Heart Foundation. *Progressive cardiac conduction defect (PCCD)*. Available at: https://www.bhf.org.uk/informationsupport/conditions/pccd

- familial hypercholesterolaemia (FH).
- aortopathies:
  - Marfan’s syndrome
  - Loeys–Dietz syndrome
  - vascular Ehlers–Danlos syndrome
  - aortic dissection in under 40 years old.

Any of these categories may cause a natural sudden death or sudden cardiac arrest (SCA). Cases may be defined and categorised as follows:

- SCA: sudden cessation of cardiac activity with haemodynamic collapse, typically due to sustained ventricular arrhythmia
- sudden unexpected death (SUD): death that occurs within 1 hour of onset of symptoms in witnessed cases, or within 24 hours of last being seen alive when it is unwitnessed
- sudden cardiac death (SCD): SUD due to a cardiac cause.

Cases that are unexplained may be further termed as follows:

- Sudden unexplained death syndrome (SUDS): unexplained sudden death occurring in an individual older than 1 year
- Sudden unexplained death in infancy (SUDI): unexplained sudden death occurring in an individual younger than 1 year with negative pathological and toxicological assessment
- SADS: unexplained sudden death occurring in an individual older than 1 year with negative pathological and toxicological assessment.

While most of these deaths may be unheralded, some deaths may be preceded by symptoms, typically palpitations, syncope, or unexplained seizures and, in the case of aortopathies, symptoms of acute aortic dissection.

Investigating and reporting deaths

The criterion for notifying a coroner applies as set out in the Notification of Deaths Regulations 2019 guidance.[5] If an SUD occurs where the cause is unexplained or uncertain, coroner notification is necessary; the coroner's pathologist will identify the ICC or if the death is a SUDI/SADS death. The coroner's office can then follow the correct pathway and samples will be obtained at post mortem. If appropriate, the coroner's office will ask if family members wish to be signposted for genetic testing and counselling. Medical examiners should also, where appropriate, ensure that clinical teams alert the family to the offer of genetic testing and counselling, to ensure this opportunity is not missed.

If an SCA occurs and the individual dies in hospital and where the cause of SCA is unexplained or uncertain, coroner notification is necessary. The coroner's pathologist will identify the ICC or if the death is a SUDI/SADS death.

- Sudden deaths will be investigated by coroners who have published guidance[6]
- In addition to review by the medical examiner or coroner, deaths in children under 18 years old must be referred to the local Child Death Oversight Panel
- Medical examiners should be alert to certain factors – for example, deaths involving ICCs where the ICCs are frequently missed.

If an SCA occurs and the cause of SCA has been diagnosed by the clinical team as likely to be an ICC, then antemortem samples will normally be preserved and relatives signposted or referred to ICC services via the ICC coordinator, if not already part of the SCA pathway.

For children, please refer to the Good Practice Paper, Medical examiners and child deaths,[7] for more information. If not a coroner referral, then the medical team looking after

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[5] UK Ministry of Justice. *Notification of Deaths Regulations 2019 guidance*. Available at: https://www.gov.uk/government/publications/notification-of-deaths-regulations-2019-guidance

[6] UK Courts and Tribunals Judiciary. *Joint Guidance for Coroners and Coroners' Officers Sudden Cardiac Death: Inherited Heart Conditions*. Available at: https://www.judiciary.uk/guidance-and-resources/joint-guidance-for-coroners-and-coroners-officers-sudden-cardiac-death-inherited-heart-conditions/

[7] The Royal College of Pathologists. *National Medical Examiner's Good Practice Series No. 6: Medical examiners and child deaths*. Available at: https://www.rcpath.org/static/7fa7a9d6-ada5-4597-b16f4602c93d3e91/Good-Practice-Series-Child-Deaths.pdf

the child would be expected to identify the potential of an ICC and apply the same processes described above. Deaths in children under 18 years old must also be referred for a child death review, which would involve the ICC coordinator.

NHS and Coronial Sudden Unexpected Death Programme

Following a pilot in 7 areas in England, the NHS and Coronial Sudden Unexpected Death Programme (NHS-C-SUD) is being rolled out nationally.

Cardiac pathology, post-mortem genetic testing and clinical familial evaluation are the 3 primary components of the programme. The NHS-C-SUD programme provides comprehensive personalised medical management of families who have suffered SUD where an inherited cardiac condition is suspected to be the cause of death.

The aim is to identify a cause of death and diagnose and treat relatives who are also potentially at risk of a genetic cardiac condition. The delivery of appropriate patient pathways requires close working between the NHS and coronial services.

Consideration is being given to the need to incorporate other pathways by which individuals/cases can enter the system, such as if a medical examiner, when carrying out scrutiny, identifies the possibility that the cardiac death could have a genetic cause.

The objectives of the programme are to:[8]

- establish consistent pathology referral practice for SUD including use of expert pathology
- establish routine tissue retention for histopathology and DNA extraction in suitable SUD cases
- establish coronial and NHS communication pathways for referrals of families for genetic testing and clinical evaluation
- establish mechanisms for standardised post-mortem genetic testing and reporting via NHS Genomic Laboratory Hubs to support timely evaluation and cascade testing of families

[8] NHS Central and South Genomics. *NHS and Coronial Sudden Unexpected Death (NHS-C-SUD) pilot*. Available at: https://centralsouthgenomics.nhs.uk/transformation-projects-patients/sudden-unexpected-death/

- develop and disseminate nationally applicable best practice pathways for NHS adoption, employing a toolkit approach
- ensure the engagement and input of patient and support groups with an interest in inherited cardiac disorders.

Conclusions

It is important for medical examiners to consider the possibility that an ICC caused or contributed to death and to be aware of the full range of heart conditions, as well as the sensitivities involved in communicating and informing relatives of any investigation and likelihood of an ICC.

Further information about ICCs can be found in the *Find out more* section of this paper.

Find out more

- Aortic Dissection Awareness UK & Ireland
  - New guide to bereavement due to aortic dissection for families launched.
- Association Of Inherited Cardiac Conditions.
- British Congenital Cardiac Association.
- British Heart Foundation:
  - UK factsheet.
  - Inherited heart conditions.
  - Inherited cardiomyopathies:
    - hypertrophic cardiomyopathy
    - dilated cardiomyopathy
    - arrhythmogenic right ventricular cardiomyopathy.
  - Channelopathies (which can cause abnormal heart rhythms):
    - long QT syndrome (LQTS)
    - Brugada syndrome
    - catecholaminergic polymorphic ventricular tachycardia (CPVT)
    - progressive cardiac conduction defect (PCCD).
  - Inherited conditions that increase your risk of heart disease:
    - familial hypercholesterolaemia (FH) – very high cholesterol levels.
  - NHS and Coronial Service Sudden Unexpected Death Programme Evaluation
  - Sudden arrhythmic death syndrome.
- Cardiac Risk in the Young – Sudden cardiac death.
- Cardiomyopathy, the Heart Muscle Charity.
- Courts and Tribunals Judiciary – Joint Guidance for Coroners and Coroners' Officers Sudden Cardiac Death: Inherited Heart Conditions.
- UK Home Office – Sudden unexpected death: medical investigation

- Central and South Genomics Service – NHS and Coronial Sudden Unexpected Death (NHS-C-SUD) Pilot.
- NHS England – The Sudden and Unexpected Deaths in Childhood (SUDC) Service – a nurse-led model of care and support.

Acknowledgements

This document was drafted following circulation to and input from the following people. The National Medical Examiner is grateful to all for their participation and support:

- Dr Alan Fletcher, National Medical Examiner, NHS England (Chair)
- Anne Marie Aherne, Deputy Head of the Chief Coroner's Office
- Elijah Behr, Professor of Cardiovascular Medicine at St George's University Hospitals NHS Foundation Trust
- Helen Briggs, Office Co-ordinator to the National Medical Examiner Office, NHS England
- Dr Graham Cooper, Regional Medical Examiner, NHS England
- Dr Frances Cranfield, Royal College of General Practitioners
- Nick Day, Policy and Programme Lead, Medical Examiner System, NHS England
- Douglas Findlay, Lay Representative
- Dr Martin Goddard, Cardiac Pathologist, Royal Papworth Hospital NHS Foundation Trust
- Natalie Harris, Healthcare Standards and Governance Lead, Welsh Government
- Andrew Leatherland, Senior Programme Manager, British Heart Foundation
- Dr Suzy Lishman, Senior Advisor for Medical Examiners, Royal College of Pathologists
- Judy O'Sullivan, Director of Innovation and Health, British Heart Foundation
- Graham Prestwich, Lay Representative
- Helen Rose, Senior Project Manager, Medical Examiner System, NHS England
- Jonny Tinsley, Head of Data and Health Analysis, Office for National Statistics
- Professor Mary Sheppard, Cardiac Pathologist, St George's University Hospitals NHS Foundation Trust, City St Georges, University of London.