

Delivering better care for families with inherited cardiovascular conditions

“...it is no longer acceptable for someone at risk of an ICC not to receive timely, expert attention” - Prof Peter Weissberg, British Heart Foundation



Inherited cardiovascular conditions (ICCs) are a group of genetic disorders that primarily affect the heart, including cardiac electrical systems and blood vessels. Sometimes, the first indication that an ICC is present in a family is the sudden and unexpected death of an apparently healthy child, teenager or young adult; other relatives may also be at risk. Recent advances in genetics and cardiology are now making it possible for health professionals to offer much more accurate diagnosis, improved clinical management, and better risk assessment for families affected by ICCs - avoiding tragic and unnecessary deaths. Steps are now needed to ensure that speedy and equitable access to these services is made available throughout the NHS.

Focus on PHG Foundation report
Heart to Heart: Services for Inherited Cardiovascular Conditions

What are ICCs?

More than 50 different ICCs have been recognised so far, with diagnostic tests increasingly available. This group of conditions is highly variable and includes some relatively common disorders such as familial hypercholesterolaemia (very high cholesterol levels) and hypertrophic cardiomyopathy, as well as some much rarer diseases, such as long QT syndrome. The conditions are highly variable in terms of the underlying genetic mutations that cause them and the resulting symptoms and clinical features. Together, inherited disorders that involve the heart affect around 340,000 people in the UK.

How is genetics changing care?

In the past, most people with cardiovascular problems were cared for primarily by cardiovascular specialists. Even when a genetic cause was known or suspected, it had little impact on clinical management. However, in the last ten years, emerging understanding of the genetic basis of many of these conditions has led to the development of new genetic tests that can not only aid diagnosis, but also in some cases inform treatment and preventative care for patients and families. Management may be more effective if begun in childhood.

Identification of a mutation in an affected person means that their relatives can be offered testing for the same mutation, an approach called **cascade testing**. Any

found to have the same mutation can be offered medical assessment and appropriate treatment or surveillance. Equally, relatives who do not have the mutation can be reassured that they are not at risk.

What does proper care include?

Besides the advances in cardiovascular genetics, in recent years there have also been major developments in electrophysiology, echocardiography and imaging techniques. There is an increasing need for specialists in different techniques in cardiology *and* genetics to work together to deliver the best care.

For many ICCs, careful clinical management can reduce risk. For arrhythmia syndromes (such as long QT syndrome) and hypertrophic cardiomyopathy, treatments may include a variety of drugs, lifestyle advice to avoid triggering events, fitting of implantable defibrillator devices or in some cases, surgical options. Familial hypercholesterolaemia can be successfully treated with statins.

Genetic testing can enable more accurate diagnosis to guide management. For example, different genetic causes of ICCs that present with similar physical symptoms may have very different sorts of ‘triggers’ that can cause a cardiac arrest (heart attack), which patients should avoid. For one ICC, this might be high-intensity exercise, whilst for another, it might be sudden loud noises such as audio alarms.

Geneticist’s perspective

“Together, inherited cardiac conditions are a common cause of preventable ill-health and sudden death, often in young people. The PHG Foundation’s report provides an invaluable and timely survey of current clinical and laboratory services for these people and their families....despite apparent current deficits in service provision, the NHS is capable of providing the most comprehensive, equitable, patient-centred network of specialist services for inherited cardiac conditions in the world”

- Professor Paul Brennan, Consultant in Clinical Genetics, Teesside Genetics Unit

What is this new report?

The new PHG Foundation report is the product of an expert Working Group, which included NHS cardiologists and geneticists, service commissioners and managers, along with representatives from several key charities:

- British Heart Foundation
- Cardiac Risk in the Young
- Cardiomyopathy Association
- Marfan Association UK
- Sudden Arrhythmic Death Syndromes UK

In addition to reviewing the current status of new and emerging knowledge and technologies for the accurate diagnosis and improved management of ICCs, it also examines wider issues that need to be addressed if patients and families are to receive optimal care within the NHS.

Patient perspective

“These tragedies with their genetic implications are catastrophic for families”

- Alizon Cox, Chief Executive of charity Cardiac Risk in the Young (CRY)

Findings - current services

A survey of NHS providers of services for ICCs revealed that capacity is insufficient to meet current needs in the UK, let alone the anticipated growth in need over the next few years. Serious inequalities were identified in both the quality and quantity of services available across the country. Service provision in London is six-fold higher per capita than elsewhere in the country.

An independent survey of lipid clinics serving patients with familial hypercholesterolaemia, carried out by HEART UK also observed wide national variation across the country, and highlighted inadequate specialist genetics input and poor provision for the management of children and young people.

Features of a specialist service

The report proposes that a specialised service for ICCs must be able to cater for the full range of conditions in both adults and children in a timely fashion; it should include cardiologists, geneticists and expert nurses, along with access to the latest laboratory and pathology services and other key investigations such as imaging, electrophysiology and exercise testing. Integration of these different elements to provide a genuinely multidisciplinary specialist service is critical, as is increased awareness of this service among wider health professionals such as GPs. Bereavement help, counselling and integration of services with voluntary organisations that provide support for families are also essential elements.

Ethical and legal issues

When an ICC is suspected in a living or deceased patient, there are some important barriers to the delivery of effective care. For example, there is a lack of clarity over tissue samples taken under the Coroner’s authority for investigating the cause of a sudden unexplained death. The default situation under the Human Tissue Act is that samples are destroyed once the Coroner’s authority lapses, unless family members consent to ongoing retention. However, they will not necessarily be aware that the death could have been from an ICC, and that testing could inform the risk to other family members.

Department of Health

“We welcome this report and will be working closely with the NHS, local healthcare services and charities, including PHG Foundation, to develop these specialist inherited cardiac services and address issues surrounding access”

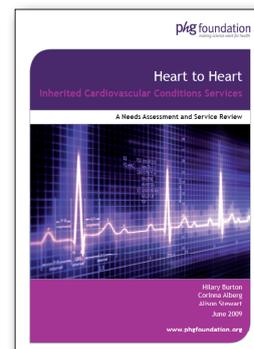
- Professor Roger Boyle, National Clinical Director for Heart Disease and Stroke

Key recommendations

The Working Group concluded that **every UK cardiac network should ensure that its population has access to specialised expert ICC services for children and adults**, although most will not have their own service. Other key recommendations included:

- Experts should develop standards for specialist ICC services, including required skills and facilities, expected activity, organisational, research and audit systems.
- Cardiac networks should ensure that ICC services meet agreed minimum standards within an appropriate timeframe.
- Requirements for nationwide access to adequate ICC services should be agreed, anticipating a steady increase in demand within the next 5 to 10 years.
- ICC service providers should work with professional organisations, regulatory bodies and educational providers to develop a workforce with the necessary specialist competences.
- ICC services should work with genetics services to develop and evaluate systems of cascade testing, including IT systems that can link individuals within families.
- Experts from ICC services should keep abreast of emerging knowledge and technologies, and work with patient groups and other organisations on research programmes to evaluate them.
- The UK Genetic Testing Network (UKGTN) should ensure effective, efficient and equitable genetic test provision in the light of current and future developments in testing technology.
- Systems should be developed and legislative changes sought to ensure the appropriate retention of samples following sudden cardiac death, the promotion of best practice regarding consent and data sharing within clinical practice, and effective approaches to cascade testing.
- The responsibility of coroners to family members who may be at risk of sudden cardiac death requires clarification and strengthening.
- Experts within ICC services should maintain an overview of emerging tests and knowledge and work with patient groups and other relevant organisations to ensure that translational research programmes are set to evaluate them.

The full report, *Heart to Heart: Services for Inherited Cardiovascular Conditions - a needs assessment and service review* and executive summary are available for free download from the PHG Foundation website:



www.phgfoundation.org

The PHG Foundation is an independent non-profit policy research and development organisation working to achieve better health via the responsible and effective application of genomic science and technology in health services in the UK and beyond.