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Cardiac Genetics Service Review

Background

There are many different forms of rare inherited disease that cause sudden cardiac death, arrhythmias and other cardiovascular conditions. In addition, a substantial number of other genetic diseases that affect different parts of the body are also associated with cardiological abnormalities. Together, individuals with these diseases represent a significant component of cardiac services in the NHS.

Surveys of service provision in cardiac genetics in 2006 showed variation across the country, which led to a request from an informal United Kingdom Cardiac Genetics Network that the PHG Foundation (formerly the Public Health Genetics Unit) lead an expert Working Group in a comprehensive needs assessment and review of cardiac genetics services. The group will also develop a service specification and commissioning framework for the genetics elements of these services

Key aims

1. To make recommendations for the development of genetics elements of cardiac genetics services which meet the requirements of service commissioners and providers, health care professionals, and patients and their families

2. To strengthen and complement the service developments arising as a result of the implementation of Chapter 8 of the National Service Framework for Coronary Heart Disease.

3. To inform a strategy for the development of cardiac genetics services in the UK and develop a programme for its implementation.

Project Scope

This work will look at the provision of services for patients and families with Sudden Cardiac Death (SCD) and Inherited Cardiac Conditions (ICCs), and of national cascade testing for Familial Hypercholesterolaemia (FH).

Both single gene disorders that primarily affect the heart and those that increase an individual's risk of cardiovascular disease are included in the scope of this project; for example, hypertrophic cardiomyopathy (HCM), familial hypercholesterolaemia (FH) and Marfan syndrome. However, common multi-factorial cardiovascular disorders (such as coronary artery disease) and their risk factors (such as polygenic hyperlipidaemia) are specifically excluded.

Biomarkers for cardiovascular disease risk estimation

The PHG Foundation is running a separate project looking at the genetic risks of coronary heart disease (CHD) which, unlike cardiac genetic diseases, is a common multifactorial condition.

The risk of an individual developing CHD can be estimated using knowledge of risk factors such as blood pressure, cholesterol levels and smoking. However, there are several models based on these risk factors, and it is not clear which performs best in contemporary British populations. It is also uncertain the extent to which genetic information and novel biomarkers could improve the accuracy of risk estimation.

This project aims to identify the best available methods to estimate risk and define those at higher risk of CHD, to explore whether new knowledge of sources or markers of risk can improve risk estimation and to consider the implications for cardiac services and public health.

Objectives

This extensive project includes review of the epidemiology of relevant single gene disorders and of the availability, evaluation and use of clinical and molecular tests and evidence-based clinical guidelines for these disorders.

Pertinent ethical and legal issues will be considered, particularly those surrounding the sharing of genetic data and obtaining and storing human tissue.

The key components of cardiac genetic services and related elements in primary and secondary care, pathology and coroner services will be set out, and a review of specialist genetic input into cardiac services and cardiac input into genetics services performed. Current and future demand for these services will be described along with patient perspectives, and the potential impact of recent and future technological developments considered.

Survey

A survey has been carried out to identify the main providers of specialist cardiac genetics services in the UK, and to determine the services offered, typical workloads, associated genetic testing and service shortfalls. Services providing specialised diagnosis and care for patients and families with suspected inherited cardiac conditions were identified through approaches to regional genetics centres and the clinical cardiac network (in England). Quantititative and qualitative information was gathered about the services, including comparative data on staffing, clinic provision and patient throughput, care pathways, and clinical guidelines used, and the criteria and mechanisms for agreeing genetic tests.

Participants

This project involves a range of experts and key stakeholders, including clinical geneticists, cardiologists, electrophysiologists and other health professionals, health service commissioners and managers, along with representatives from organisations including the British Heart Foundation, the Cardiomyopathy Association, the Arrhythmia Alliance and the Marfan Association UK.

The findings and recommendations of the Working Group to the United Kingdom Cardiac Genetics Network will be available in 2009.

The PHG Foundation is an independent multidisciplinary policy research organisation working to achieve better health via the responsible and effective application of genome-based science and technology in health services.

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