Mending Broken Hearts

Tools to diagnose people with life-threatening inherited heart conditions are now available, thanks to blue-skies research and motivation to change medical practice by Professor Hugh Watkins from the Department of Cardiovascular Medicine at the University of Oxford.

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With more than 100,000 affected individuals in the UK, hypertrophic cardiomyopathy (HCM) is a relatively common heart disease. HCM is the most common cause of sudden cardiac death in children and young adults and, because it is a genetic disorder, it can affect whole families with sometimes more than one family member suddenly dying.

The inner workings of the heart were not fully understood when Professor Watkins started his research on HCM. In the 1990s, genetic mapping of the disease became possible; from his investigations Professor Watkins found that the genetic mutations associated with the disease caused the motor proteins of heart muscle to misalign. HCM was the first heart condition to be understood genetically and the revolutionary understanding of the disease, combined with the ability to genetically detect it, changed national and international medical guidelines. It is now recommended that genetic testing is considered as a standard aspect of care for families suspected of having HCM. This is one of the first examples of high throughput genetic testing in use in the NHS and more than 2,000 families have now been screened at the Oxford Radcliffe Hospital's NHS genetics laboratory.

HCM is a difficult disease to treat because it is caused by a mechanical problem in the heart which makes the heart inefficient. However, now the specific problem has been identified, there is more chance of developing a cure. Professor Watkins is currently working on developing drugs or methods to lower the risk of death, improve quality of life for sufferers or even prevent people who carry the genetic defect from ever developing the condition.

"Professor Hugh Watkins has led the way in finding the genetic mutations that cause one of the most common – and potentially life-threatening – inherited heart conditions: hypertrophic cardiomyopathy."

Professor Peter Weissberg, Medical Director, British Heart Foundation

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