

Genetics and Hypertrophic Cardiomyopathy

Who should have a genetic test for Hypertrophic Cardiomyopathy?

Hypertrophic cardiomyopathy (HCM) occurs in approximately 1 in 500 adults and is the most common genetic cardiac disease. Almost 2/3 of patients have a family history and HCM is inherited in an autosomal dominant fashion which means that an affected person's children are generally considered to be at 50% risk of being affected. Approximately 1/3 of cases have no previous family history of hypertrophic cardiomyopathy.

HCM shows a high degree of variability particularly in age of onset and clinical severity. Over 150 different variations (mutations) have been identified in ten different genes which makes detection of a mutation in an affected individual extremely difficult. However some research laboratories do offer screening in some of the most frequently involved genes and once a mutation has been identified in a family other at risk individuals in that family may be offered testing to see if they are at increased risk.



Stephanie, a specialist heart nurse, lives in Surrey with her teenage son James. When she was 26 years old she had a cardiac arrest (her heart temporarily stopped beating), after which she was diagnosed with Hypertrophic Cardiomyopathy, an inherited disease of the heart muscle.

Stephanie's condition was diagnosed after doctors carried out an echocardiogram (ECHO) - an ultrasound scan of the heart - and noticed slight thickening of the heart muscle.

'It was a shock to everybody, this was the first presentation in the family. The diagnosis was made clinically -there wasn't a gene test available then - then we heard it was hereditary, and it could be passed on to my son, who was two and a half...obviously I did care about myself, but I was more concerned about the same thing happening to my son.'

Following her diagnosis, Stephanie was at first given tablets to prevent another cardiac arrest, but seven years ago, her medication was replaced with an implantable cardioverter defibrillator (ICD) and a pacemaker. People with a heart muscle disorder can have an ICD put into their bodies which, if their heart beat becomes irregular, delivers shocks to restore a normal rhythm. Stephanie says she has been fit and well since having hers fitted:

'I haven't had a cardiac arrest - I've had a few palpitations and things, but I'm OK, I can live my life now how I like....knowing that

with this ICD, if anything happens to me I'll be OK, which is great, because I know that I'm at high risk of sudden death...I can live a normal life without taking tablets, without restricting my lifestyle'.

Stephanie's main concern was to find out if her son was at risk of the same condition, before he reached puberty, when he might start to show symptoms. The genetic change responsible for Hypertrophic Cardiomyopathy in Stephanie's family was identified when James was six, and the family were offered genetic testing as part of a research trial:

'If the gene had been available when he was two and a half, then I would've had him tested, because I was worried from day one. I didn't realise how worried I was until I had the results of the genetic test, and he was negative. And the stress flew out the window'.

James' test result means that he no longer has an increased risk of developing heart problems, and so will not need to have regular electrocardiogram (ECG) or ECHO screening.

'That would be the option for him, had he had the gene. He's 15 now, so he would be being looked at every 6 months if he had it. In fact he was screened - he did have an ECHO, he did have an ECG - until we had the genetic results'.

Stephanie says that although having hypertrophic cardiomyopathy changed her life, she doesn't live every day as though it was her last. And as for getting an early, accurate diagnosis she says:

'It's got to be good. In families where people are at high risk of sudden death, it's good because there is an intervention.'