DIAGNOSING
FAMILIAL HYPERCHOLESTEROLAEMIA

British Heart Foundation

Familial hypercholesterolaemia (FH) is a genetic condition affecting approximately 1 in 250 people, resulting in high cholesterol levels from childhood and a high risk of early heart disease.

As the high cholesterol levels occur at a very young age, it is not unusual for a person with FH to have a heart attack as early as in their 20s or 30s. Children have a 50 per cent chance of inheriting the condition if one of their parents has FH. In the UK, FH affects an estimated 250,000 individuals, the majority of whom will not have been diagnosed. BHF-funded research carried out by BHF Professor Steve Humphries at UCL, and supported by BHF-funded research at Queen's University Belfast, has led to the identification of specific genetic mutations that cause FH, and to the development of effective genetic tests. Thousands of people are now accessing genetic testing through BHF-funded testing schemes.

Impact

BHF Professor Steve Humphries has dedicated his life to uncovering the causes of familial hypercholesterolaemia- a dangerous genetic condition that greatly increases the risk of premature death from heart disease. Thanks to Professor Humphries' research, and to BHF-funded nurses managing genetic testing services around the country, thousands of people have been diagnosed, and many lives have been saved.

More work is needed to ensure that access to FH services becomes a national priority and everyone eligible for genetic testing is able to seek support.

FIGHT FOR EVERY HEARTBEAT

bhf.org.uk

DIAGNOSING FAMILIAL HYPERCHOLESTEROLAEMIA

1980s-1990s

BHF Professor Steve Humphries collects DNA samples from hundreds of FH patients to investigate the genetic mutations causing their disease

1993

Between 1993 and 2013, the BHF provides over £6m of funding for Professor Steve Humphries to lead research into the genetic causes of cardiovascular disease – around £2m of this is for research into familial hypercholesterolaemia (FH)

1996

A database of all the genetic mutations linked to FH is curated by the UCL Cardiovascular Genetics Group

1996

Researchers at Queen's University Belfast identify a new genetic mutation linked to FH in the Northern Irish population

1997

A genetic diagnostic service is established at Great Ormond Street Hospital

1999

Researchers from Queen's University Belfast publish a paper describing improved methods for FH testing based on their research into mutations causing FH in the Northern Irish population

1999

The BHF provides almost £90,000 for further study of the genetics of FH in the Northern Irish population

2000

FH is shown to be under-diagnosed, particularly in young adults where early treatment with statins could be highly beneficial

2002

Cascade screening, whereby relatives of people diagnosed with FH are tested, is shown to be the most cost-effective method for testing for FH in the population

2004

Ethical issues surrounding genetic testing are overcome – genetic tests are shown not to affect patients' perceptions of control over their condition

2005

Researchers at Queen's University Belfast publish an updated version of the Northern Irish methodology paper, incorporating new genetic mutations linked to FH

2008

NICE guidelines are updated to include genetic and cascade testing for FH cases

2009

FH testing is included in the NHS Vascular Checks guidelines

2010

A testing kit is developed that can quickly and easily identify 20 different types of genetic mutation known to cause FH

2010

A national audit led by BHF Professor Steve Humphries shows that NICE guidelines for FH testing have not been widely adopted, the resources for genetic and cascade testing have not been provided, and people with FH still remain undiagnosed.

2010

The BHF invests £450k over a 3-year period in developing an FH cascade testing model in Wales.

Research · Funding · Medical Milestone · Impact

2011

Over 3,000 genetic tests for FH are performed in labs across the UK

2013

Further research increases understanding of FH with no discernible genetic cause, offering the potential to further improve diagnosis in the future

2014

The Welsh cascade testing service is well established – genetic testing is completed for 1,400 new FH patients

2014

The BHF invests a further £1m in setting up cascade testing services across England and Scotland (8 NHS Trusts receive funding for FH nurses)

2015

The BHF funds a national register of children with FH, directed by BHF Professor Steve Humphries, to monitor the effects of treatment on growth and puberty, support health audits, and provide anonymised data for further research in the field

2016

Hundreds of people have been diagnosed with FH through BHF-funded testing schemes across England and Scotland

Research Funding Medical Milestone Impact