

**Patient experience of Familial
Hypercholesterolaemia (FH) within the all-Wales
cascade genetic testing programme**

Common disease in UK

1:500 within UK population (Heterozygous FH)

Wales 6 000

Estimated that 80% do not know about FH

Dominant inheritance: 50% among 1st degree relatives

Untreated: > 50% risk of CHD by 50 years of age (men)

> 30% risk of CHD by 60 years of age (women)

Also can lead to sudden death before 40 years of age in men

Treatment with statins – risk becomes that of the general population

2003 White Paper 'Our Inheritance, our Future'

'This programme offers a paradigm for how to use genetics to improve healthcare'

Department of Health 2005

Pilot studies inc South Wales

NICE guidelines 2008

Advised cascade testing using genetic testing

Treatment should begin at 10 years of age

'The use of a nationwide, family-based, follow-up system is recommended to enable comprehensive identification of people affected by FH'

NICE guidelines August, 2008

2009: Welsh Assembly Government agreed to fund FH testing programme in conjunction with the British Heart Foundation

Specialist nurses and genetic counsellors appointed

Database established to manage the cascade

programme: genetic test results

who has been contacted

who has replied

who requires follow-up

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Heart attack risk test rolled-out

A screening programme will be rolled out across Wales to identify people at risk of early heart attacks due to high cholesterol.

Around 5,000 people in Wales are estimated to have Familial hypercholesterolaemia (FH), which can lead to heart attacks in young people.

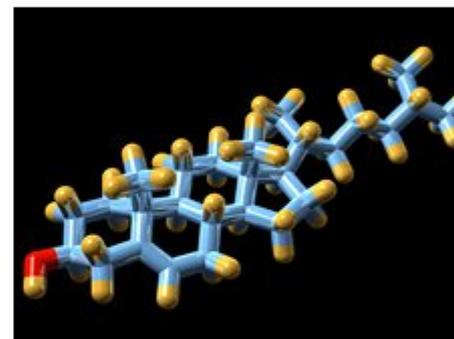
The assembly government and the British Heart Foundation have brought in a testing programme for people at risk.

It is hoped it will improve the quality of life for people with FH.

The screening programme will be rolled out across Wales later this year, funded by the assembly government in what it claims is a first for the UK.

Health Minister Edwina Hart said the tests would also help to reassure the families of people with the condition.

"It is important that we are able to identify people early to give them the help and support to enable them to live life to the full," she



High cholesterol can cause heart attacks if not managed



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Cascade testing: a method for identifying people at risk of a genetic condition by a process of family tracing

Family tree from initial diagnosed patient

1st Degree relatives identified

Patients can then contact their relatives and advise them to be tested - the clinic can also give them information to pass on to their relatives

Depending on where the relative lives, they can then go to their GP or the specialist clinic where the initial patient was identified

If the patient prefers, the clinic can contact the relatives directly and advise them to go for testing at the specialist clinic or GP

Semi structured interviews – approx 1 hour in a site of the patient's choice – hospital, their home, my office or their office

Patients recruited through the specialist clinics

Diagnosed > 6 months

Age: 18 – 70

South Wales: 7

North Wales:6

Interview questions developed through : literature review

clinic observation

attending FH support group

discussions with clinic staff & database coordinator

Augmented after initial interviews when new issues were raised by interview subjects

Interview Themes

GP problems

Particularly in young people – refusal to perform test

‘why are we testing the cholesterol of a 20 year old....and within 24 hours I had a phone call, can you come down straight away, my cholesterol was about 13, and all of a sudden they were interested’

Refusal to believe cholesterol results

She ‘looked me up and down and she said there’s no way your cholesterol is 11’ ‘I’ve never seen one that high..you’ll be one of the highest in North Wales’

Male: heart attack aged 43

‘I’ve always had a clean bill of health from me GP saying I was fit and all that. And really I wasn't to be honest. I had this underlying problem.’

Genetic Test

‘it didn’t make feel differently about FH’ but

‘I was surprised when, I was quite upset when they said, yeah, we’ve found the gene...it was a little bit upsetting, even though I’ve been living with it for 8 years’

‘the reason I want the genetic test was for me to know for future generations’

‘it’s quite tangible, isn’t it, because to say to people, I’ve got this disease rather than, oh I might have or I might not’

‘it was my way of going to the doctors saying I’ve got it, I had to convince them and I shouldn’t have to do that’

‘so I think it’s helped me a little bit to accept that, to say – well this is the reason, you know’

Cascade testing process

‘From a family point of view, it’s important to make sure that everyone gets the right medicine and to try and control the condition’

‘I spoke to them directly and said they were happy to be contacted by one of the researchers to explain to them what was involved’

‘no problem, we’re not hiding anything, you know if it helps in research’

‘I phoned them all up first anyway and warned them and they said yeah. No trouble’

'I should have thought that's fairly straightforward isn't it? They want to live and they want their children to live'

'I was met with complete apathy [from 2 sisters] and I was a bit disgusted with them really. I said you know it's quite important so yeah, well they just bumped me off and that's it'

'it's very much a latent thing..which is what my sisters are probably thinking, you know he looks alright...you know he hasn't changed or anything'

Testing of children

'how do you bring up a child to say right, when you're 10, you're going to have this test, you might have[it], you might not'

'if they have siblings then how do you deal with it if one of them has it and one of them hasn't'

'I think catching [them] before the whole smoking and drinking thing. I think that would be a key factor in catching them at 10'

Daughter-in-law (pharmacist)

'she said oh no they're much too young, they don't need to, blah blah, so rather than get bombarded in arguments with her, I just let it go because it was the hospital that said they'd do it then'

Genetic counselling

‘do you mean in a sort of emotional support?’

[explanation of role given]

‘I don’t think that would be particularly useful’

‘No, I’ve never thought it would be useful personally...I think I understand what’s involved and I’m comfortable with it’

‘I’m not sure I’ve necessarily been offered the services of one, but I probably would have declined anyway’

‘it would have been...if we’d all just been diagnosed, clearly that would have been important’

‘I can think of all the things that would have been helpful at different stages in my life and I’d like to think that soon all those things will be in place but for me they weren’t’

[Diagnosed 30 years ago]

Insurance

Genetic diagnosis 'will make things easier' - previously 'saying well it's a potential FH, with a genetic diagnosis I think it will make things a bit clearer on that side as well'

'It's a grey area, isn't it, because when you do the life insurance policies now, there's no option for FH and ...they say have you got high cholesterol, and they write high cholesterol down but you can have high cholesterol and not have FH, they're different things'

‘when you do your insurance on the website, it does ask you about your cholesterol if it’s FH or not, so it obviously does make a difference’

‘a little bit of one of my concerns as regards to my DNA’s now on the system ...in the future will they – these general insurance companies be looking at people’s DNA and saying oh, he’s got a defective gene; we’ll give him sort of a loading on simple car insurance or something like that’

Dietician

‘I eat a pretty healthy diet, I exercise regularly and her advice was avoid pies and pastries and that was it’

[aged 20 – had been vegetarian since 11 years old]

‘the thing is you can’t teach an old dog new tricks, you know I was 50 when they tried to change my diet’

‘I think it would have been useful...if you’ve got children you need to ask questions like actually well they need a certain amount of fat in their diet, how much do they need?’

After seeing dietician, his 'wife said they didn't believe us' 'they just thought I was saying what I thought they wanted to hear which wasn't the case'

'basically my wife, she's fully aware of the condition. She wants to keep me going as long as possible so we do eat healthy'

A lot of negotiation with GPs regarding FH especially when young people involved

Several instances in the past when diagnosis made because someone in family worked in healthcare

Newly diagnosed patients had better experience of FH

Family cascade testing not problematic in most cases

Timeline:

2003 White Paper

2005 Pilot studies

2008 NICE guidelines

2009 Cascade testing begun in Wales



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