



Improving healthcare for families with genetic conditions

Preventing heart attacks due to inherited high cholesterol.

Can NHS Wales lead the way?

What is inherited high cholesterol – familial hypercholesterolaemia (FH)?

- It is a common but potentially fatal condition. It is passed on from parent to child with a 1 in 2 chance of inheritance.
- FH is characterised by high cholesterol levels in the blood which causes premature coronary heart disease (CHD) leading to ill-health and premature death. Young adults with FH are at particularly high risk.
- In Wales, it is estimated that there are **6000 people affected with FH** (1 in 500 people). Currently, only 1200 are known, meaning **4800 people** have no idea they have this genetic condition and are not receiving any management or treatment.
- There is no Wales-wide service in NHS Wales working to identify and diagnose those affected. They are often only identified after developing CHD and suffering a heart attack. **People with FH need to be identified early, before they develop CHD and whilst they are still in good health.**

What needs to be done?

- A **nurse led 'Cascade Screening' (family tracing)** system starting from known cases is an effective and inexpensive method to identify and diagnose people and families with FH using cholesterol & DNA testing. Once diagnosed, individuals can be carefully managed and treated effectively and inexpensively and stay in good health.

The evidence & support

- A Cascade Screening pilot project based in Bridgend & Cardiff involving a specialist nurse is identifying and diagnosing people and families with FH. This project is very successful and has the potential to be rolled-out across Wales, so that all who are at risk of this condition may benefit from being diagnosed and treated,
- **NICE guidelines** for the diagnosis of FH supports cascade screening services and its health economics analysis is also (final recommendations will be published in August 08, they are currently open to consultation).
- The **National Service Framework for Coronary Heart Disease** supports a cascade screening service for FH diagnosis.
- **Families** within the South Wales FH Forum support the establishment of a service that will diagnose their relatives and other families like their own.
- The **Cardiac Networks Coordinating Group in Wales**, the **Medical Genetics Service for Wales**, the **British Heart Foundation**, **HEART UK & Genetic Interest Group**, and many other professional groups support the establishment of a Wales-wide service for the diagnosis of FH.

The Solution

- A Cascade Screening Service needs to be commissioned across Wales.
- The establishment of a national team of 6 nurses and 2 support staff would potentially **prevent heart attacks for the 4800 people across Wales** who currently have no idea they are affected.
- **This population group is at high risk of developing CHD and premature death, which if diagnosed is completely preventable.** NHS Wales needs to address this inequity so that families with FH do not suffer adversely.

Key Asks

- Funding for 6 specialist FH nurses on an all-Wales basis.
- Funding for a project manager and a data base administrator.
- Support for development of patient information and implementation of IT systems.
- Laboratory funding for genotyping

How can Assembly Members help?

- **Please support the Statement of Opinion by Janice Gregory AM to support families with Familial Hypercholesterolaemia. OPIN-2008- 0026**

For further information, please contact:

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The Genetic Interest Group

- **Working to benefit all people affected by genetic disorders.**

The Genetic Interest Group (GIG) is a national alliance of organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders. Its primary goal is to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them.

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