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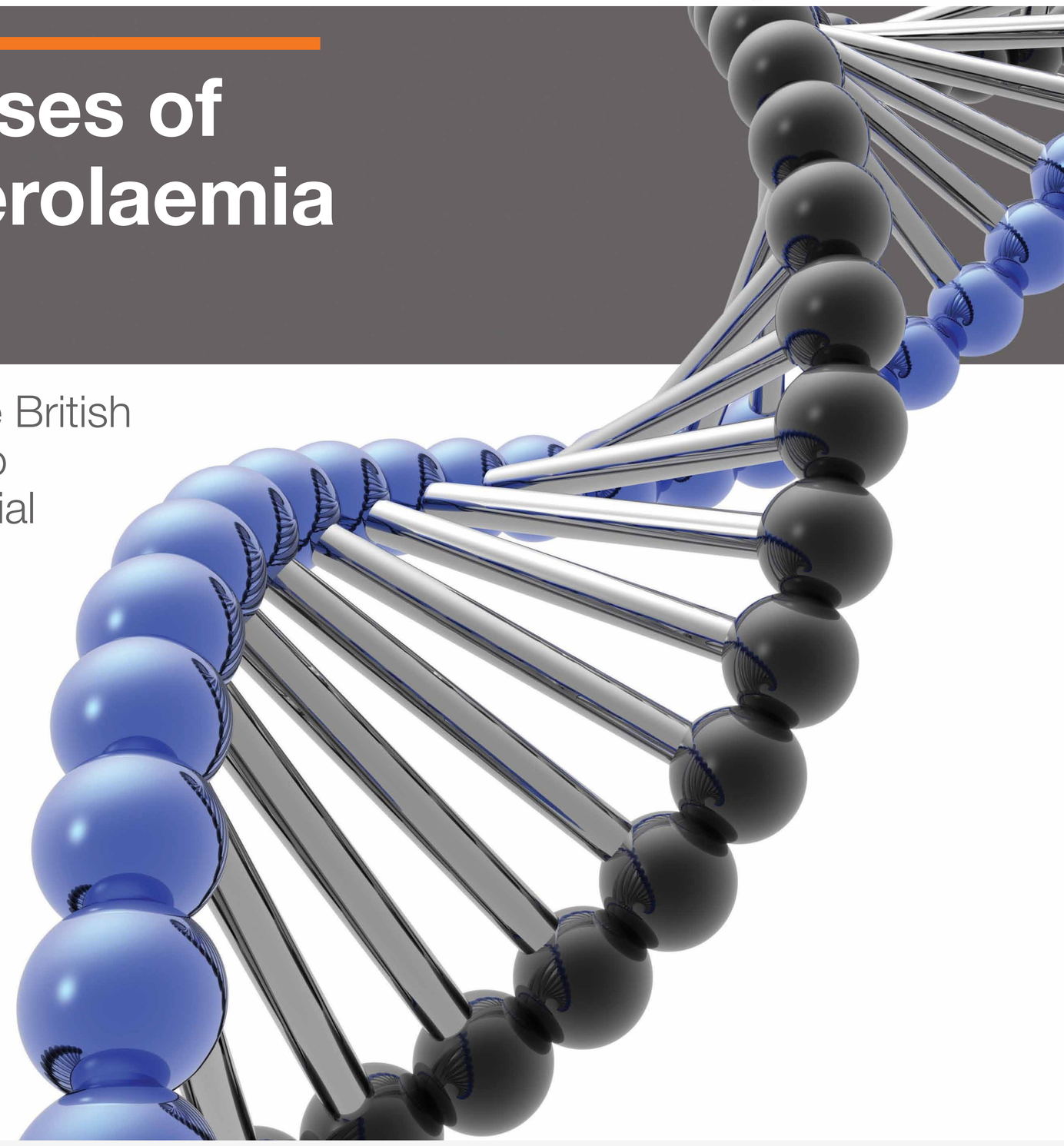
Finding the hidden cases of familial hypercholesterolaemia in Wales



University Hospital Wales, supported by the British Heart Foundation, implemented a service to diagnose individuals and families with Familial Hypercholesterolaemia (FH) across Wales. The service was based on the NICE FH clinical guideline. Day to day delivery is carried out by British Heart Foundation specialist nurses and genetic counsellors working with primary care, hospital physicians and laboratory staff, working closely with a family support group.

Everybody's problem but nobody's problem

FH is a single gene disorder affecting 1 in 500 of the population. It causes high blood cholesterol levels which can lead to premature coronary heart disease.

Prior to this service being set up, individuals were suffering early onset coronary disease and nothing was being done to prevent this in other family members.

The problem was that FH was being missed because it fell between professional stools and there was lack of clarity and focus about the service that was clinically needed. Lipid clinic provision was patchy and uncoordinated, and Wales had no system for family cascade testing.

It was everybody's problem but nobody's problem. Cardiology teams would treat the premature coronary disease if the patients reached them, but not recognise the cause. Physicians and medical biochemists would treat the cholesterol, but not make a genetic diagnosis or be able to follow up families, and GPs and paediatricians would not be aware of the condition and would not think of it in younger people.

NICE guidelines a blueprint for change

The NICE guideline on FH, published in 2008, gave a blueprint that we could follow for the service that was needed in Wales. The strong clinical and economic evidence base from the NICE guideline was the foundation and focus for this service implementation.

We identified the need for a multidisciplinary service that had local and regional components. The local components were in primary care (case recognition) and secondary care (specialist diagnosis). The regional components were DNA diagnosis and the family cascade testing.

We demonstrated feasibility by carrying out pilot projects on DNA testing and the role of FH specialist nurse. We also learnt lessons from a successful service in the Netherlands and in collaboration were able to procure and adapt a bespoke IT system for FH and cascade testing.

In October 2010, we launched a service for FH. The FH database now includes 1,147 individuals with possible FH. Of these, 471 have received a DNA diagnosis. Cascade testing from index patients using DNA testing has led to 222 relatives being diagnosed with FH and 181 being reassured that they do not have the condition.

Multi-professional support needed

The need for multi-professional support was identified as a barrier to the successful rollout of the FH service.

To overcome this, we engaged clinical stakeholders across a range of professional and managerial organisations by setting up an "FH special interest group" under the oversight of Cardiac Networks Coordinating Group.

A dedicated "ambassador" for the service met all the key stakeholders and worked to define the precise clinical requirements by focusing on the patient pathway for individuals and families.

Working across professional boundaries can be challenging. Early communications, groundwork and repeatedly making the clinical and economic case in a range of professional settings were essential to bringing clinicians and others on board.

Working in partnership with 'third sector' organisations such as British Heart Foundation, Heart UK and the Genetic Alliance has also been very positive not just for funding but also for expertise, training and patient focus.

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