Implementation of a Multidisciplinary Approach to Diagnosis and Management of Familial Hypercholesterolaemia (FH) in Wales: the Role of the FH Specialist Nurse


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The Wales FH service, launched in Sept 2010 has been developed based on NICE guidance to provide a coordinated approach to cascade testing for FH and other aspects of diagnosis and clinical management. Prior to this there was no cascade testing offered and inequitable service provision.

BHF FH nurses have been central to a variety of different developments including:
- Increased awareness of FH and access to specialist services, including genetic testing, for index patients and their families
- Multi-disciplinary partnership working across lipidology, genetics, cardiology and primary care.
- Development of an FH primary care pack and web-based teaching guide to support primary care
- A nationwide electronic patient pathway, utilising an IT system to ensure an equitable and systematic approach to patient care.

Results: Three specialist BHF FH nurses have helped to develop lipid services across Wales, doubling numbers of FH/lipid clinics from 8 to 16. Referral rates from primary/secondary care have increased by 19% in the first year and 13.2% in the second year, with over 1000 index patients assessed clinically for FH. Of these, 780 have been genotyped and 270 have received a genetic diagnosis (35%). Family cascade testing by genetic counsellors, using identified FH genetic mutations, has led to 222 relatives being diagnosed with FH and 181 being reassured that they do not have FH.

We conclude that FH specialist nurses have a key role to play in the development and delivery of FH services.

Implementation of Familial Hypercholesterolaemia (FH) Nurse Led Assessment Clinics in South Wales


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Aneurin Bevan Health Board (ABBB) has an approx population of 561,000. At the time of the launch of the all Wales FH testing service, December 2010, there were three lipid clinic sessions a month, seeing on average 12 new patients per month. New patient waiting lists were in excessive of 6 months, managed only by initiative clinics, twice a month.

Patients who were being seen in Lipid clinic, who met the criteria for FH genotyping, were seen by the FH specialist nurse and offered counselling and the option of consenting for the FH genetic test, either during their lipid clinic appointments or at specific community based nurse-led genotyping clinics. Due to the excessive lipid clinic waiting list times, no FH awareness sessions were carried out in primary care, to promote referral for FH assessment/genetic testing.

Since March 2012, ABHB has initiated nurse-led FH assessment clinics that run alongside consultant led clinics. By identifying patients for FH assessment at the time of referral, waiting times for FH assessment/genotyping have reduced from 6 months to 6-8 weeks. This has affected general lipid referral times with waiting list held at 6 months, without the use of initiative clinics. Reduction in waiting times have allowed for the promotion of FH within primary care. This improves access to FH genetic testing and the cascade testing of family members, with known pathogenic variants.

Two Cases of Tangier Disease Mutation: Clinical Relevance for Vascular Risk

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Tangier disease (TD) is an autosomal codominant disorder that causes a complete absence or extreme deficiency of high density lipoprotein (HDL). It is caused by mutations in the adenosine triphosphate binding cassette transporter 1 (ABCA1) which plays a role in the cholesterol efflux pathway. Two siblings initially presented with undetectable HDL cholesterol (HDL-C) levels. We received the samples through our Supraregional Assay Service (SAS) centre for Cardiac Biomarkers. The index case was under the care of dermatologists for acne and a lipid profile was ordered as oral retinoid therapy was considered. Repeat analysis confirmed a low HDL-C of 0.1 mmol/L in both siblings together with low total cholesterol but normal triglycerides. Apolipoprotein A1 (APO A1) was <0.05 g/L. Lipoprotein electrophoresis showed absence of alpha band which represents the HDL fraction. Lipid profile testing of other family members (both parents and a younger sibling) was normal with a normal lipoprotein electrophoresis pattern. We proceeded to genetic analysis of this family. Both siblings with low HDL-C were found to be compound heterozygotes for ABC-1 mutations described as ABCAI R 587W CGG to TGG in position 1759 in exon 14 (allele 1) for the mother and ABCAI G851R GGA to AAA on position 2552 in exon 18 (allele 2) for the father. Both the parents had HDL-C levels >1 mmol/L. Studies showed a 54% risk of developing peripheral neuropathy and a 20% risk of developing cardiovascular disease (CVD) in patient with TD compared with <1% and 5%, respectively for controls. In patients with TD care should be taken when administering drugs that may induce an atherogenic lipid profile. The 2 siblings with compound heterozygote ABC-1 mutations should be monitored for CVD risk factors.

Risk Equivalents in Hyperlipidaemia

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