Innovation Library NENC

Health Innovation North East and North Cumbria

Validating a regional model of familial hyper-cholesterolaemia diagnostic and cascade testing

Programmes

- Community/Out of Hospital
- Evaluation
- Spread and Adoption

People with the inherited medical condition Familial Hypercholesterolaemia (FH) are highly likely to develop early heart disease which can lead to a heart attack. Medical treatments and changes in diet are very effective at controlling the condition. These need to be started as early as possible but 85% of people with FH don't even know that they have it. A new state of the art genetic test, developed specifically for the North East population, can rapidly diagnose the condition in those suspected of having FH. Once FH is confirmed, the test can then be cascaded to relatives of the affected person to determine who else in the family has inherited the condition.

Around 1 in 500 people have FH which makes it one of the most common inherited conditions. The new genetic test can be used to detect FH in affected families, before the symptoms start to show. Early treatment can then be started to prevent heart disease in later life.

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> FH is a silent killer…Having a test which can diagnose a gene alteration means treatment can be started in time to prevent early deaths from heart disease.

Challenge/Problem Identified

Familial Hypercholesterolaemia (FH) is an inherited condition where a faulty gene causes high levels of cholesterol in the blood. Excess cholesterol can stick to blood vessel walls blocking the flow of blood to the heart. Consequently, FH is linked to a higher risk of early coronary heart disease.

FH can't be cured but it can be managed well using medical treatments (such as statins) and diet. These treatments dramatically reduce the risk of heart disease and death. However, they are most successful if they are started early, before the high blood cholesterol levels have caused permanent damage.

If one parent has FH then there is a 50% chance that one of their children will also have it. Genetic testing can confirm a person has a faulty FH gene. Further genetic testing would identify all the other family members with the condition. Treatment can then be started promptly in all those affected, before any adverse symptoms emerge.

Testing at risk relatives of those with genetically confirmed FH is called "Cascade" testing. It is the most effective way of finding un-diagnosed people and is recommended by NICE. However, confirming an FH diagnosis using full-gene sequencing and providing cascade testing to family members is expensive and has not routinely been available in the North East and North Cumbria.

Overview of innovation

- The Northern Genetics Service and NewGene (an NHS partner company) have developed a cost effective FH genetic test specific for the North East region.
- A clinical scoring system has been set up to identify likely sufferers of FH. This has led to over 350 patients being tested for the FH mutations most particularly associated with the North East region. Sixty-four family members have been cascade tested for known family mutations.
- The testing method looks for common regional mutations first and only does full sequencing if no common mutations are found. This saves both time and money. The majority of affected individuals (58%) are found in the first stage which is rapid and low cost. This reduces the diagnosis time for these patients as well as the overall cost of the test.

Outcome

Strong collaborations with local clinics together with AHSN (NENC) funding made it possible to turn this test into a regional service. The service was launched in April 2014.

The testing service was successfully handed over to the CCGs in July 2015 and is now completely CCG funded.

The British Heart Foundation (BHF) has funded two FH nurses who are now fully trained and delivering clinics and testing in the region. The BHF have also funded an administrative assistant who has been in post since August 2015.

A partnership has been established with the cholesterol charity, HEART UK. They produce useful literature about FH and genetic testing. HEART UK have helped to set up patient support groups in Newcastle and Sunderland to spread

awareness of the testing service.

Impact

- Patient group meetings and patient ambassadors offer crucial support across the region.
- The prevalence of positive mutations (detected by the 2 stage test) is 37% which is higher than in most other regions using full sequencing.
- Adoption of the 2 stage approach has so far resulted in a cost saving of >£24k and equates to a £65.91 reduction per patient.
- The concept of looking for regionally specific mutations first (before investing in costly sequencing), is generating interest in other regions.
- The service has prevented an estimated 12 deaths associated with this disease.
- Additional funding of >£174k has been achieved.
- Figures from the FH Guideline Implementation team suggest the diagnostic service has saved the NHS an estimated £716k.

Benefit

- Patients – Early diagnosis and getting the right medication as early as possible.
- Local GPs and CCGs – More accurate knowledge about the prevalence of FH in the region. Ability to provide appropriate care to as many patients as possible and prevent serious symptoms with early treatment.
- Local NHS Trusts – Early FH diagnoses should lead to a reduction in premature cardiovascular deaths and a reduced burden on cardiac services.

Plans for the future / spread and adoption

- The aim is to use cascade genetic testing to increase the diagnosis rate of FH in the region from 15% to 50% within 5 years.
- Further development of the genotyping assay and clinical selection criteria could make it even more effective and cost efficient.
- The majority of referrals currently come from the North of the region and Cumbria. More work needs to be done to promote testing, improve uptake of the service and ensure equal access across the whole AHSN region.
- Patient support groups are needed in every town that has a cholesterol clinic so that they are available to everyone that undergoes FH testing.
- A strategy is being developed to increase the referrals from primary care.

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