QS 41

The identification and management of heterozygous familial hypercholesterolaemia in adults, young people and children (QS41)

THE | COMMISSIONING review

NICE

QUALITY STANDARD

Introduction

In August 2013 NICE published its quality standard on the identification and management of heterozygous familial hypercholesterolaemia (FH) in adults, young people and children. The quality standard for FH specifies that services should be commissioned from and coordinated across all relevant agencies encompassing the whole FH care pathway. A person centred, integrated approach to providing services is fundamental to delivering high quality care to people with FH.

Quality statements

What is familial hypercholesterolaemia?

FH is an inherited condition caused by an alteration in a gene, which results in a high cholesterol concentration in the blood. Raised cholesterol concentrations are present from birth and lead to early development of atherosclerosis and coronary heart disease. The condition is transmitted from generation to generation in such a way that siblings and children of a person with FH have a one in two chance (50:50 risk) of also having FH.

Most people with FH have inherited an altered gene for FH in an autosomal dominant pattern from only one parent and are therefore 'heterozygous'. Occasionally, a person will inherit an altered gene from both parents and will have 'homozygous' FH or 'compound heterozygous' FH. Homozygous FH is rare, with an incidence of approximately one in a million.

The prevalence of heterozygous FH in the UK population is estimated to be one in 500, which means that approximately 120,000 people are expected to be affected. However, more than 80% of these are currently undiagnosed and untreated. If left untreated, more than 50% of men with heterozygous FH will develop coronary heart disease by the age of 50 years and more than 50% of women by the age of 60 years. Life expectancy is restored to near normal with early preventive treatment, particularly statin treatment and smoking cessation.

The importance of better identification of families/individuals at very high risk of cardiovascular disease, including those with FH, is recognised in the Department of Health's Cardiovascular Disease Outcomes Strategy.1

About NICE quality standards

NICE quality standards are a concise set of prioritised statements - each quality standard has an average of six to eight statements - designed to drive measurable quality improvements within a particular area of health or care.

Quality standards are derived from high quality evidence-based guidance, such as NICE guidance or guidance from NICE accredited sources, and are produced collaboratively with health care professionals, social care and public health practitioners, along with their partner organisations, patients, carers and service users.

NICE quality standards are not mandatory but they can be used for a wide range of purposes both locally and nationally. For example, patients and service users can use quality standards to help understand what high-quality care should include. Health care professionals and social care and public

health practitioners can use quality standards to help deliver high quality care and treatment.

The Health and Social Care Act 2012 sets out a clear expectation that the care system should consider NICE quality standards in planning and delivering services, as part of a general duty to secure continuous improvement in quality.

Commissioners and providers of health and social care should refer to the library of NICE quality standards when designing high quality services. Quality standard topics are formally referred to NICE by NHS England for health-related areas, and by the Department of Health and Department for Education for areas such as social care and public health.

What does the NICE quality standard say?

The NICE quality standard on FH consists of 8 quality statements covering diagnosis, specialist referral, DNA testing, cascade testing, drug treatment and annual review.

List of quality statements

Statement 1

Adults with a baseline total cholesterol above 7.5 mmol/l are assessed for a clinical diagnosis of FH.

Most of the 120,000 people estimated to have FH are undiagnosed and untreated. Because untreated FH carries a very high risk of cardiovascular disease, it is important that every opportunity is taken to identify people with FH and offer them treatment. Considering a clinical diagnosis of FH in people with high cholesterol will result in greater identification of FH and support cascade testing of their relatives. This will lead to more treatment to reduce cholesterol levels and prevention of coronary events among people with FH.

Statement 2

People with a clinical diagnosis of FH because they have high cholesterol and family history or other signs are referred for specialist assessment.

Diagnosing and managing FH in an individual and their relatives can be complex, and is best achieved when there is access to specialist services. Specialist assessments which include DNA testing should be performed by a healthcare professional with expertise in FH with access to the wider skills of a MDT (including a dietician, cardiologist and clinical geneticists). Once an accurate diagnosis has been made, people with FH can receive appropriate treatment, and cascade testing can be started to identify affected family members.

Statement 3

People with a clinical diagnosis of FH because they have high cholesterol and family history or other signs are offered DNA testing as part of specialist assessment.

DNA testing is important because it increases the certainty of a diagnosis of FH and allows the identification

of affected and unaffected relatives through cascade testing.

Statement 4

Children at risk of FH are offered diagnostic tests by the age of 10 years.

Children at risk of FH are those who have 1 parent with the condition. Children with FH begin to develop cardiovascular disease before clinical signs appear, with thickening of the carotid artery wall identifiable by the age of 10 years. Diagnosis by the age of 10 years – by a healthcare professional with expertise in FH in children who has access to the wider skills of a MDT - allows lifestyle changes and tailored therapy if indicated, which will reduce

long term problems associated with high cholesterol and improve long term health.

Statement 5

Relatives of people with a confirmed diagnosis of FH and a known genetic mutation are offered DNA testing through a nationwide, systematic cascade process.

Most people in the UK with FH are undiagnosed. Cascade testing has been shown to be effective for identifying people with FH, especially when provided nationwide. Nationwide cascade testing ensures that all family members can access DNA testing wherever they live.

Statement 6

Adults with FH receive lipid modifying drug treatment to reduce LDL C (low-density or "bad" cholesterol) concentration by more than 50% from baseline.

Lipid modifying drug treatment reduces LDL C levels and prevents the development of cardiovascular disease. Studies indicate that treatment that lowers LDL C levels by more than 50% from baseline offers greater benefit for plaque stabilisation than treatment that is less effective at reducing LDL C.

Statement 7

Children with FH have an assessment for possible drug treatment to reduce the low-LDL-C in their blood by a specialist in a children's department, by the age of 10 years.

Once a child is diagnosed as having FH, it is important they are assessed for lipid modifying drug treatment as soon as possible. The assessment should include a discussion of the harms and benefits of different treatments. This allows children to start treatment as soon as it is appropriate and before significant atherosclerosis has developed.

Statement 8

People with familial hypercholesterolaemia are offered a detailed review of their condition at least once a year.

Regular structured review enables treatment to be monitored and adjusted to achieve the recommended LDL C concentration. It also enables monitoring for the possible development of symptoms and signs of coronary heart disease and optimising management. In addition, records can be maintained of affected family members and information can be tailored to individual circumstances. Progress with cascade testing of at risk relatives can also be monitored.

The NICE quality standard on familial hypercholesterolaemia is available on the NICE website at http://guidance.nice.org.uk/QS41 The NICE clinical guideline on familial hypercholesterolaemia is available at http://guidance.nice.org.uk/CG71

References

1. www.gov.uk/government/publications/improving-cardiovascular-disease-outcomes-strategy

I  MY WALKS

When managing dyslipidaemia, the heart is not the only muscle that needs loving.

For the treatment of hypercholesterolaemia.

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