NATIONAL INSTITUTE FOR HEALTH AND CARE EXCELLENCE

INDICATOR DEVELOPMENT PROGRAMME

Consultation report

Indicator area: Familial hypercholesterolemia (FH)

Consultation period: 17 April – 16 May 2019

Date of Indicator Advisory Committee meeting: 5 June 2019

Output: New indicators for general practice

Contents

Introduction

In some people high total cholesterol levels are caused by an inherited gene defect: familial hypercholesterolemia (FH). A raised cholesterol concentration is present from birth and may lead to early development of atherosclerotic disease.

Left untreated FH can lead to premature coronary heart disease. However, with appropriate lipid-lowering treatment, care is highly effective and life expectancy can return to normal (Qureshi et al. 2016)

The prevalence of heterozygous FH in the UK population is estimated to be 1 in 250. Currently it is estimated that up to 80% of people with FH are undiagnosed and therefore untreated.

Diagnosis is based on the Simon Broome criteria or the Dutch Lipid Clinic Network (DLCN) criteria which includes information on family history, total and LDL cholesterol concentrations, clinical signs such as tendon xanthomata and DNA testing.

Considering a diagnosis of FH in primary care will result in greater identification and support cascade testing of relatives. It will lead to more treatment of high cholesterol and the prevention of CHD amongst people with FH.

Summary of indicators included in the consultation

ID	Indicator wording	Evidence source
IND8	The percentage of people aged 29 years and under, with a total cholesterol concentration greater than 7.5 mmol/l that are assessed against the Simon Broome or Dutch Lipid Clinic Network (DLCN) criteria.	Familial hypercholesterolaemia: identification and management (2017) NICE guideline CG71, section 1.1 Familial hypercholesterolaemia (2013) NICE quality standard QS41, statement 1
IND9	The percentage of people aged 30 years and older with a total cholesterol concentration greater than 9.0mmol/l that are assessed against the Simon Broome or Dutch Lipid Clinic Network (DLCN) criteria.	Familial hypercholesterolaemia: identification and management (2017) NICE guideline CG71, section 1.1 Familial hypercholesterolaemia (2013) NICE quality standard QS41, statement 1
IND10	The percentage of people with a clinical diagnosis of FH referred for specialist assessment.	Familial hypercholesterolaemia: identification and management (2017) NICE guideline CG71, recommendations 1.1.6, 1.1.8 and 1.2.2 Familial hypercholesterolaemia (2013) NICE quality standard QS41, statement 2

IND8: FH - People aged 29 years and under with a high total cholesterol

The percentage of people aged 29 years and under, with a total cholesterol concentration greater than 7.5 mmol/l that are assessed against the Simon Broome or Dutch Lipid Clinic Network (DLCN) criteria.

Rationale

This indicator is intended to increase identification of those aged 29 years and under with undiagnosed FH.

Summary of consultation comments

Stakeholders made the following positive comments in relation to this indicator:

- This indicator was supported to improve early FH identification, referral and treatment rates and will have a significant impact on reducing mortality and morbidity
- It was strongly recommended that this indicator is developed into a full QOF indicator with appropriate achievement points.

Stakeholders outlined the following concerns about this indicator:

- It would add a substantial burden to primary care with limited clinical benefit
- Small indicator denominators at individual GP practice level negatively impact on the validity of the indicator
- FH screening is not currently approved by the National Screening Committee
- Triglyceride levels will need to be considered as well as high cholesterol. Ideally patients with secondary causes should also be excluded before Simon Broome or DLCN assessment is carried out
- This indicator will require a Read/SNOMED code for assessment by Simon Broome or DLCN criteria, if not already available.

Considerations for the advisory committee

The committee is asked to consider:

- The concerns raised around the burden of testing and links to screening, possible misunderstanding around how the indicator works?
- Stakeholder concerns around patient numbers:
    - ◊ small indicator denominators at individual GP practice level
    - ◊ burden of work for general practice
- Additional assessments for triglycerides and exclusion of secondary causes.
- Codes required for GP systems.

IND9: FH - People aged 30 years and older with a high total cholesterol

The percentage of people aged 30 years and older with a total cholesterol concentration greater than 9.0mmol/l that are assessed against the Simon Broome or Dutch Lipid Clinic Network (DLCN) criteria.

Rationale

This indicator is intended to increase identification of those people aged 30 years and over with undiagnosed FH.

Summary of consultation comments

Stakeholders made the following positive comments in relation to this indicator

- This indicator will support improved diagnosis and treatment rates to prevent premature death.

Stakeholders outlined the following concerns about this indicator:

- This indicator does not support the wider identification of these people
- Testing would add a substantial burden to primary care with limited clinical benefit
- Small indicator denominators at individual GP practice level negatively impact on the validity of the indicator
- FH screening is not currently approved by the National Screening Committee
- This indicator's age range needs to be reviewed as testing lipids for patients aged 30 and over would add a substantial burden to primary care. Is this proportionate to the benefit to patients?
- Triglycerides and the need to exclude secondary causes of high cholesterol should be considered
- Appropriate coding will be required for GP systems.

Considerations for the advisory committee

The committee is asked to consider:

- Stakeholder concerns around patient numbers:
  ◊ small indicator denominators at individual GP practice level
  ◊ burden of work for general practice
- Suggestion to construct one indicator based on IND8 and IND9 for all age ranges to avoid the challenge of small numbers.

IND10: FH – Referral for specialist assessment

The percentage of people with a clinical diagnosis of FH referred for specialist assessment.

Rationale

Diagnosis and management of FH can be complex and is best achieved in specialist services. Referral from primary care for specialist assessment, including DNA testing can confirm a diagnosis. 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.

Summary of consultation comments

Stakeholders made the following positive comments in relation to this indicator:

- It was strongly recommended that this indicator is developed to help drive identification of people with suspected FH in primary care and ensure appropriate management
- Treating FH with specialist involvement was supported based on the complexity of the condition and its management.

Stakeholders outlined the following concerns about this indicator:

- Clarification needed on which specialist assessment service people with FH should be referred to and what this entails
- Accuracy of diagnosis coding
- This indicator should be changed from people with a clinical diagnosis of FH who are referred to a lipid or endocrine clinic to those who have been genetically screened as the purpose of the referral to secondary care is to get diagnostic confirmation through genetic testing however this is not current national practice. By April 2020 genomic hubs will come online and this should be the end point. Arguably as GPs cannot currently refer FH patients directly for genomic testing the measure in QOF is restricted. Knowing and managing genomic risk is vital.

Measuring referrals without measuring those having genetic testing may add increased cost and little benefit

- It was assumed 'referred for specialist assessment' includes phone call assessment. Could this description be expanded to include face-to-face specialist assessment?

Considerations for the advisory committee

The committee is asked to consider:

- Which specialist assessment service people should be referred to
- Accuracy of diagnosis coding
- Focussing on a combined measure of referrals and genetic testing
- The description of specialist assessment.

General comments on FH indicators

The following is a summary of general comments on the 3 draft FH indicators:

- The numbers per GP practice are too low for these indicators to provide an acceptable indication of care standards
- A number of stakeholders highlighted [The NHS Long Term Plan](#) which supports 10 year cardiovascular disease prevention and includes key risk factors such as cholesterol, raised blood pressure and atrial fibrillation
- One stakeholder raised concern about the lack of indicators for cholesterol identification and management in people who have already suffered a cardiovascular event or are at the highest risk of further cardiovascular events. It was felt that these current indicators do not go far enough to improve outcomes in line with aspirations of [The NHS Long Term Plan](#)
- One stakeholder highlighted the need for a national FH registry for people and families. This would also include service provision management and cascade testing
- Self-care education for people with FH was highlighted as a general consideration as different people have different symptoms
- Ensure data and definitions link to those used in the CVDPrevent audit.

Suggestions for additional indicators

- Suggestion to include additional indicators to identify and manage people with FH.

Appendix A: Consultation comments

ID	Indicator	Stakeholder	Comment
1	General	British Cardiovascular Society (BCS)	The BCS Clinical Standards group have reviewed the proposed new indicators for FH and we are very positive about the 3 suggested FH indicators and have no comments. We feel it is a good step and supports the 10 year CVD prevention plan.
2	General	PHE	PHE welcomes the development of indicators to support Familial Hypercholesterolaemia testing IND 8 & IND 9 to support NICE guidance and the NHS Plan commitment. We would like to highlight ongoing work (which NICE is linked with) on the NHS funded **CVD prevent audit** to ensure there is a consistent approach to data definitions for these two pieces of work.
3	General	Elcena Jeffers Foundation	There is a wish to know about self-care in general public education in life as different people has different diseases or ailments
4	General	Royal College of Nursing	The Royal College of Nursing (RCN) welcome the consultation on the listed NICE QOF indicators. The RCN invited members who care for people with the listed conditions to review the draft indicators on our behalf. The comments below reflect the views of our reviewers.
5	General	SANOFI	Background / Rationale for piloting and consulting on the new indicator Would a reference need adding to the prevalence of heterozygous Familial Hypocholesteraemia and estimated % of people with Familial Hypocholesteraemia? Numbers vary depending on the evidence referenced.

6	General	SANOFI	Recommendation to include two further indicators to support the CVD prevention plan within the long term plan and current NICE guidelines to identify and manage patients with Familial Hypocholesteraemia
			1) The percentage of people aged 60 years and younger who have had an emergency admission for a cardiovascular event in the last year, that are assessed against the Simon Broome or Dutch Lipid Clinic Network (DLCN) criteria.
			   - **Evidence base - NICE guidance CG71 (2017) Familial Hypercholesterolaemia: identification and management.**
			   - *1.1.3 For people with a personal or family history of premature coronary heart disease (an event before 60 years in an index individual or first-degree relative), but whose total cholesterol is unknown, offer to measure their total cholesterol*
			2) The percentage of people who have had an emergency admission for a cardiovascular event in the last year and are on maximum statin therapy, that are assessed against the Simon Broome or Dutch Lipid Clinic Network (DLCN) criteria.
			   - **Evidence base - NICE guidance CG71 (2017) Familial Hypercholesterolaemia: identification and management.**

			- *1.1.3 For people with a personal or family history of premature coronary heart disease (an event before 60 years in an index individual or first-degree relative), but whose total cholesterol is unknown, offer to measure their total cholesterol* - **Evidence base - NICE guidance CG181 (2016) Cardiovascular disease: risk assessment and reduction, including lipid modification** - *1.3.21 Do not delay statin treatment in secondary prevention to manage modifiable risk factors.* - *1.3.22 If a person has acute coronary syndrome, do not delay statin treatment. Take a lipid sample on admission and about 3 months after the start of treatment*
7	General	AMGEN Ltd	The three current indicators under consultation, referenced above, appropriately seek to improve the identification and management of patients with suspected FH. Based on the prevalence of FH, however, there will be a significantly larger population of patients who have already suffered cardiovascular events, without FH, who remain at very high risk of further avoidable events and who should be appropriately managed. Given the complete lack of indicators for identification and management of patients' cholesterol in those who have already suffered a cardiovascular event (secondary prevention) these current indicators do not go far enough. QOF currently supports improved management of raised blood pressure and atrial fibrillation, but notably does not address the issue of raised cholesterol in secondary prevention patients and as such is out of step with the NHS Long Term Plan. As demonstrated in the report that accompanied the QOF review [1], removal of indicators incentivising active identification and management has led to a drop off in performance in this indicator for secondary prevention patients putting them at risk of avoidable CVD events. In addition, recent UK figures highlighted by the British Heart Foundation (BHF) have shown that deaths from heart and circulatory diseases among people under 75 is on the rise for the first time in 50 years [2]. **Indicators should therefore include factors such as:** % patients who have suffered an MI or stroke in the previous 12 months that are taking a high intensity statin / atorvastatin 80mg. % patients who have suffered an MI or stroke in the previous 12 months whose total cholesterol is below 5mmol/l. **We therefore suggest that additional NICE and QOF indicators are introduced to ensure patients at the highest risk of suffering further cardiovascular events are appropriately managed in order to improve outcomes in line with aspirations of the NHS Long Term Plan.** [1] IMPACT OF REMOVING INDICATORS FROM THE QUALITY AND OUTCOMES FRAMEWORK: RETROSPECTIVE STUDY USING INDIVIDUAL PATIENT DATA IN ENGLAND. Policy Research Unit in Commissioning and the Healthcare System, June 28th, 2018. [2] 13 May 2019 https://www.bhf.org.uk/what-we-do/news-from-the-bhf/news-archive/2019/may/heart-and-circulatory-disease-deaths-in-under-75s-see-first-sustained-rise-in-50-years

8	IND8	Royal College of Nursing	This is an important new indicator as currently the number of people who have been identified with FH is very low. Early identification and referral for treatment will have a significant impact on reducing mortality and morbidity. The difficulty will be in ensuring that people under the age of forty get their cholesterol levels checked and identifying those in which this check should be done could be a challenge.
9	IND8	AMGEN Ltd	Familial Hypercholesterolaemia (FH) leads to raised cholesterol levels from birth, which cause the early development of atherosclerotic disease. This in turn increases the risk of premature stroke and coronary heart disease with significant reductions in both the quality and quantity of life years. The need to address patients with this genetic condition has also been recognised in the NHS Long Term Plan published in January this year. Included within the plan is a recognition that cholesterol, alongside raised blood pressure and atrial fibrillation, is a key modifiable risk factor to be addressed if the NHS is to achieve its goal of preventing 150,000 **heart attacks, strokes and dementia cases over the next 10 years.** To help achieve this figure, the Long Term Plan also highlights the need to increase identification of patients with FH from 7% currently to 25% in the next 5 years. At current levels of identification, it is almost certain that this target will be missed by some distance. Introducing indicators to support and drive this activity is absolutely necessary and a hugely positive step. Given the number of often competing therapeutic priorities faced by general practitioners, it is often those which are not incentivised that are missed. A recent analysis [1] conducted to support NHS England's Report of the Review of the Quality and Outcomes Framework in England [2] demonstrated the impact a Quality and Outcomes Framework (QOF) indicator can have. It showed that following the removal of CHD003 (percentage of patients with coronary heart disease whose last measured total cholesterol, measured in the preceding 15 months, is 5mmol/l or less) there was a drop in indicator performance and increase an in missed cholesterol readings. We believe that the removal of cholesterol as a measurable, modifiable risk factor from this and other CVD related QOF domains puts patients at risk of suffering heart attacks and strokes. Cholesterol is now only present in the diabetes domain of QOF, whereas it was once present in several CVD related ones, and we believe this has been a serious omission given levels of stroke and coronary heart disease remain the single biggest contributor to premature mortality and morbidity in England. NICE Guideline CG71 brings together a comprehensive suite of evidence and recommendations to identify and manage patients with suspected FH, but without incentives and drivers in the system implementation of such guidance often does not happen.

			We therefore fully support introduction of this indicator and strongly recommend that it is developed into a full QOF indicator with appropriate points for achievement to help drive activity in primary care to identify patients with suspected FH. [1] IMPACT OF REMOVING INDICATORS FROM THE QUALITY AND OUTCOMES FRAMEWORK: RETROSPECTIVE STUDY USING INDIVIDUAL PATIENT DATA IN ENGLAND. Policy Research Unit in Commissioning and the Healthcare System, June 28th, 2018. [2] Report of the Review of the Quality and Outcomes Framework in England. NHS England, July 4th 2018.
10	IND8	British Medical Association	While agreeing that this represents best practice, we cannot support this as an indicator of quality as the numbers in each practice will be too low to provide an acceptable indication of standards of care.
11	IND8	National Pharmaceutical Advisers Group (PAG)	Agree with rationale and should improve rates of diagnosis and treatment to prevent premature death.
12	IND8	Royal College of General Practitioners	We are concerned that the indicator to test lipids for patients aged 29 and under would add a substantial burden to primary care and question whether this is proportionate to the benefit to patients. Screening for FH is not currently approved by the National Screening Committee and is likely to cause a large degree of extra work with limited clinical benefit https://legacyscreening.phe.org.uk/screening-recommendations.php
13	IND8	Heart UK	Consideration needs to be given to triglycerides. Many of those with high cholesterol will also have high trigs and may not have FH and require assessment by Simon Broome or DLCN. Ideally patients with secondary causes should also be excluded before assessment by SB/DLCN is done. If the code for potential heterozygous familial hypercholesterolaemia (HeFH) was produced and used then the indicator would need revising to: "The percentage of people…, with a total cholesterol concentration greater than…, where secondary causes and significant hypertriglycaeridaemia have been excluded ie have potential FH, that are assessed against the Simon Broome or Dutch Lipid Clinic (DLCN) criteria."
14	IND8	Heart UK	Will require a READ/SNOMED code for assessment by SB/DLCN, if not already available
15	IND9	Heart UK	Consideration needs to be given to triglycerides. Many of those with high cholesterol will also have high trigs and may not have FH and require assessment by Simon Broome or DLCN. Ideally patients with secondary causes should also be excluded before assessment by SB/DLCN is done.

			If the code for potential heterozygous familial hypercholesterolaemia (HeFH) was produced and used then the indicator would need revising to: "The percentage of people…, with a total cholesterol concentration greater than…, where secondary causes and significant hypertriglycaeridaemia have been excluded ie have potential FH, that are assessed against the Simon Broome or Dutch Lipid Clinic (DLCN) criteria."
16	IND9	Heart UK	Will require a READ/SNOMED code for assessment by SB/DLCN, if not already available
17	IND9	National Pharmaceutical Advisers Group (PAG)	Agree with rationale and should improve rates of diagnosis and treatment to prevent premature death.
18	IND9	AMGEN Ltd	We believe that a separate indicator based around a different threshold in IND9 compared to IND8, i.e. 9mmol/l versus 7.5mmol/l, is not required and should be removed. Having a higher cut-off will without doubt mean a cohort of patients with FH whose total cholesterol is between 7.5mmol/l and 9mmol/l will not be identified if this indicator is used. It will effectively discriminate against those patients above 29 years old who are in this cholesterol range. As it stands, IND9 does not support the wider identification of these patients **We therefore strongly suggest that this indicator be merged with IND8 and in effect a single indicator produced that would not prejudice patients falling in this cholesterol range over the age of 29 leaving them without a potential diagnosis and appropriate treatment for FH.**
19	IND9	British Medical Association	While agreeing that this represents best practice, we cannot support this as an indicator of quality as the numbers in each practice will be too low to provide an acceptable indication of standards of care.
20	IND9	Royal College of General Practitioners	We are concerned that the indicator to test lipids for patients aged 30 and over would add a substantial burden to primary care and question whether this is proportionate to the benefit to patients. Screening for FH is not currently approved by the National Screening Committee and is likely to cause a large degree of extra work with limited clinical benefit https://legacyscreening.phe.org.uk/screening-recommendations.php We suggest that the age range is reviewed.
21	IND10	AMGEN Ltd	Once patients have been identified through the Simon Broome criteria or the Dutch Lipid Clinic Network (DLCN) criteria as being suspected FH it may be that primary care healthcare professionals do not feel equipped to manage them appropriately. Encouraging appropriate referral of all clinically diagnosed patients for specialist assessment is essential in ensuring that patients can receive appropriate, optimised therapy to prevent avoidable cardiovascular events.

			We therefore fully support introduction of this indicator and strongly recommend that it is developed into a full QOF indicator with appropriate points for achievement to help drive activity in primary care to identify patients with suspected FH and ensure appropriate management.
22	IND10	British Medical Association	While agreeing that this represents best practice, we cannot support this as an indicator of quality as the numbers in each practice will be too low to provide an acceptable indication of standards of care.
23	IND10	Clinical Genetics Society	The document states that patients with a diagnosis of familial hypercholesterolaemia should be referred for specialist assessment including DNA analysis. It should be explicit which service patients should be referred to, as currently many patients are referred into genetics services for genetic testing. Genetics services are not commissioned to perform genetic testing for this group of patients and they should be referred directly to their local lipid clinic who can both request the DNA analysis if indicated and manage the patient. Referral to the genetics service often leads to delays in the patient being seen, with the potential for detrimental health consequences.
24	IND10	Heart UK	Important to reinforce use of correct coding for possible HeFH (ie clinical diagnosis). The separate code for definite (He)FH should only be used once diagnosis is confirmed.
25	IND10	Heart UK	HEART UK supports a single, national registry for FH to register patients and families and manage their service provision, particularly cascade testing. In England, the licenses required by Trusts to use the PASS database are largely funded through HEART UK and has been a particularly useful tool in screening for FH. "The percentage of people with a clinical diagnosis of FH referred for specialist assessment and included on the national FH registry."
26	IND10	National Pharmaceutical Advisers Group (PAG)	Involvement of a specialist to treat FH makes sense given complexity of disease and importance of the condition to be managed well.
27	IND10	Royal College of General Practitioners	While agreeing that this represents best practice, we cannot support this as an indicator of quality as the numbers in each practice will be too low to provide an acceptable indication of standards of care.
28	IND10	NHS Medway CCG	I'd propose that this indicator is changed from those referred to a lipid or endocrine clinic to those who have been genetically screened. I understand SNOMED codes exist to record this. The purpose of the referral to secondary care is to get diagnostic confirmation, which is by way of genetic testing, but we know that this is not happening in many parts of the country. As the genomic hubs come online, and will be by April 2020 if not before, this should be our end point. It could be argued that as GPs cannot as yet refer directly for genomic testing of FH patients the measure in QOF is restricted to what is in the gift of the GP, but what matters to patients, and importantly for their relatives if they are mutation positive for a FH gene, is that we know and manage their genomic risk. Measuring referrals without measuring those having genetic testing may add great cost and little benefit.

29	IND10	SANOFI	We assume 'referred for specialist assessment' includes phone call assessment, could this description be expanded to include face to face specialise assessment to increase the likelihood of diagnosing FH