

# Consultation report: Familial hypercholesterolaemia

Consultation period: 13 April 2023 – 15 May 2023

Date of Indicator Advisory Committee meeting: 06 June 2023

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## **FH assessment and diagnosis**

IND2022-130: The percentage of patients with a total cholesterol reading greater than 7.5 when aged 29 years or under, or greater than 9.0 when aged 30 years or over, who have been:

- diagnosed with secondary hyperlipidaemia or
- clinically assessed for familial hypercholesterolaemia or
- referred for assessment for familial hypercholesterolaemia or
- genetically diagnosed with familial hypercholesterolaemia.

### **Rationale**

Familial hypercholesterolaemia (FH) is a genetic disorder that causes a high cholesterol level. This increases the likelihood of coronary artery disease, heart attacks and sudden cardiac death. Early detection and genetic diagnosis will lead to provision of appropriate lipid-lowering treatment to lower these risks and improve outcomes.

### **Summary of consultation comments**

Responses were received from 5 stakeholders. They noted the potential to improve diagnosis rates and the health benefits of treatment for those subsequently identified as having FH.

At consultation NICE asked whether it was acceptable for the denominator to include any historical reading and not be limited to recent readings (for example, in the preceding 12 months) until performance improves. Most stakeholders agreed that this approach was acceptable however one felt that the denominator should be restricted to recent high readings only (the past year).

Clarification was sought on the specific type of cholesterol test that would be required for the reading (HDL, non-HDL, LDL).

There was mixed response on the potential workload implications. The data should be available within general practice IT systems, but concerns were

raised that the indicator could increase workload within specialist services who already have limited capacity.

One stakeholder suggested that additional indicators would be needed to ensure that people are treated appropriately when diagnosed with FH. It was also felt that there would be benefit to an additional focus on people with a learning disability or people who are underserved.

## Considerations for the advisory committee

The committee is asked to consider:

- The acceptability of including any historically high reading in the denominator.
- Whether the type of cholesterol test needs specifying.
- The potential increased workload for specialist services.
- Whether supplementary indicators are needed on diagnosis in specific groups (such as people with a learning disability or people who are underserved).
- Whether supplementary indicators are needed on treatment once FH is diagnosed. The existing [NICE indicator](#) on provision of lipid lowering therapies for people with CVD does not include FH. However, there are likely to be small numbers per practice until diagnosis rates improve.

## Appendix A: Consultation comments

ID	Stakeholder organisation	Comment	NICE response
1	Amgen Ltd	Introduction of this indicator should support the identification and treatment of people with familial hypercholesterolaemia (FH) who are currently going undiagnosed. Referral for FH testing or subsequent diagnosis does not necessarily translate to treatment however, so it is important that this indicator is not considered in isolation but that it is accompanied by measures and incentives beyond QOF that enable higher rates of treatment within this population.	<p>Thank you for your comment.</p> <p>New indicators on treatment for people with familial hypercholesterolemia were considered by the committee. It was agreed that diagnosis rates should be improved before the numbers of people with diagnosed FH would be enough to assess variation in treatment across practices.</p>
2	Amgen Ltd	Where cholesterol readings are being taken, the indicator as it is written should not place much of an additional burden on general practice, as it just requires the onward referral of patients with readings above the stated levels. Amgen believes that additional clarity over the type of cholesterol test required (HDL, non-HDL, LDL) is necessary to ensure consistency in application and mitigate any confusion about the workload implication where additional testing might be perceived. There is no suggestion in the indicator as it is written that any additional data collection will be required.	<p>Thank you for your comment.</p> <p>The indicator is based on NCD102 from the Network Contract DES 2023/24 and would use the same codes for total cholesterol recordings:</p> <ul style="list-style-type: none"> <li>• Plasma total cholesterol level (observable entity)</li> <li>• Substance concentration of cholesterol in plasma (observable entity)</li> <li>• Cholesterol level (observable entity)</li> <li>• Total cholesterol level (observable entity)</li> <li>• Serum cholesterol level (observable entity)</li> <li>• Serum fasting total cholesterol level (observable entity)</li> <li>• Substance concentration of cholesterol in serum (observable entity)</li> </ul>

ID	Stakeholder organisation	Comment	NICE response
			<ul style="list-style-type: none"> <li>• Serum total cholesterol level (observable entity)</li> </ul> <p>Accompanying guidance has been updated to clarify that ideally a fasting sample should be used but the indicator searches for any total cholesterol readings.</p>
3	Amgen Ltd	<p>All patients within the denominator have a very high cholesterol level, so their referral to specialist services or for further (genetic) testing is entirely appropriate. Should they test negative for FH, it is likely that they will require some lipid lowering therapy and ruling out FH may help in making decisions about treatment options.</p> <p>With FH incidence in the UK estimated at 1 in 250 there is a risk that the introduction of this indicator would see a rise in referrals to specialist lipid clinics which are already experiencing challenges around capacity. Adding this burden without considering ways to improve the management of existing capacity could see a negative impact on those clinics and patients waiting for care. Similar challenges may also exist for genomic laboratory hubs responsible for testing around FH. This is not a reason not to implement the indicator, however it is a live challenge for health systems.</p>	<p>Thank you for your comment.</p> <p>The committee agreed that in the first years of implementation there could be increased workload implications for primary care and specialist services. The accompanying documentation has been updated to highlight this risk and note that consideration needs to be given if implemented in practice.</p>
4	Amgen Ltd	<p>FH is equally prevalent across all demographic and socio-economic groups so there should not be any specific differential impact in introducing this indicator.</p>	<p>Thank you for your comment.</p>

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5	Amgen Ltd	<p>FH is equally prevalent across all demographic and socio-economic groups so there are no inherent issues resulting from the condition's epidemiology. Underlying challenges around engaging certain groups with healthcare services will apply to this indicator. With FH being a genetic condition, healthcare providers will be able to consider ways to proactively engage people to optimise success with this indicator.</p> <p>Directing communications to families of previously identified FH patients is the most effective way to find FH patients, identification via General Practice records will enable personalisation of communication to those patients, rather than 'catch all' recalls which are more likely to exacerbate inequalities.</p> <p>Work has already been undertaken by the Academic Health Science Networks alongside Primary Care Networks, voluntary and charity sector in this disease area to reduce inequalities.</p> <p>This builds on work already undertaken in other ICBs to engage with the most vulnerable, isolated and highest risk patient groups, for example there have been some useful initiatives whereby children are engaged through schools, and they support education across their families where there are potentially language, barriers and/or low health literacy.</p>	Thank you for your comment.

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6	Amgen Ltd	<p>Amgen believes that Question 6 in the consultation is ambiguous when referring to the terms 'new' and 'historic'. Clarification would be helpful over whether 'new' is time-bound, for example within the current QOF year, or refers to the most recent reading.</p> <p>In seeking to maximise the identification of FH to achieve expected levels and given the nature of the condition as a chronic genetic one, maintaining the denominator as historic high cholesterol readings appears appropriate. Where more recent readings drop below the indicator thresholds, investigation for FH may still be appropriate depending on individual circumstances, and to maximise identification further investigation would appear to be appropriate. In cases where FH has been ruled out this should be recorded and used to exclude patients from searches.</p> <p>People with very high cholesterol levels will retain the risk of cardiac events. Amgen suggests that wherever a patient is identified as having high cholesterol it is important that they are initiated on the most appropriate treatment as soon as possible. Regarding this indicator, while referral for investigation or testing is important to establish the presence or absence of FH, it should not result in any delays to initiation on lipid-lowering therapies.</p>	<p>Thank you for your comment.</p> <p>The accompanying documentation has been updated to highlight that the indicator does not include a specific time period for the cholesterol reading.</p> <p>An example has also been added to the validity assessment to clarify that the indicator looks for the earliest reading on the patient's record and does not look for newer or more recent cholesterol readings only (for example, within the preceding 12 months only).</p> <p>New indicators on treatment for people with familial hypercholesterolemia were considered by the committee. It was agreed that diagnosis rates should be improved before the numbers of people with diagnosed FH would be enough to assess variation in practice across practices.</p>
7	NHS England	<p>CVD prevention is especially important for people with a learning disability as it was identified as one of the leading causes of death in last year's LeDeR.</p>	<p>Thank you for your comment.</p> <p>If data was extracted at patient level we would expect that local systems and providers could</p>

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		<p>It would be interesting to understand proportion of people with a learning disability (or autistic people) being identified as having FH.</p> <p>Can part of this indicator also being identification of which underserved groups are most likely to be impacted so can be targeted in primary care.</p>	interrogate it further to understand performance within particular groups.
8	NHS England	<p>Keeping the patient included in the denominator has the benefit that they are always included and as such you will not omit them from relevant searches. The downside is that as the number of treated or excluded individuals increase the denominator will get larger and larger and as such new patients with a raised level will not have such an impact on the overall percentage. This may mean that there is less of a drive to address these patients. On balance keeping it as per the advice is sensible.</p>	<p>Thank you for your comment.</p> <p>The accompanying documentation has been updated to highlight that the denominator will include some of the same patients for the same high reading each time data is extracted and therefore the indicator should be reviewed once diagnosis rates have improved.</p>
9	Primary Care Cardiovascular Society	<p>Diagnosis and subsequent treatment of FH is important as the health benefits are substantial.</p> <p>However, there is a capacity issue. As things currently stand many ICSs do not have the infrastructure to deal with this.</p> <p>There is also variance in the provision of services to support cascade testing of relatives such that there is a risk of widening health inequalities.</p> <p>We agree that the denominator should include any historically high cholesterol result (and not only new high cholesterol results) as this should have been</p>	<p>Thank you for your comment.</p> <p>The committee agreed that in the first years of implementation there could be increased workload implications for primary care and specialist services. The accompanying documentation has been updated to highlight this risk and note that consideration needs to be given if implemented in practice.</p> <p>The accompanying documentation has been updated to highlight that the denominator will include some of the same patients for the same high reading each time data is extracted and</p>



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		assessed at some point and the outcome appropriately coded.	therefore the indicator should be reviewed once diagnosis rates have improved.
10	Royal College of General Practitioners	<p>The RCGP is calling for an immediate suspension of QOF during the current crisis with the need of a review to identify 5-10 indicators that have the greatest evidence of impact on patient outcomes that could be retained once QOF is re-introduced. Over the years, QOF has become painfully detailed in terms of reporting, both clinically and administratively, causing increasing frustration for GPs. This can divert the attention of GPs away from the patients sitting in front of them in consultations. It is also likely to be driving an increase in the number of unnecessary appointments, which may be more about ticking a box to reach a target rather than looking at what is needed by the individual patient.</p>	Thank you for your comment.
11	Royal College of General Practitioners	<p>There is a risk with this indicator that to achieve QOF, patients could be referred on to lipid clinics as the simplest way of meeting the target at a time of immense pressure in primary care and to avoid additional appointments being used within primary care.</p> <p>If all historical high cholesterol are included, then there is a risk of over diagnosing and over reporting of FH. We would recommend that the high cholesterol should be limited to a specific time period e.g., 1 year, to ensure this is achievable</p>	<p>Thank you for your comment.</p> <p>The committee agreed that in the first years of implementation there could be increased workload implications for primary care and specialist services. The accompanying documentation has been updated to highlight this risk and note that consideration needs to be given if implemented in practice.</p> <p>The committee noted that including historical high readings would help improve case-finding and diagnosis rates. However, an additional indicator focused on new high readings in the preceding 12</p>

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		within primary care and that the results accurately reflect those not on treatment.	months was agreed by the committee and progressed for inclusion on the NICE menu. Likely low numbers per practice would mean it would not be suitable for use in the QOF.
12	British Medical Association's General Practitioners Committee	<p>In the opinion of BMA's General Practitioners Committee (England), QOF needs a wholesale review, and introducing new indicators and tinkering with old ones does not fit with the agreement to carry out a wholesale review made by NHSE and DHSC.</p> <p>In addition, when patients have multiple co-morbidities, single disease measures can be challenging. It would be helpful if NICE could advise whether there are conditions or medications for other conditions, that commonly occur with the single disease, that will result in a caution flag when co-prescribing, and if there are, provide guidance on whether to prescribe.</p>	Thank you for your comment.
13	British Medical Association's General Practitioners Committee	<p>The guidance is not entirely clear when trying to consider the implications of this indicator, e.g. this section</p> <p>1.5 Use the Simon Broome criteria (see appendix F of the full guideline) or Dutch Lipid Clinic Network (DLCN) criteria to make a clinical diagnosis of FH in primary care settings. This should be done by a</p>	<p>Thank you for your comment.</p> <p>The indicator is based on NCD102 from the Network Contract DES 2023/24 and would use the same codes for "assessed for familial hypercholesterolemia":</p> <ul style="list-style-type: none"> <li>Simon Broome diagnostic criteria for familial hypercholesterolaemia result (observable entity)</li> </ul>

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		<p>healthcare professional competent in using the criteria.</p> <p>Is this what is meant by making an assessment? In addition, how is HCP competency defined in using the criteria, or are there universally available FH services, without restriction criteria, to which GPs can refer? Without knowing this we cannot comment on whether there are system barriers to achieving this.</p> <p>Regarding the consultation question, have NICE modelled the potential workload of addressing historic results and generated a cost/risk: benefit analysis? The cost used must be the cost of delivering the service, not the cost to the commissioner.</p> <p>The guidance also states that it should ideally be a fasting sample. What if there's a historic non-fasting result and subsequent fasted results are below the cut off? We are unclear what the clinical implications of a raised value in this context and whether we would be adding value in carrying out an assessment/referring, if not done at the time, or not?</p>	<ul style="list-style-type: none"> <li>• Assessment using Simon Broome diagnostic criteria for familial hypercholesterolaemia (procedure)</li> <li>• Dutch Lipid Clinic Network diagnostic criteria for familial hypercholesterolaemia score (observable entity)</li> <li>• Assessment for familial hypercholesterolaemia (procedure)</li> <li>• Assessment using Dutch Lipid Clinic Network diagnostic criteria for familial hypercholesterolaemia (procedure)</li> </ul> <p>The committee agreed that in the first years of implementation there could be increased workload implications for primary care and specialist services. The accompanying documentation has been updated to highlight this risk and note that consideration needs to be given if implemented in practice.</p> <p>The committee noted the risks of spurious results being included but balanced this with the potential benefit of increasing diagnosis rates. They recommended that the indicator be reviewed once diagnosis rates improve. The accompanying documentation highlights that personalised care adjustments or exception reporting should be considered to account for situations where the</p>

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			patient declines assessment or if further assessment is not appropriate.