Health Innovation
North East and North Cumbria

CVD lunch and learn session - Familial Hypercholesterolaemia

21st May 2024 12.15-13:00

House Keeping

- Please ensure your microphone and video are turned off during the session. This is to avoid any disruption during presentations and to assist with the quality of the connection.
- If you need to take a break, please feel free to drop off the call at any time and re-join.
- Live captions are available if required.
- The event is being recorded and will be shared.
- Please ask any questions you have through the chat facility. We will try to address questions during the event, but if we don't manage to do this we will follow up after the event.
- If you cannot see the chat, please email your question/s to sarah.black@healthinnovationnenc.org.uk

The Health Innovation Network

The Academic Health Science Network for the North East and North Cumbria has changed its name to Health Innovation North East and North Cumbria (HI NENC).

The new name – which came into effect on 1st October following the start of our new five-year licence – reflects the organisation's key role to continue to support the development and spread of innovation across the region's health service.

But while our name has changed, our vision remains the same: to improve health outcomes, reduce inequalities, and boost the regional economy. Working alongside partners across the system, we will continue to accelerate health innovation in the region, and beyond.

Established in 2013 by NHS England we are one of 15 Health Innovations.

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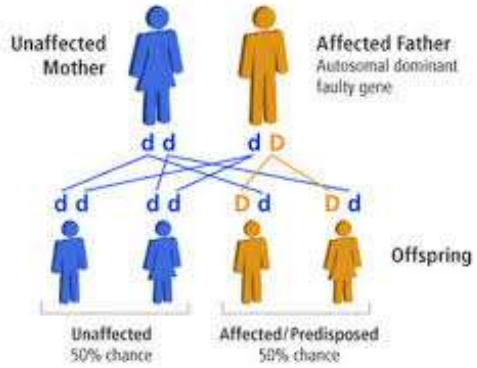
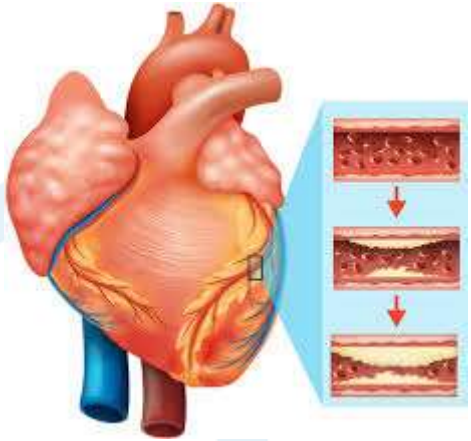
Familial Hypercholesterolaemia

Stewart Pattman, Consultant Chemical Pathologist
Catherine Tucker, Senior Clinical Pharmacist

Northumbria NHS Foundation Trust Lipid Clinic

Familial Hypercholesterolaemia (FH)





- Autosomal dominant
- Family History important

- 1 in 250
- 208

Doubles cholesterol
Early vascular disease

FH national drivers

NICE National Institute for Health and Care Excellence

NICE guideline

Familial hypercholesterolaemia: identification and management

Clinical guideline
Published: 27 August 2008
Last updated: 4 October 2019

www.nice.org.uk/guidance/cg71

NHS

The NHS Long Term Plan

CHOL001 Percentage of patients on the QOF Coronary Heart Disease, Peripheral Arterial Disease, Stroke/TIA or Chronic Kidney Disease Register who are currently prescribed a statin, or where a statin is declined or clinically unsuitable, another lipid lowering therapy	14	70-95%
CHOL002 Percentage of patients on the QOF Coronary Heart Disease, Peripheral Arterial Disease, or Stroke/TIA Register, who have a recording of non HDL cholesterol in the preceding 12 months that is lower than 2.5 mmol/L, or where non HDL cholesterol is not recorded a recording of LDL cholesterol in the preceding 12 months that is lower than 1.8 mmol/L	16	20-35%

FH – how are we doing with identification?

FH identification – May 2024

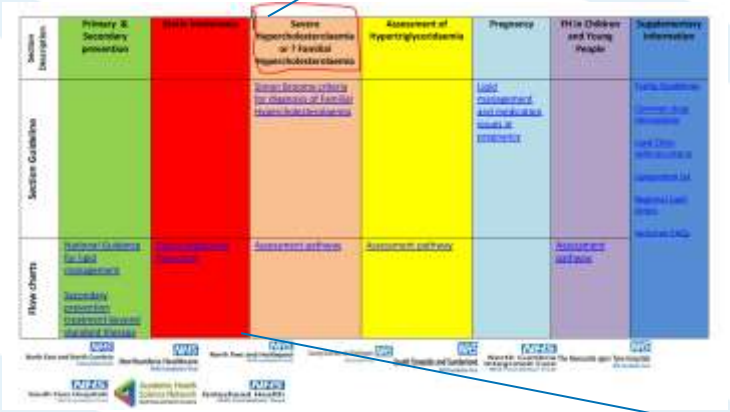
NHS

North East Quality Observatory Service

	Service 4					Comparators from various time points (not routinely updated)				
	To Nov 2020	1 Nov 2020 to 31 Mar 2021	1 Apr 2021 to 31 Mar 2022	1 Apr 2022 to 31 Mar 2023	1 Apr 2023 to 31 Mar 2024	England	NI	Wales	Scotland	UK
Year testing commenced						historic	2000	2005	2008	historic
% patients identified (using 1 in 270 estimate using ICB September 2022 population)	**8.6%**	**9.0%**	**9.8%**	**11.3%**	**12.7%**	**5.8%**	**21.4%**	**12.2%**	**10.6%**	**7.7%**
Index patient with **positive** genetic testing result	448	26	48	62	67	3,059	343	643	1,081	5,126
Index patient with **negative** or variant of uncertain significance (VUS) genetic testing result	745	77	138	252	293	8,423	2,797	2,297	5,942	19,459
Diagnostic yield	**37.6%**	**25.2%**	**25.8%**	**19.7%**	**18.6%**	**26.6%**	**10.9%**	**21.9%**	**15.4%**	**20.9%**
Positive relatives	554	22	49	117	97	3,232	1,136	792	1,028	6,188
Negative relatives	538	23	56	95	110	3,174	1,237	661	1,073	6,145
Relatives tested per positive index patient	2.4	1.7	2.2	3.4	3.1	2.1	6.9	2.3	1.9	2.4
Positive relatives per positive index patient	1.2	0.8	1.0	1.9	1.4	1.1	3.3	1.2	1	1.2
Total positive tests	**1,002**	**48**	**97**	**179**	**164**	**6,291**	**1,479**	**1,435**	**2,109**	**11,314**

Source: NEQOS

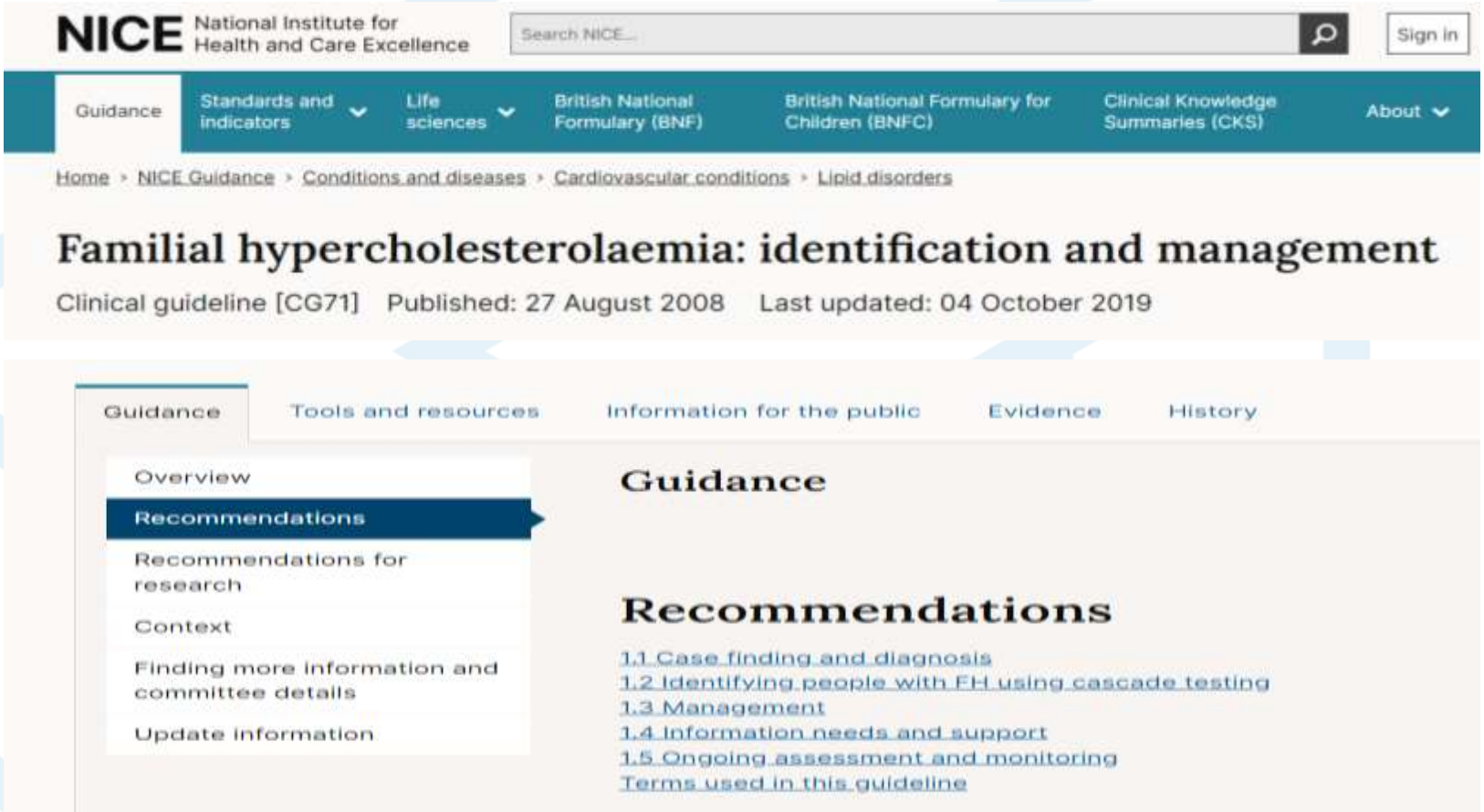
FH – Identification of patients



Severe Hypercholesterolaemia or ? Familial Hypercholesterolaemia

Simon Broome criteria for diagnosis of Familial Hypercholesterolaemia

Assessment pathway


NICE National Institute for Health and Care Excellence
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Familial hypercholesterolaemia: identification and management
Clinical guideline [CG71] Published: 27 August 2008 Last updated: 04 October 2019
Guidance
Tools and resources
Information for the public
Evidence
History
Overview
Recommendations
Recommendations for research
Context
Finding more information and committee details
Update information
Guidance
Recommendations
1.1 Case finding and diagnosis
1.2 Identifying people with FH using cascade testing
1.3 Management
1.4 Information needs and support
1.5 Ongoing assessment and monitoring
Terms used in this guideline

Primary care: Digital searches

UCLPartners

ardens
HEALTHCARE INFORMATICS

Name	Population Count	%
FH Case Finding - Combined (DLCNS >=5 Adj OR NICE Plus Adj)	54	1%
FH Case Finding: 0. DLCNS Possible FH >=5 TG Adj OR >8 #	32	1%
FH Case Finding: 1. DLCNS Definite FH >6 TG Adj #	4	1%
FH Case Finding: 1. DLCNS Definite FH >8 TG Adj #	1	1%
FH Case Finding: 1. DLCNS Definite FH>8 #	1	1%
FH Case Finding: 2. DLCNS Probable FH 6-8 #	27	1%
FH Case Finding: 2. DLCNS Probable FH 6-8 TG Adj #	16	1%
FH Case Finding: 3. DLCNS Possible FH 5 #	33	1%
FH Case Finding: 3. DLCNS Possible FH 5 TG Adj #	15	1%
FH Case Finding: 4. DLCNS Possible FH 3-4 #	437	4%
FH Case Finding: 5. DLCNS Possible FH 3-5 #	470	4%
FH Case Finding: NICE Total Plus	109	1%
FH Case Finding: NICE Total Plus (taking TGs into consideration)	46	1%
FH Case Finding: Simon Broome 1 - Definite FH #	0	0%
FH Case Finding: Simon Broome 2 - Possible FH #	40	1%

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Lipids, Familial Hypercholesterolaemia, PCSK9i & Inclisiran Overview

Report Name	Report Returns	Action
? 2.0 Case Finding – Consider screening for familial hypercholesterolaemia	Patients who have a significant chance of familial hypercholesterolaemia	Screen for FH – see below
? 2.01 Case Finding – Consider screening for familial hypercholesterolaemia (also eligible for IF FH referral)	Patients in 2.0 who are also appear in the IF CVD04 'consider for FH assessment' denominator	Screen for FH – see below
? 2.02 Case Finding – Consider screening for familial hypercholesterolaemia – highest risk patients	Patients in 2.0 who are at highest risk of FH – for areas with limited resources – concentrate on these patients	Screen for FH – see below
? 2.1 Case Finding – Eligible for IF FH referral but FH less likely	Patients who appear in the IF CVD04 'consider for FH assessment' denominator, who are less likely to have FH	Screen for FH – see below; but likely to have a secondary cause of hyperlipidaemia
? 2.2 Case Finding – Code for FH but not genetic code – consider need for genetic testing	Patients with a code suggesting FH e.g. *Possible FH* who don't have a definitive FH code	Review record and consider: Adding definitive code if appropriate Referral for genetic testing Removal of code if incorrect – e.g. secondary hyperlipidaemia

Example based on PCN of 50,000

? Lipids 1 CONSIDER STARTING/RESTARTING LIPID LOWERING		
? Lipids 2.2 Case Finding - Code for FH but not genetic code - consider need for genetic testing	59	0.1%
? Lipids 2.1 Case Finding - Eligible for IF FH referral but FH less likely		
? Lipids 2.02 Case Finding - Consider screening for familial hypercholesterolaemia - highest risk patients	77	0.2%
? Lipids 2.01 Case Finding - Consider screening for familial hypercholesterolaemia (also eligible for IF FH referral)		
? Lipids 2.0 Case Finding - Consider screening for familial hypercholesterolaemia #	321	0.6%
? Lipids 2 SCREENING FOR FAMILIAL HYPERCHOLESTEROLAEMIA		

Recognising Familial Hypercholesterolaemia:

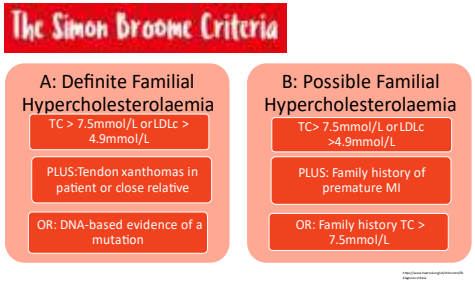
using searches……

Desktop review

- Check clinic letters for previous lipid clinic involvement: coding not always accurate/ specific
- Highest cholesterol result ever (even if on treatment now)

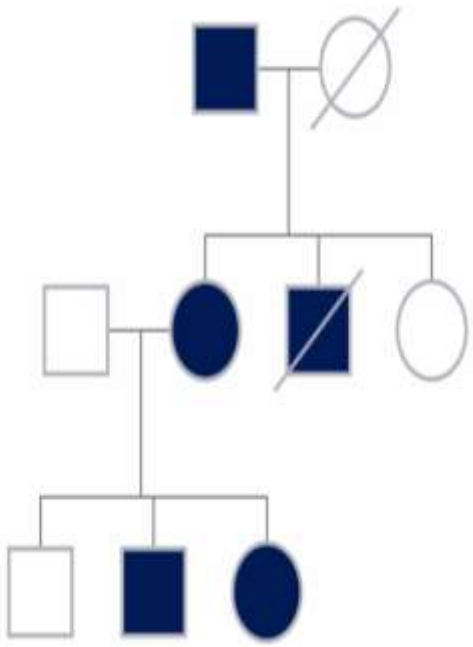
Discuss

- Personal and family history of CVD (MI)<60y
- Exclude secondary causes
- Use Simon Broome criteria: refer to lipid clinic query FH



Communication of genetic test results

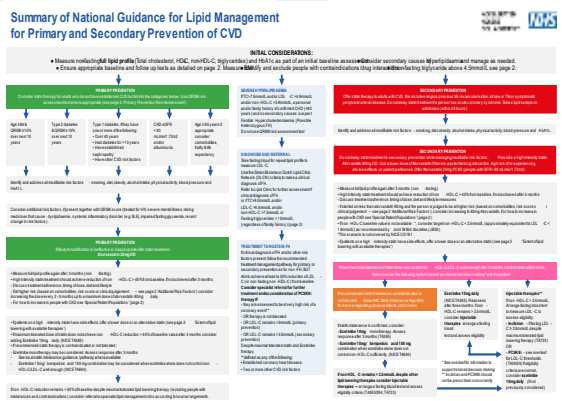
- Centre for Life Genetic service:
  - Results letter to GP / Lipid clinic
- Clearer coding on Lipid clinic letters
  - support coding in primary care record
  - digital searches rely on coding
- Ongoing treatment and follow up
- Family cascade



On going treatment: Lipid targets

- Primary or secondary prevention?

- Document a cholesterol target
  - Primary prevention
    - NICE (2014) : 40% reduction in non-HDL cholesterol
  - Secondary prevention
    - JBS-3 (2013): Non-HDL-c < 2.5mmol/L



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NHS England Lipid management pathway (the blue section- FH)

TREATMENT TARGETS IN FH

If clinical diagnosis of FH and/or other risk factors present, follow the treatment pathway for
primary or secondary prevention

Aim to achieve at least a 50% reduction of LDL-C (or non-fasting non-HDL-C) from baseline.

Consider specialist referral for further treatment and/or consideration of PCSK9i therapy IF

- assessed to be very high risk of a coronary event**
- OR therapy is not tolerated
- OR LDL-C remains >5mmol/L (primary prevention)
- OR LDL-C remains >3.5mmol/L (secondary prevention)

Despite maximal tolerated statin and Ezetimibe therapy.

FH Annual Review (NICE CG71)

primary or secondary care

- Cholesterol treatment target
- Access to further medications:
  - PCSK9i injections
  - Medication: side effects/ adherence
  - Pregnancy/ breastfeeding: need a plan to stop statin and when to re-start
- Low threshold for CVD investigations:
  - Chest pain/ SOB
- Check for other risks (lipoprotein a)
- Lifestyle/ dietary review
- Cascade testing in families: confirm

What are the outputs of the lipid clinic?

Assess for genetic testing

Set lipid target

Optimise/access medication (LDLc>5)

Tips for using the NEELI guideline

Total Cholesterol > 7.5mmol/L and/or LDL-C (fasting) > 4.9mmol/L and/or non-HDL-C > 5.9mmol/L

Need two lipid profiles -Biological and analytical variability

Take fasting blood for repeat lipid profile AND

Blood and urine samples for secondary hyperlipidaemia profile (U+E, LFT, TFT, HbA1c, Urine ACR)

ASSESS

Current drug treatment

Lifestyle including diet (note any fad diets) and physical activity

Alcohol history

Glycaemic control if diabetic

Secondary causes

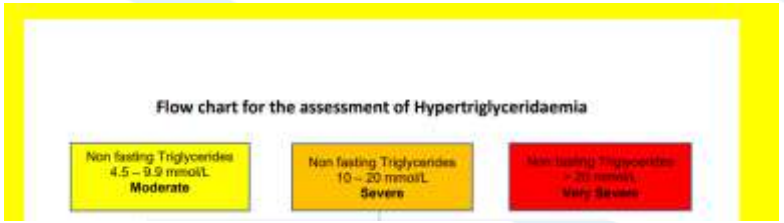
Diabetes – *need to optimise*

Diet – *Heart UK diet leaflet, avoid keto diet*

Drugs – *medications*

Check urine Albumin /creatinine ratio! (vascular risk factor plus high in nephrotic syndrome)

High triglycerides = yellow section



Are tendon xanthomata (visible and/or palpable) present and/or is there a personal and/or family history of confirmed CHD/raised cholesterol

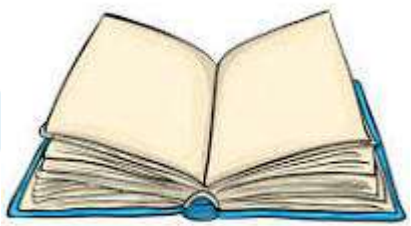
Family history of IHD/high cholesterol crucial
If no knowledge get adult 1st degree relatives cholesterol checked!

1st degree relative with;

- IHD <60yrs
- total cholesterol >7.5mmol/l
- or has a genetic diagnosis of Familial Hypercholesterolaemia

=> Advice & Guidance route is the most flexible

Genetic testing



Variants
- Apo B
- LDL
- PCSK9

Absent
- No cascade testing
- Possible FH diagnosis only, aim to decrease nHDLc by 50% from maximum
- Eligible for PCSK9i if LDLc>5mmol/l

Present
- Family genetic cascade testing via centre for Life
- Genetic FH diagnosis, aim to decrease nHDLc by 50% from maximum
- Eligible for PCSK9i if LDLc>5mmol/l

Advice & Guidance correspondence

Flags other issues

Secondary cause present
Different approach e.g. hypertriglyceridaemia
Possible FH criteria not fulfilled
Pattern of cholesterol not in keeping with FH

Recommend next steps

Still need to risk assess (QRISK), may we worth getting family cholesterol checked, may need to check lipoprotein (a), advice would be given

Resources

RC GP LEARNING

Genomics Education Programme

Health Innovation
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Q&A time

- Any questions?

Upcoming events…

Health Innovation
North East and North Cumbria

Cardiovascular medicines in palliative and end of life care

Monday, 10 June 1-2pm

Optimising the management of patients with Chronic Kidney Disease

Tuesday, 16 July 12-1pm