Health Innovation East Midlands

Local change, national impact

FAMCAT2 – early detection of familial hypercholesterolaemia

Tina Kundi, Project Manager – Health Innovation East Midlands

14th October 2025

Welcome

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- Today's session will last approx. one hour – with approx. 40mins presentation, and time at the end for Q&A.
- Please keep your cameras and microphones off until the Q&A.
- Enter your questions in the chat as we go, and they will be answered at the end.
- We will record the webinar to share afterwards, along with the slides.

Agenda

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- About FH (Kerry Oliver)
- The evidence base for FAMCAT2 (Prof. Nadeem Qureshi)
- FAMCAT2 implementation options and case studies (Kerry Oliver)
- Norfolk and Waveney Pilot recording (Dr Javier Gomez & Shelina Rajan)
- Q&As (joined by Dr Sarah Rodgers and Dr John Robinson)
- Close

About us

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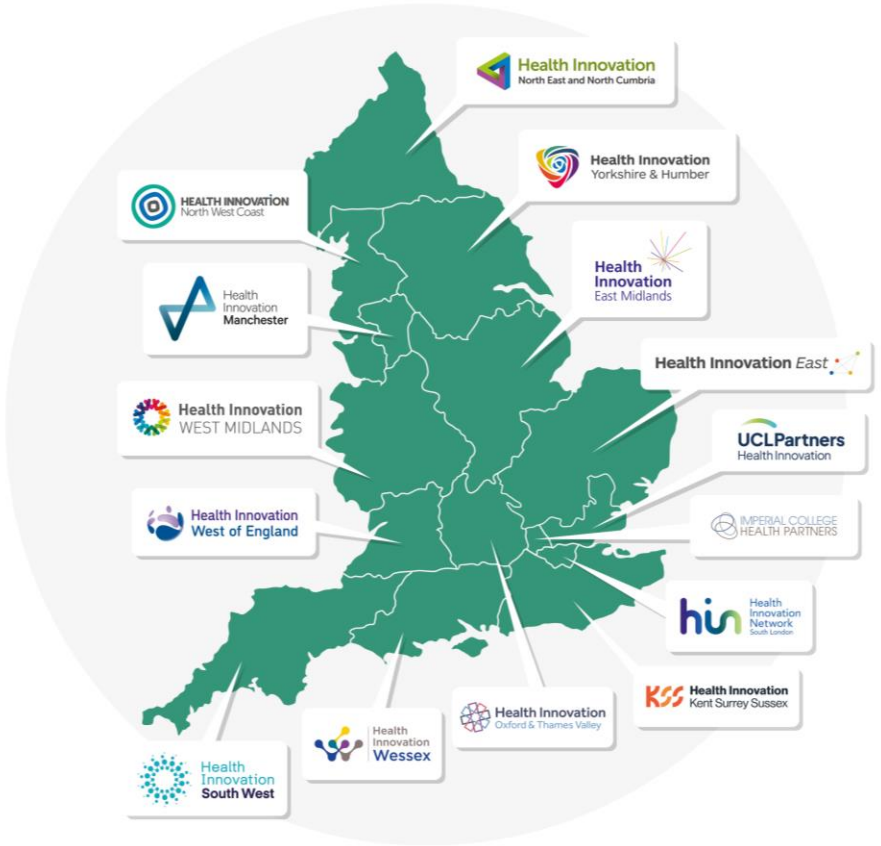
- HIEM is one of 15 Health Innovation Networks (HINs) – commissioned to bring together the NHS, industry, academia and the third sector to help drive health innovation across the NHS, at pace and scale.
- We work both nationally across the network, and locally with health and care systems.
- Each health innovation network is fully-embedded in their local health and research ecosystem.



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Kerry Oliver, PRIMIS, University of Nottingham

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About FH

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- Familial hypercholesterolaemia (FH) is a highly prevalent, treatable genetic disorder that increases the likelihood of cardiovascular disease (CVD) at a premature age.
- However, only 20% of FH individuals identified in the UK.
- There is a need for the identification and treatment of FH.
- Requires cost-effective and accurate approaches to casefinding.

NICE recommendations & issues

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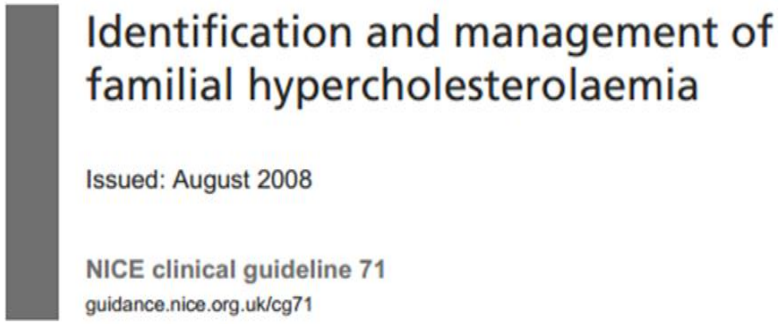
NICE National Institute for Health and Care Excellence



- **Simon Broome criteria**: workload and high referral rates to lipid clinic (11% detection rate)
- **DLCN criteria**: workload (examination) (35% detection rate)
- **Total cholesterol** > 9 mmol/l in 30 years+ or > 7.5mmol/l < 30 years: low false positive but high false negative (< 28% detection rate)

FAMCAT2: new evidence based algorithm

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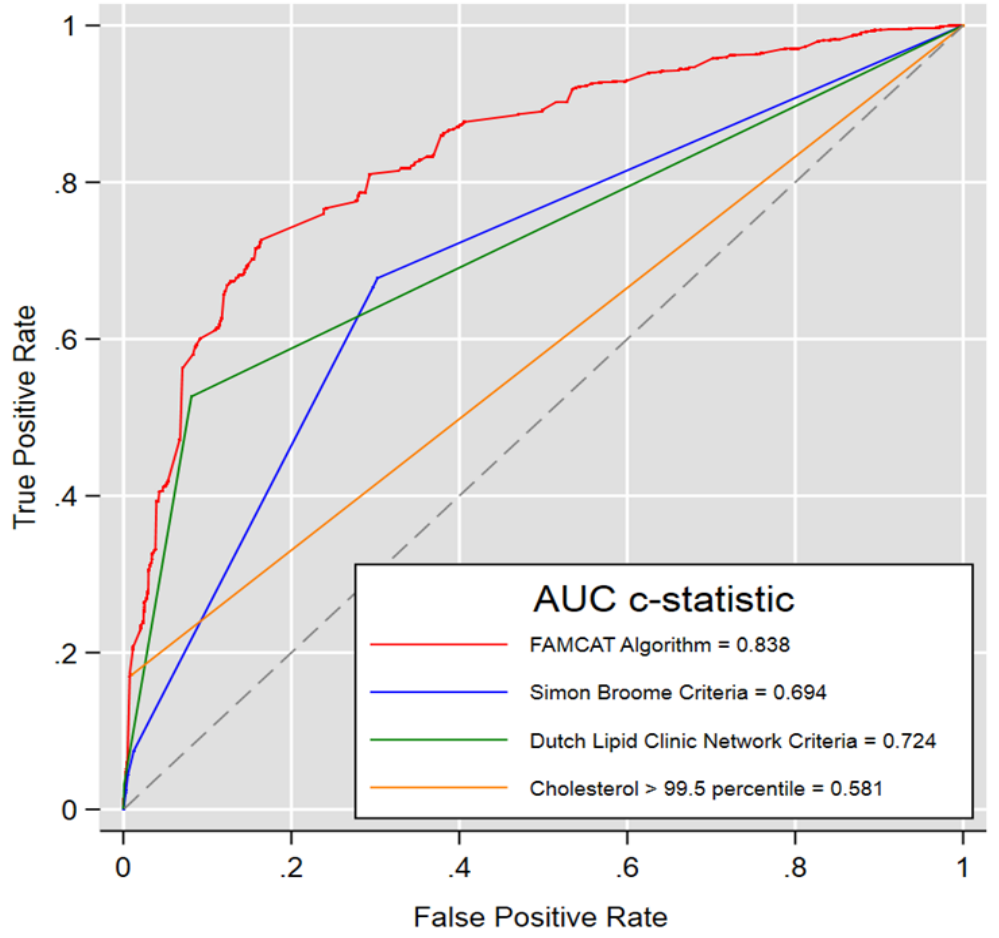
- **Superior performance** to SB, DLCN, Total cholesterol
- Improved **detection rate** (baseline 46% detection rate)
- Prioritise **workload & referrals** by adjusting FAMCAT threshold
- **Cost effective** approach



Evidence base

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- Electronic casefinding and genetic testing in primary care could improve identification of FH; and the better targeting of patients for specialist assessment. http://dx.doi.org/10.1136/heartjnl-2021-319742
- FAMCAT identifies FH with greater accuracy than currently recommended approaches. https://doi.org/10.1016/S2468-2667(19)30061-1
- FAMCAT algorithm had significantly better sensitivity than DLCN, MEDPED, and Simon Broome (SB) criteria. FAMCAT has the best potential for FH casefinding using EHRs. https://doi.org/10.1093/ehjdh/ztac059
- Large population cohort study of >1 million patients confirmed better performance of FAMCAT, compared with other approaches for casefinding of FH in primary care, such as SB criteria, DLCN criteria or very high cholesterol levels. https://doi.org/10.3399/bjgpopen20X101114
- In primary care, in patients with documented cholesterol, FAMCAT2 performs better than other casefinding criteria for detecting genetically confirmed FH, with no prior clinical review required for casefinding. https://dx.doi.org/10.1136/openhrt-2021-001752
- In terms of cost-effectiveness, SB identified the most FH cases but required 102 genetic tests to identify one FH patient. FAMCAT2 identified fewer but only required 23 genetic tests. FAMCAT2 considerably cheaper and a good combination of sensitivity and specificity. https://doi.org/10.3390/jpm12030330

About FAMCAT2

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- FAMCAT2 is a validated clinical algorithm developed by academics at the University of Nottingham that identifies individuals with the highest probability of having familial hypercholesterolemia (FH).
- It uses routinely collected primary care data to calculate the FAMCAT2 score and ranks patients based on their likelihood of having FH.
- It supports early detection and intervention, aligning with national cardiovascular prevention strategies and enhancing patient outcomes.
- FAMCAT2 is best deployed at population level.

Flexible implementation

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PRIMIS supported the research and has led on developing approaches to implementation, including compliance with medical device regulations:

1. Practice-based model:
   - Available as a standalone tool for use within the EMIS Web clinical information system.

2. Integrated model:
   - Using an Application Programming Interface (API).
   - Or replicate the algorithm directly within their systems.

Case studies

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1.

Deploying the FAMCAT2 algorithm to accurately identify patients at high risk of FH in Wessex – A GP practice case study.

https://healthinnovationwessex.org.uk/img/projects/FAMCAT%20Case%20study%20Final%2012-7-24.pdf

Key findings:

- The FAMCAT2 Tool was well received by 55 GP practices using EMIS Web.
- Use of the tool led to the identification of over 262 patients with a confirmed diagnosis of FH who could be treated to prevent future cardiac related events.
- Led to not only an increase in the diagnosis of FH but also raised awareness of FH amongst primary care health care professionals.

2.

Pilot to permanent: the FH Hub's success story

https://healthinnovationeast.co.uk/pilot-to-permanent/

Key findings:

- 141 patients underwent genetic testing, with 24 (17%) confirmed to have FH.
- Without risk stratification, identifying the same number of FH cases would have required screening 6,250 people, a far greater strain on NHS resources.
- Identifying one positive FH case for every six genetic tests – outperformed comparative approaches.
- Today the FH Hub is a permanent service supporting the Norfolk & Waveney population

Feedback

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"A real life-saver! The FAMCAT2 tool provided the impetus and process for practices to look at patients with a diagnosis of FH as well as find those who were undiagnosed. It raised awareness of this genetic condition and put in place safety mechanisms in primary care to save lives. A win-win in busy practices!"

Liz Corteville – Deputy to Associate Director Medicines Optimisation, Hampshire and Isle of Wight ICB

"Using the FAMCAT2 evidence-based tool to identify and prioritise patients for review and referral for FH has helped us to find a significant number of patients in Hampshire who we can now support to effectively manage their FH."

Rachel Howard – Head of Medicines Optimisation, Hampshire and Isle of Wight ICB

"Identifying a high percentage of FH patients demonstrates that using the FAMCAT2 algorithm embedded within ECLIPSE on behalf of primary care is a cost-efficient and effective model of FH casefinding."

Shelina Rajan, Clinical Nurse Specialist in FH, Norfolk and Norwich University Hospitals NHS Foundation Trust

Key advantages

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- Enables proactive casefinding rather than opportunistic detection.
- Earlier identification of FH cases leading to improved outcomes for patients and their families.
- Helps avoid unnecessary referrals and invasive tests, reducing patient worry and stress.
- High degree of accuracy, granularity, and specificity.
- Cost-effective solution that maximises resources and reduces unnecessary testing and referrals.
- Enables alignment with genomic service capacity.
- Promotes a standardised approach to referral and supports equity of access.
- Designed for systematic, large-scale use across entire practice populations or systems.
- Recommended case finding approach by Genomics Education Programme.

Insights

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- One size doesn’t fit all.
- Flexible approach.
- Involvement of the key stakeholders in the existing pathway.
- Primary care data quality.
- Population level implementation.
- Translating evidence-based research into everyday practice.
- Academic, clinical, and technical maintenance.

FAMCAT2

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Norfolk and Waveney example

Dr Javier Gomez, Consultant Chemical Pathologist & FH Service Lead

Shelina Rajan, Clinical Nurse Specialist - FH

Norfolk and Norwich University Hospital

Norfolk and Waveney pilot

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Thank you

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- Please complete the poll to tell us what you thought about today's webinar

Thank you

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- And don’t forget to click on the QR code to register your interest in a follow up call.

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Any questions?

Please raise your hand or add them to the chat

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Further information

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