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Health Innovation North East and North Cumbria

Improving access to FH Genetic Testing: GENinCode

capacity.

FH is a potentially preventable cause of premature coronary artery disease and stroke, yet most affected people remain unidentified. The NHS Long Term Plan aims to identify 25% of all FH patients in England, through genetic testing, the gold standard for diagnosing this condition. However, health system structures and capacity issues present challenges for meeting this goal. We sought to evaluate whether genetic testing offered by a commercial provider offers advantages in testing efficiency compared to the current clinical standard, offered through the NHS Genomics Laboratory Hub (GLH) at a reduced cost to the NHS, with comprehensive testing and a fast and efficient reporting time frame.

Since May 2023 GENinCode have provided the NENC GLH with the following support enabling them to begin to meet the NHS 10 year plan:

Total number of FH samples tested: 1334

Total number of NENC referring clinicians supported: 37

Number Positive (pathogenic) FH patients diagnosed: 189

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> We are delighted to support the NHS North of England through our collaboration with Health Innovation North East and North Cumbria by reducing the cost of genetic testing and improving turnaround times to identify individuals in the population with high levels of cholesterol FH giving rise to accelerated onset of heart disease over their lifetime. By identifying FH patients earlier in life we can effectively educate, manage and treat patients to reduce their risk of heart attack, heart disease and stroke.
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> **— MATTHEW WALLS, CEO GENINCODE**

Challenge/Problem Identified

Familial Hypercholesterolemia (FH) is a hereditary disorder characterised by markedly elevated levels of low-density lipoprotein cholesterol (LDL-C), predisposing affected individuals to premature cardiovascular events. Despite its significant impact on public health, under diagnosis and delayed intervention remain persistent challenges. Identifying those with FH and treating the associated high lipid levels reduces cardiovascular disease risk dramatically.

Overview of innovation

By early identification of those suffering with FH, the test identifies a patient cohort at high risk of a cardiovascular event who will benefit most from lipid reduction/optimisation. Ultimately this leads to more precise and efficient treatment targeting those individuals most at risk through a reduction of cholesterol.

Action taken

Results from a baseline audit explored clinician and patient experience with the testing technology 'LIPID inCode' provided by GENinCode, a commercial FH test which provides an alternative to the current service delivered via the Genomic Laboratory Hubs.

This led to two pilots aimed at understanding 'direct to test' delivered in primary care and secondary care and one pilot study from the secondary care aimed at reducing the lipid clinic waiting list. The aim in all 3 studies was to reduce clinic waiting times using a test that costs less than the current NHS test, provides comprehensive test results with faster turnaround times and in-depth reporting capabilities.

Working collaboratively with local clinicians we have supported the implementation of LIPID inCode and the development of bespoke reporting to meet the needs of the NHS North of England clinical teams. This has allowed the North of England to begin to meet its NHS 10 Year Targets for FH population testing (current total to June 2024 – 1334 tests).

In addition, we have undertaken and supported three pilot projects:

1. The acceptability of LIPID inCode testing for patients and clinicians in secondary care. LIPID inCode can also deliver FH testing using both saliva and blood (in addition to conventional blood only). This was highly acceptable to patients (particularly as it allowed test kits to be sent via the post). Clinicians found the reports both acceptable and informative. Turnaround times were much improved compared to samples sent to the GLH.
2. A primary care pilot was performed where those at risk of FH were identified from primary care searches. Direct to test was offered via a GP with a special interest in cardiology. This pilot, although successful in its aim and purpose, suggested that further work is needed to refine the eligibility criteria for direct to test before widespread adoption of FH testing in primary care.
3. Waiting lists seen in secondary care lipid clinics are long. To address this, we piloted an opportunity to offer direct testing to those referred as ' Questionable FH' who were on the waiting list for the secondary care lipid clinic as a means of reducing waiting times.

Outcome

The case studies and results showed successful delivery of the objectives and the LIPID inCode test has now been implemented in the NHS North of England (Newcastle. Leeds and Sheffield).

The LIPID inCode NHS test results are now being provided to physicians as part of the normal NHS FH services. The results of the LIPID inCode clinical adoption have enabled the NHS North of England to begin to meet its 10 Year Plan targets as well as providing physicians with fast turnaround of patient test results and more comprehensive (hypercholesterolemia related) risk analysis.

LIPID inCode enables the NHS to identify patients at high risk of CVD earlier as well as providing additional polygenic hypercholesterolemia information enabling more precise and efficient (pharmacogenomic) treatment with testing provided at a lower cost than in-house NHS testing.

In LIPID inCode testing carried out by GENinCode in the past 15mths the following data has been recorded:

		Number
P VARIANTS, FH POSITIVE	Pathogenic Likely	189
LP VARIANT, FH probable	Pathogenic	65
VUS	VUS	21
NMD	NMD	1059
ARH		0
LALD		
Total		1334

Impact

More effective use of limited NHS expert resource and prioritisation:

Release of limited expert resources within the GLH; these resources can be used for other NHS disease areas requiring prioritisation of limited expert resource.

e.g. A sample run of 25-30 FH samples would be expected to release:

- 3 full lab days for 2 Genetic Technicians
- 3-4 full days for 2 Clinical Scientists (longer if more comprehensive analysis of new variants is required).

Comprehensive Test Service:

Unlike the current NHS standard Monogenic FH report, GENinCode LIPID inCode test includes

- Monogenic diagnosis
- Hypercholesterolaemia Polygenic Risk (LDL-C)
- Cardiovascular Risk Stratification
- Predisposition to raised levels of LP (a)
- Pharmacogenetics of Statins

Faster turn around times (TAT):

NHS: TAT 3-4 months from sample in to report out

GENinCode: 10-15 days from sample in to report out

Online direct reporting and online report repository for NHS clinicians

Most cost effective:

GENinCode provide LIPID inCode FH testing service at a reduced cost and well below that of the NHS equivalent testing.

Plans for the future / spread and adoption

The pilot project demonstrates the feasibility of using a simple algorithm to identify patients with a high pre-test probability of FH and the use of postal saliva samples for genetic index testing, allowing best use of limited Lipid clinic resources.

Incorporating Lipoprotein (a) measurement and Coronary Genetic risk scores maximises identification of patients at increased risk of CVD

Genetic testing for FH provided by a commercial partner, GENinCode Plc, is feasible, efficient, accurate, and represents a viable option for increasing NHS genetic testing capacity, in order to meet the NHS Long Term Plan target for identification and treatment of people with FH.

- The use of LIPID inCode has been adopted as the FH diagnostic for NENC. This has been delivered with clinician support via the Northern Lipid Forum.
- Two abstracts have recently been presented at the HEART UK conference 2023/2024
- Discussions are on-going with other GLH's

The NHS North of England implementation of LIPID inCode and successful case studies provide growing evidence of the need to cross apply LIPID inCode testing to other areas of NHS England to help support these areas achieving the NHS 10 Year Plan. With faster turnaround times and delivery at lower cost than the NHS, it would also release NHS/GLH expertise to prioritise in other areas.

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