NICE Shared Learning Awards 2019

Royal Brompton & Harefield NHS
NHS Foundation Trust

A telephone appointment service to improve access to familial hypercholesterolaemia cascade screening

20 YEARS OF NICE
1999-2019

First, second and third-degree biological relatives of people with familial hypercholesterolaemia (FH) should be offered cascade testing, as recommended by NICE. In 2012, when this project was launched, genetic testing centres in the UK for FH were limited.

To make sure that relatives could access services and not be missed because of geographical location, we started a telephone cascade screening service specifically for relatives of those who already have a diagnosis of FH.

"Telephone cascade testing is accessible and convenient, increasing the detection rate of FH."

Laura Davis, FH clinical nurse specialist,
Royal Brompton and Harefield NHS foundation Trust

What we did and why

In 2012, the provision of genetic testing centres for FH across the UK was limited. To access these services many patients had to travel long distances with time and cost implications, or were not able to attend at all. The ability to cascade screen family members of patients with a molecular diagnosis of FH was difficult to implement because of the dispersion of families throughout the UK.

We aimed to:

- Increase the diagnosis of FH by offering a convenient way for patients to access cascade screening (NICE CG71).
- Enhance the continuity and quality of care for patients who need cascade screening (NICE CG71)
- Reduce the need for venepuncture for the comfort of patients.

A telephone appointments service for cascade screening was started, using a buccal swab for DNA collection. We:

- Sought trust approval.
- Liaised with a genetic laboratory to extract DNA from buccal swabs.
- Sourced buccal swabs.
- Sourced packaging compliant with P1650 packaging instructions and UN3373 regulations to transport specimens.
- Created an appointment pack.

Outcomes and impact

In 2011, before the implementation of the telephone appointment service, 25 relatives of DNA FH-positive patients were screened. In 2013 we saw a 168% increase, which has continued to increase with further service expansion.

Since it's introduction, 551 relatives have been screened, 185 of these have a confirmed molecular diagnosis of FH. This increases opportunities to reduce the risk of premature cardiovascular disease with treatment and lifestyle advice in this high risk group.

Patient satisfaction levels from everyone who took part in this cascade screening service:

- 96.8% of respondents were happy with the appointment method.
- 3.2% did not answer.

Patient comments:

"Saved time off school."

"Process works well – quick, simple and efficient."

"Avoided travel to Harefield."

"Much easier."

Clinical commissioning groups benefited from an 84% reduction in appointment cost.

What we learnt

- The telephone appointment system provides a convenient, cost-effective method of genetically screening relatives for FH, regardless of geographical location.
- Uptake of FH genetic screening increases.
- A systematic approach and continued care for a family is possible.
- Disruption to everyday life is minimised with reduction in time off work, school and higher education.
- Multiple members of the same household can be screened at the same time using a speakerphone facility.
- Venepuncture can be avoided through the use of buccal swabs.
- Buccal swabs are an effective method of DNA collection for FH cascade screening.
- Patients like this appointment method.

NICE National Institute for Health and Care Excellence

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