Lynch syndrome and cascade screening

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What is Lynch syndrome?

Bowel Cancer UK
Beating bowel cancer together

- Lynch syndrome is a genetic condition which increases a person's risk of developing bowel cancer by up to 80% as well as increasing the risk of other cancers
- It is estimated to cause around 3% of bowel cancer cases in the UK every year, many of them in people under the age of 50.
- Yet currently, less than 5% of people with Lynch syndrome have been diagnosed
- Around 175,000–200,000 people across the UK have Lynch syndrome and are unaware

What causes Lynch?

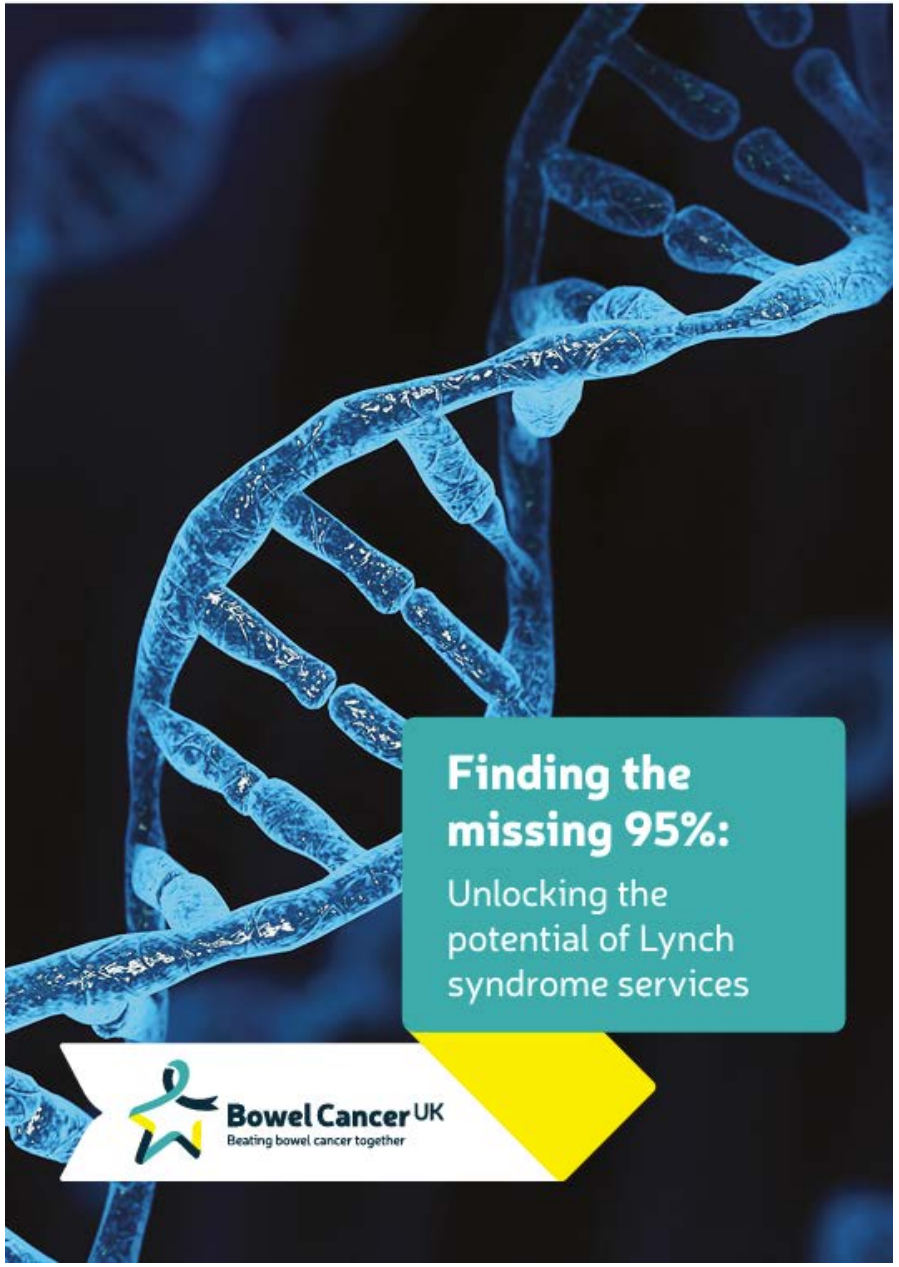
- Mismatch repair (MMR) genes are responsible for fixing DNA errors in the body
- If these genes have faults, DNA mistakes can build and lead to cancer over time
- Around 15% of people with bowel cancer are estimated to have defects to these genes
- Approximately 25% of those with MMR defects have Lynch syndrome
- Because these faults can happen in cancers unrelated to Lynch syndrome, two more tests are carried out
- If no changes are found, further testing of DNA called germline testing, is necessary to definitively diagnose Lynch syndrome

Management of Lynch

- Before getting a diagnosis, every patient must undergo genetic counselling
- Genetic counselling is one example of a wraparound care measure. Wraparound care consists of providing information, support, and the network needed to help patients through their Lynch syndrome journey
- To find the missing 95% of undiagnosed people with Lynch, family members of patients who test positive for Lynch should be offered the same genetic tests
- Routine surveillance colonoscopy reduces the risk of dying of bowel cancer by as much as 72% because it can detect bowel cancer at an earlier stage when it is treatable and curable, and in some cases even prevent it from developing
- Some people with Lynch syndrome may benefit from other ‘risk reducing strategies’ – aspirin, hysterectomy etc

NICE guidance

- The National Institute for Clinical Excellence (NICE) is responsible for evaluating the clinical and cost effectiveness of new drugs and treatments
- In 2017, they recommended that everyone diagnosed with bowel cancer is tested for Lynch syndrome, at the time of diagnosis
- Since the initial guidance was published, NICE have also adopted the British Society of Gastroenterology (BSG) and Association of Coloproctology of Great Britain and Ireland's (ACPGBI) recommendations on routine colonoscopic surveillance for those with Lynch syndrome
- This guidance applies to England and Wales, and equivalent guidelines have been endorsed in Scotland and Northern Ireland



Our FOI and 2024 report

Findings

To understand how well the national guidance on Lynch testing and surveillance has been implemented, we sent Freedom of Information requests to health authorities across the UK.

Our 2023 data found that, across the UK:

• Significant improvements in genetic testing with an average of 9 in 10 newly diagnosed bowel cancer patients being tested

•Half of health authorities who responded, reported that family members of people with Lynch syndrome aren't offered letters they can take to their GP, which is one route to accessing genetic counselling and testing

•A postcode lottery for life-saving routine surveillance colonoscopies exists. Only 6 in 10 health authorities across Scotland, Wales and Northern Ireland offer surveillance colonoscopies in line with clinical guidance

•Major gaps in data collection and reporting on Lynch guidance and services are holding back Lynch syndrome care

What we're looking for

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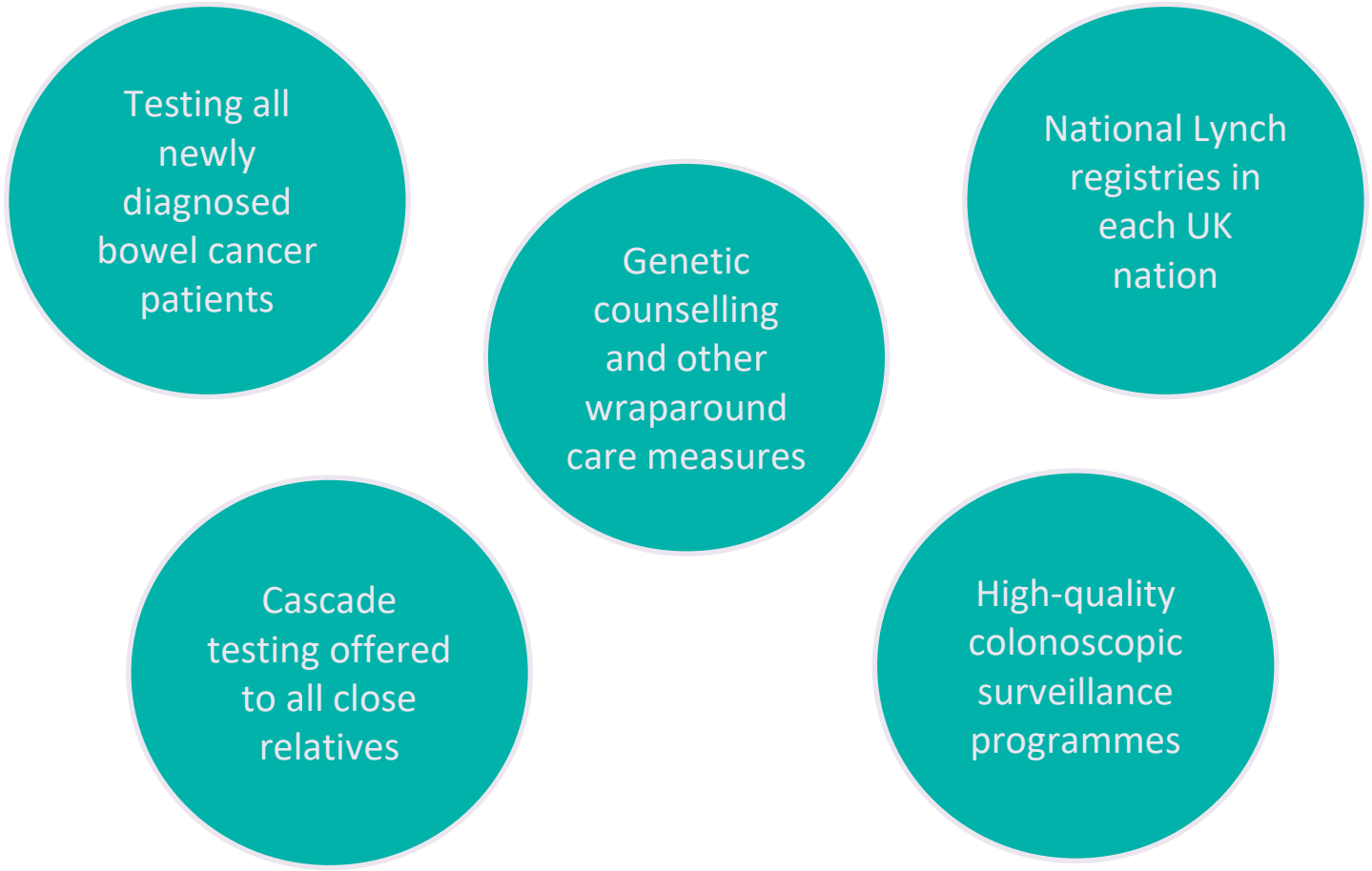

What lies ahead?

A **targeted screening programme for Lynch syndrome in people with colorectal cancer** based on molecular diagnostic tests followed by offering risk-reducing strategies in those who test positive, and for others who are diagnosed with Lynch syndrome **including their family members**.

The purpose of this programme would be to **identify those with the inherited genetic condition** associated with an increased risk of several cancers to improve outcomes in terms of disease specific mortality by **preventing the associated cancers or increasing earlier diagnosis of the disease.** This is predicated on the observation that **survival after treatment of this disease is highly stage dependant**. Risk-reducing strategies to help prevent or increase early diagnosis of associated cancer could include routine colonoscopic surveillance for colorectal cancer.

In the long-term, a secondary outcome would be in **reduction in disease mortality of family members with Lynch syndrome** as cascade testing would allow individuals at high risk to be identified and offered appropriate surveillance and strategies to reduce their risk.

Why screening?

- Screening offers the opportunity to establish a robust framework for the diagnosis, management and support of people affected by Lynch syndrome
- Strengthened data collection alongside nationally agreed programme standards
- Ability to screen a population that have a higher risk of developing colorectal cancer to improve outcomes

Any questions?