

Improving services for Lynch syndrome: who's responsible?



Thank you

At Bowel Cancer UK we strive to ensure that patients are at the heart of everything we do and that we listen to and consult them about their experiences and priorities. This report aims to share the experiences of people diagnosed with Lynch syndrome and we would like to thank the 178 people who responded to our survey and shared their experiences with us. Special thanks must also be given to Lynch Syndrome UK for promoting the survey and Dr Kevin Monahan, Consultant Gastroenterologist at West Middlesex University Hospital, and Dr Fiona Laloo, Consultant Clinical Geneticist at Central Manchester University Trust, for their clinical expertise.

About Bowel Cancer UK

We are the UK's leading bowel cancer research charity and we are determined to save lives and improve the quality of life for all those affected by bowel cancer. We support and enable research, educate patients, public and professionals about bowel cancer and campaign for early diagnosis and best treatment and care for all those affected.

Find out more at bowelcanceruk.org.uk

About bowel cancer

Bowel cancer is the fourth most common cancer in the UK and the second biggest cancer killer, affecting both men and women. Every 15 minutes in the UK someone is diagnosed with bowel cancer. That's over 41,000 people every year. Bowel cancer is treatable and curable especially if diagnosed early. Nearly everyone diagnosed at the earliest stage will survive bowel cancer. However, this drops significantly as the disease develops. Early diagnosis really does save lives.

Contents

If you only have time to take a quick look, we would recommend that you read the executive summary (p.4) and recommendations (p.25).

Executive summary	4
Method	6
Survey results	7
1 Understanding and identifying Lynch syndrome	7
An inherited condition	8
Lynch syndrome cancers	9
2 Testing for Lynch syndrome	11
Molecular screening	12
Genetic counselling	13
Genetic testing	13
Sharing a diagnosis with your family	14
The case for a genetics registry	15
3 Managing Lynch syndrome	17
Access to regular colonoscopy	17
Developing a high quality surveillance programme	18
Aspirin: reducing the risk of bowel cancer	19
4 A personalised approach to care	21
Care plan	21
A dedicated healthcare professional	22
5 Access to information and services	23
Conclusion and recommendations	25

Executive summary

Lynch syndrome (formerly known as hereditary non-polyposis colorectal cancer) is caused by a defect in the mismatch repair (MMR) genes, which usually works to repair damage to DNA. This defect in DNA repair means that people who have Lynch syndrome are much more likely to develop a range of cancers.

Bowel cancer is the most common cancer caused by Lynch syndrome. In some cases, the risk of developing bowel cancer is as much as 80%. An estimated 1,100 cases of bowel cancer can be attributed to Lynch syndrome each year – many of them under the age of 50. Despite this fewer than 5% of individuals with the condition have so far been diagnosed in the UK and many people do not receive the clinical management they need to reduce their risk of developing bowel cancer.

We have undertaken extensive research to understand how people are diagnosed and managed for Lynch syndrome within the healthcare system. This report presents the patient perspective through findings from a comprehensive survey that was carried out in July 2016. In this survey we asked people with Lynch syndrome to share with us their experiences across the care pathway from being initially tested and then diagnosed, to their access to regular colonoscopy and about the information and support they received.

In addition to this survey we have also:

- Worked in partnership with professional bodies to hold an expert meeting with 35 leading UK clinicians and scientists to consider how we can improve the way people with Lynch syndrome are diagnosed and managed.
- Conducted a Freedom of Information (FOI) request to all hospitals in the UK to determine adherence to the Royal College of Pathologists Colorectal Cancer dataset on testing bowel cancer patients under the age of 50 for Lynch syndrome.
- Actively engaged with the NICE guideline development process on molecular testing for Lynch syndrome and were represented on the Committee by our Expert Advisor, Dr Kevin Monahan.
- Held numerous meetings with policy makers, people with Lynch syndrome and professionals to identify barriers and develop solutions.

81%

81% were diagnosed with Lynch syndrome under the age of 50 years

 **30%**

30% had been waiting 4 months to over a year from having a blood sample taken for genetic testing to being informed of their diagnosis

 **11%**

11% waited more than 6 months to see a genetic counsellor

 **87%**

87% would consent to being part of a national registry of people diagnosed with Lynch syndrome

Our survey results show that whilst some people with Lynch syndrome have received exceptional care there are still too many experiencing delays right across the pathway. In particular we found:

- 81% were diagnosed with Lynch syndrome under the age of 50 years.
- 30% had been waiting 4 months to over a year from having a blood sample taken for genetic testing to being informed of their diagnosis.
- 11% waited more than 6 months to see a genetic counsellor before having their genetic test; and 28% were not offered a follow-up appointment once their diagnosis had been confirmed.
- 87% would consent to being part of a national registry of people diagnosed with Lynch syndrome.
- 49% had experienced a delay to their planned colonoscopy with 78% waiting more than 6 weeks beyond their planned appointment date.
- Over 70% had not been provided with a written personalised care plan, which sets out their treatment options.
- 43% had not been given enough useful and relevant information on Lynch syndrome.

To address these issues we are calling for the following recommendations to be implemented as matter of urgency. We believe, if adopted, these recommendations could reduce the significant variation in how people with Lynch syndrome are managed in the UK.

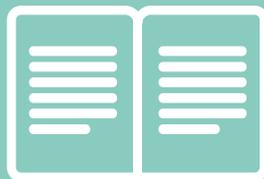
Top recommendations:

- A **national registry** of people identified as having Lynch syndrome must be developed. This should be a systematic process with consent obtained by a clinician at the point of diagnosis. Research linked to the registry would increase our knowledge and understanding of Lynch syndrome, including clarifying how many people are affected by the genetic condition and the regional differences in treatment and care which need to be addressed to improve outcomes.
- A **national surveillance programme** for people with Lynch syndrome should be established to reduce the vast variation in access, quality and timeliness of regular colonoscopy. A surveillance programme, using data from the registry, will need to be quality assured and have recall capabilities so people are sent colonoscopy appointments on time.
- **Comprehensive UK guidelines** should be developed that set out best practice for the clinical management of people with Lynch syndrome.

49%



49% had experienced a delay to their planned colonoscopy with 78% waiting more than 6 weeks beyond their planned appointment date



70%

Over 70% had not been provided with a written personalised care plan



43%

43% had not been given enough useful and relevant information on Lynch syndrome

Method

In July 2016 we ran an online self-selecting survey asking people with Lynch syndrome to share with us their experiences of being diagnosed and managed for the genetic condition. The survey was made up of over 55 questions that encompassed the pathway from diagnosis, to treatment options for cancer (if applicable) and their experience of accessing information and support services.

The survey was promoted online across all four nations through the Bowel Cancer UK website, social media channels and emailed to our supporter base. The survey was also promoted through a number of third party organisations, charities and patient forums. This included Lynch Syndrome UK, local Healthwatch organisations, health councils, clinicians and genetic centres.

The survey collection continued for the duration of six weeks. In total we received 178 responses. These responses have helped us create a detailed picture of the experiences of people with Lynch syndrome across the care pathway and helped identify which services require improvement.

All quotes used throughout this report have been taken from the survey.

This report and the final recommendations were further informed by:

- Literature research we conducted.
- Our FOI requests from 2015 and 2016, which identified hospitals that followed Royal College of Pathologists Colorectal Cancer dataset guidelines to test all patients diagnosed with bowel cancer under the age of 50 for Lynch syndrome at diagnosis.
- Recommendations from our clinical expert meeting held in March 2016, which brought together leading clinicians, researchers and people with Lynch syndrome to discuss how these individuals can be better identified and managed.
- Clinical experts were consulted on the survey questions, findings and recommendations and checked the report for clinical accuracy.

Part 1 – Understanding and identifying Lynch syndrome

Lynch syndrome is a genetic condition that can significantly increase the risk of developing bowel cancer by as much as 80%, as well as other cancers such as womb and ovarian. It is estimated that there are 175,000¹ people who have Lynch syndrome in the UK, however less than 5% have been identified². This means 95% do not know they are at significant increased risk of developing cancer over the course of their lifetime.

Lynch syndrome is caused by a change in one of the mismatch repair (MMR) genes, which usually works to repair damage to DNA. Many people with Lynch syndrome may not yet have had a diagnosis of cancer, and those who have a diagnosis of cancer may not have any obvious distinguishable features from other cancer patients. However, Lynch syndrome is usually suspected in the following circumstances:

- 1. A person is diagnosed with a Lynch syndrome related cancer at a young age**
- 2. There is a strong family history of Lynch syndrome-related cancers**

There is clinical guidance in place to assist clinicians to identify this group of individuals. Most notably, these are the Royal College of Pathologists Colorectal Cancer Dataset³, which recommends everyone diagnosed with bowel cancer under the age of 50 is tested for Lynch syndrome, and the Amsterdam/Bethesda guidelines, which sets out the criteria for a strong family history. Our survey shows that 81% of our respondents were diagnosed with Lynch syndrome under the age of 50 years. However Lynch syndrome also commonly affects older people too, which means people over 50 with Lynch syndrome are potentially being missed under current guidelines.

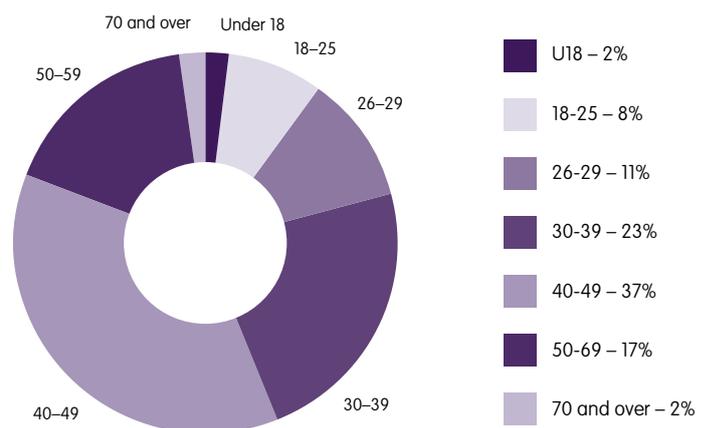
Lynch syndrome cancer risks⁴

- 50–80% bowel cancer
- 30–60% womb (endometrial) cancer
- 10–20% stomach cancer
- 10–12% ovarian cancer
- 5–20% urological cancer

Research from the Mallorca Group⁵ has shown that people with Lynch syndrome also have an increased risk of skin, prostate and breast cancers.

However the risk level varies with the mutation of the genome. In female Lynch syndrome gene carriers the risk of developing endometrial (womb) cancer is also very high. The group recommends an endometrial biopsy or transvaginal ultrasound from the age of 35 but the value of surveillance for endometrial cancer is still unknown.

Age at diagnosis of Lynch syndrome

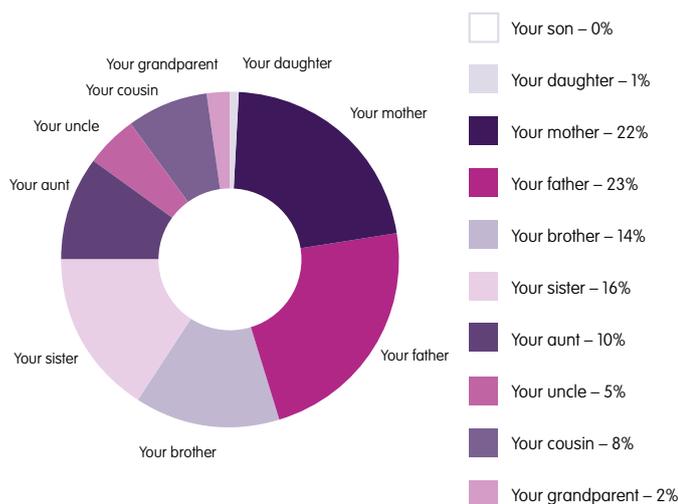


An inherited condition

Lynch syndrome is an inherited condition, with a 50% chance that the children of an affected individual may also have the condition. People have two copies of each MMR gene. If someone has a Lynch syndrome gene mutation they will have one healthy copy of the MMR gene and one faulty copy. As a result there is a 50:50 chance they will pass on the faulty gene. Our survey shows the extent to which Lynch syndrome can affect multiple generations of families, with just over half (52%) stating that other family members had been diagnosed with the genetic condition first:

- 75% a first degree relative (a parent, sibling or child)
- 17% a second degree relative (aunt, uncle, grandparent)
- 8% a third degree relative (cousins)

The first person diagnosed with Lynch syndrome in a family



Amsterdam criteria for family history⁶

The Amsterdam criteria, set by the International Collaborative Group on Hereditary Non-Polyposis Colorectal Cancer, are used to help clinicians assess and identify Lynch syndrome families. All of the criteria below must be met to suspect Lynch syndrome.

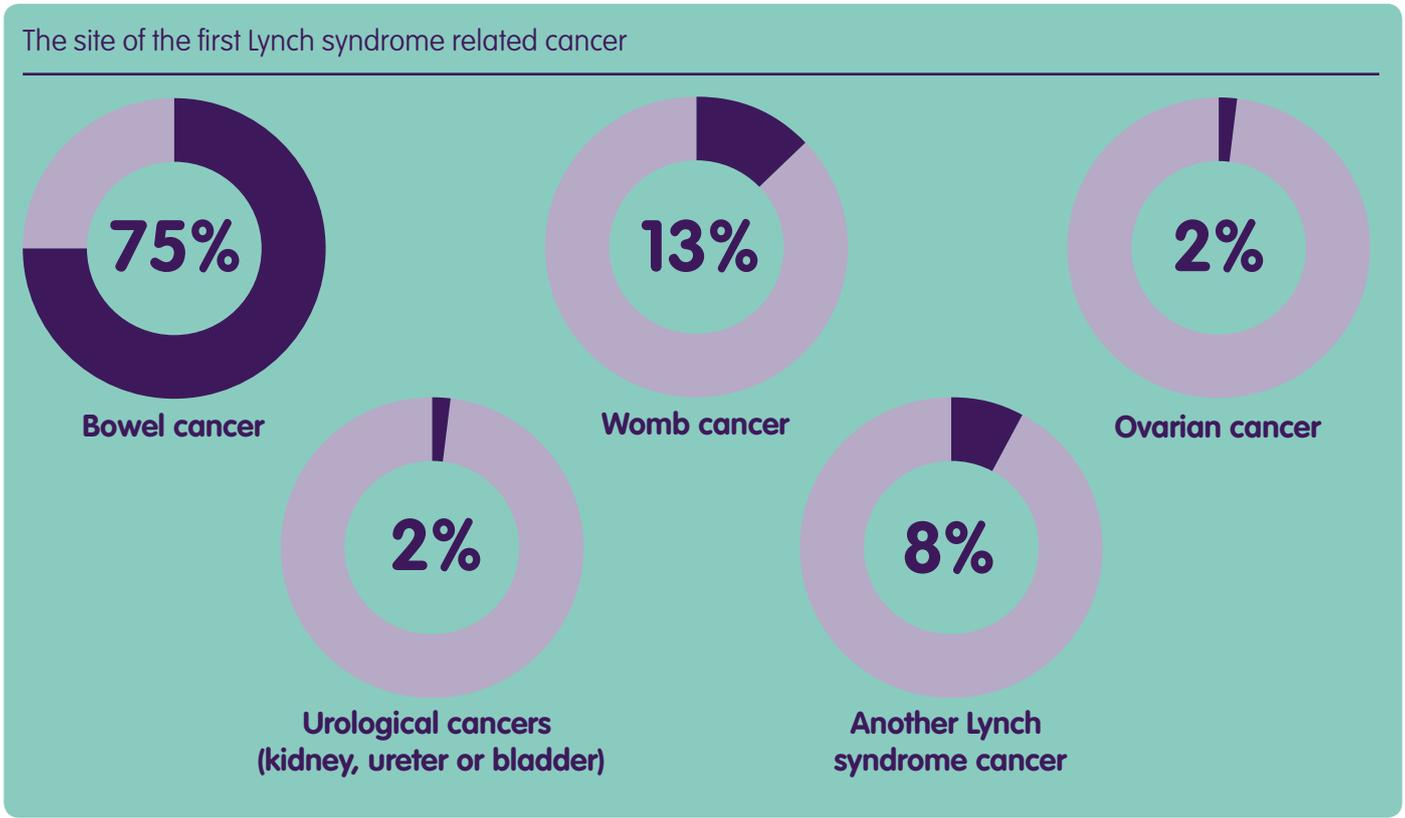
- There should be at least three relatives with a Lynch associated cancer (colorectal, womb, small bowel, ureter or renal pelvis)
- One should be a first-degree relative
- At least two successive generations should be affected
- At least one should be diagnosed before age 50
- Familial adenomatous polyposis should be excluded
- Tumours should be verified by pathological examination

However the Amsterdam Criteria has often been criticised for being too restrictive and is not commonly used by clinicians.

Lynch syndrome cancers

People who have Lynch syndrome are much more likely to develop bowel cancer at a younger age because of having a faulty gene. The average age of being diagnosed with bowel cancer in people with Lynch syndrome is 45.⁷ Just over 60% of our survey respondents said they had been diagnosed with a Lynch syndrome related cancer and 75% of these listed bowel cancer as their first Lynch syndrome related cancer. The majority of these (81%) were first diagnosed with bowel cancer under the age of 50. A third of these developed a subsequent cancer after their first diagnosis.

While we know that bowel cancer is the most common cancer caused by Lynch syndrome, with up to 1,100⁸ cases each year, people with Lynch syndrome can develop a number of cancers. Our survey demonstrates the wide range of cancers that people with Lynch syndrome can develop.



Caroline Backhouse, 51, from Hampshire



I was diagnosed with bowel cancer when I was 49 years old and I was referred to a geneticist after I had chemotherapy, where I was diagnosed with Lynch syndrome.

Before I could have the genetic test, I had to see a genetic counsellor to make sure I fully understood what was happening and why. I waited 7 months to see one. I don't know why it took so long. At the appointment we discussed my family history and she said I most likely had Lynch syndrome. A blood sample was taken to confirm the diagnosis but I had to chase and chase to get the results. Eventually, after 8 months my diagnosis was confirmed. In total, the whole process from referral to receiving my diagnosis took 15 months. Having bowel cancer is stressful enough and it's not helpful having to chase healthcare professionals.

In the 3 years it took to diagnose me with bowel cancer, I was never asked once about my family history by any of the doctors who I saw. My oncologist was the first to mention the possibility of a genetic link at my first chemotherapy appointment. I was not advised of MMR testing either. Had this screening test been carried out at diagnosis of bowel cancer, perhaps I would've known sooner about my diagnosis of Lynch syndrome.

I feel very lucky to know I have the condition as it explains so much for my family. My mum died of ovarian cancer, her mum died of bowel cancer, my mum's brother died from liver cancer, her sister died from ovarian cancer and her other brother died from lung cancer. Looking at my family tree and with my diagnosis of Lynch syndrome, the genetics team determined that they all had the genetic condition. My whole family has been devastated by cancer because of Lynch syndrome.

I've discussed my diagnosis with some family members and they are very keen to be tested. I also recently sent a copy of a letter sent to me, to my 12 first cousins on my mother's side. The letter invites their GP to contact my genetics team for further information and informs them that a member of their family has been diagnosed with Lynch syndrome.

I could view it that I'm a ticking time cancer bomb but I choose not to and keep going. The way I look at it is that I'm now monitored closely and more importantly my girls, age 11 & 13, now have the knowledge not to have to go through what myself and others in the family have. They're fully aware of Lynch syndrome and are being taught it's nothing to be scared of but something that can be managed.

I went back to my GP and told him about Lynch syndrome, and in his 30 year career he had never heard of it. More information needs to be provided to healthcare professionals, so it's not the patient informing them. I feel my experience could have been improved if healthcare professionals understood and knew about Lynch syndrome. I had to educate myself and them.

Part 2 – Testing for Lynch syndrome

Once Lynch syndrome is suspected, testing for the condition usually involves a three-stage process:

1

Provisional molecular screening test

If Lynch syndrome is suspected a simple screening test (also known as a molecular test) can be carried out on the patient's tumour tissue to identify if they are likely to have Lynch syndrome. This test looks for changes in the mismatch repair proteins.

2

Genetic counselling

Consent is needed before genetic testing can take place. Before consent can be given a person suspected of having Lynch syndrome is required to see a genetic counsellor who will talk through the process and offer information, advice and support.

3

Genetic testing

If changes to the genes are detected by the provisional screening test, the individual will be referred for full genetic testing to have their diagnosis confirmed. This involves a simple blood test and confirms that a change in the gene is present. Usually this takes around 6–8 weeks.

However the route to diagnosis can be both extremely difficult and time consuming. This is because it involves a number of steps, all of which can suffer delays.

“From initial consultation to eventually being offered the genetic test took 5 years. The professionals were helpful in providing information; however, there was no support or referral to deal with the emotional impact of a positive test”

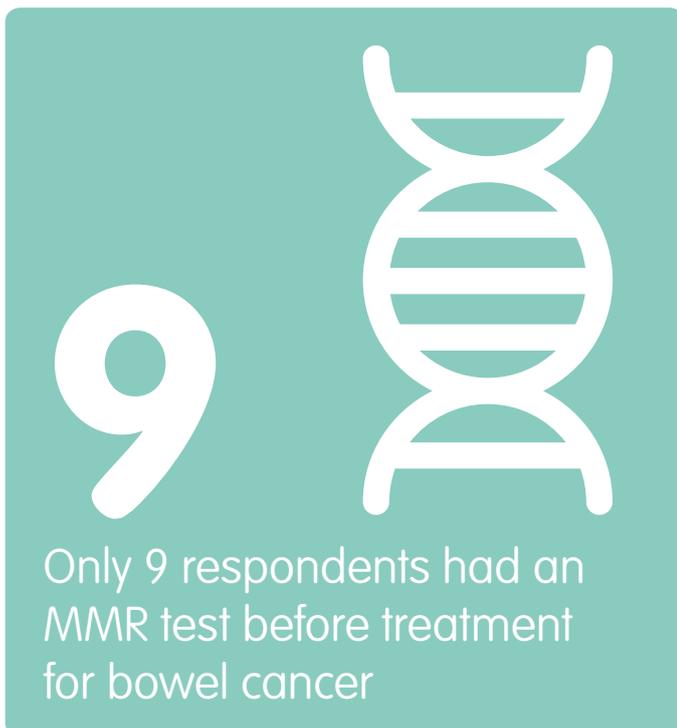
Molecular screening

Both Royal College of Pathologists⁹ (RCPATH) and British Society of Gastroenterology¹⁰ (BSG) guidelines recommend that people diagnosed with bowel cancer under the age of 50 have a provisional molecular screening test carried out automatically, as a reflex test, at diagnosis of bowel cancer. We recently carried out an FOI request¹¹ to identify the number of hospitals that currently follow the RCPATH guidelines. We found that while 71% were testing patients under the age of 50, only half were carrying out reflex testing, and less than one in ten were carrying it out at diagnosis, before treatment takes place. Our patient experience survey found that only one third of patients were aware of having this screening test and only nine respondents reported having this test carried out before treatment for bowel cancer. It is important that this test takes place at diagnosis, as having Lynch syndrome can affect treatment options for bowel cancer. We know from research¹² that patients are put at risk of not receiving the best treatment on offer to them if molecular testing for Lynch syndrome is delayed.

The National Institute for Health and Care Excellence (NICE)¹³ has published draft guidance which recommends expanding testing to all those with a diagnosis of bowel cancer. This is also known as

universal testing. The economic and health benefit for testing everyone is strong. Knowing if a person has Lynch syndrome means they can be placed in a surveillance programme to receive regular colonoscopy. This can help detect bowel cancer at an earlier stage when it is less costly to treat. Previous studies¹⁴ have also concluded that the total net health benefit for molecular testing for Lynch syndrome in all bowel cancer patients would be a cost effective use of NHS resources. Furthermore, NICE has calculated that if universal testing is adopted over 300 bowel cancers are expected to be prevented each year in the UK.¹⁵ The full guidance is set to be published in early 2017. Should these recommendations be adopted, they will have implications for the pathology laboratories involved in the testing and for clinical genetics services who advise patients and families.

It is crucial the guidelines to identify people with Lynch syndrome are adhered to as systematic testing can detect people at greater risk of bowel cancer and recurrence, inform treatment options and help identify those with family members who may also have the condition and be at risk of bowel cancer. By testing everyone we can ensure that no one slips through the diagnostic net.



Role of genetic counselling

It is mandatory to see a genetic counsellor prior to undergoing genetic testing. This is because genetic counselling plays a crucial role in providing people with the necessary support, information and advice on Lynch syndrome, the implications of genetic testing, how to cope with a diagnosis and communicating the condition and the implications of this to family members. It is encouraging that 80% of our respondents reported seeing a genetic counsellor before they were genetically tested. Many reported a positive experience of having access to genetic counselling, demonstrating the value and importance of having timely access to this service.

Currently genetic services aim to see individuals within 18 weeks¹⁶ of receiving the result of the initial MMR screening test but our survey found that one in ten respondents waited as long as six months to be seen. This may be because many genetic centres are already working to full capacity. But as genetic testing cannot be carried out until a person has been seen by a genetics counsellor, the 18-week pathway only adds to the delay to receiving a confirmed diagnosis of Lynch syndrome. This means, in some cases, people are waiting more than a year to be diagnosed. Those who test positively for an MMR defect should be seen urgently by genetic services in order to expedite the pathway to diagnosis. We also found that following a diagnosis of Lynch syndrome nearly one third of respondents were not offered a follow-up appointment with the genetics service to discuss their diagnosis, but over half of these would have wanted a follow-up appointment.

Given the valuable role that clinical genetics services and counselling plays in helping come to terms with, and understand a diagnosis of Lynch syndrome, it is crucial that have timely access to these services and that they are given the option of a follow-up appointment once their diagnosis has been confirmed.

Genetic testing

It is important that once a patient receives a positive molecular screening test they are referred for genetic testing quickly. We know that in some cases, due to the size of the gene, it can be difficult to find the mutation but patients are also kept waiting because of delays in processing of testing, which are often linked to capacity issues. This means that patients can be kept waiting unacceptable lengths of time to have their diagnosis shared with them. We found that while 70% of our survey respondents had their diagnosis confirmed and shared with them within three months of having the genetic test, 30% waited four months to over a year.

“The genetic counsellor provided brilliant information, support and advice. I felt at ease throughout the whole process and the information provided did help me come to a decision about whether to follow through with the test”

“Genetic counselling helped me to face my diagnosis and helped give me hope when I felt hopeless”

33%

33% were not offered a follow up appointment with genetic services

“Good service but it takes a long time to get an appointment. There are also regional differences in response rates which is frustrating for family members”

“Questions were answered at the time but no further follow-up was allowed or a contact for further advice. You’re just left to find out further information for yourself”

“A responsible dedicated genetic counsellor is key. Sounds reasonable but it is extremely hard to find”

Sharing a diagnosis of Lynch syndrome with family

While it can be difficult to come to terms with a diagnosis of Lynch syndrome, it can be even more difficult sharing this diagnosis with family members who may also have the condition. Although over 90% of our respondents shared their diagnosis with their family, many commented on the difficulty in doing this. In particular, feelings of guilt for potentially passing on the faulty gene to their children were mentioned.

It is important that the right information and support is readily available as soon as a diagnosis has been made. Our survey respondents said the following could have helped them to better communicate their diagnosis of Lynch syndrome to their family:

- The use of accessible and lay language in information booklets
- Basic age related information to share with their children
- An explanation on the implications of the diagnosis and what next steps should be taken
- Written information provided in a timely way about their diagnosis

Recommendations:

- All hospitals should ensure that molecular testing for Lynch syndrome takes place automatically, at **diagnosis**, as a **reflex** test on **all** patients diagnosed with bowel cancer and that the results of this test is communicated to patients.
- All hospitals should carry out **regular audits** to determine whether molecular testing of tumours for Lynch syndrome is taking place at diagnosis of bowel cancer and should ensure that clinical and laboratory teams are fully engaged with the task of delivering guidance on molecular testing for Lynch syndrome.
- A follow-up appointment with a genetic counsellor should be **standard practice after a diagnosis of Lynch diagnosis has been confirmed**.
- The **development of a streamlined and expedited route** to diagnosis to help prevent delays in the pathway and to ensure the individual receives their diagnosis as quickly as possible.
- **Better information and support** should be made available to people with Lynch syndrome to enable them to feel confident and prepared when explaining and sharing their diagnosis with family members.

The case for a national registry for people with Lynch syndrome

Right now our understanding of the number and experiences of people with Lynch syndrome in the UK is limited – only around 6,000 people with Lynch syndrome are currently known. This is estimated to be just 5% of all people with the condition. However this will begin to change with the introduction of universal testing but we need to create a national registry to enable us to conduct research and deliver the care that people with Lynch syndrome need.

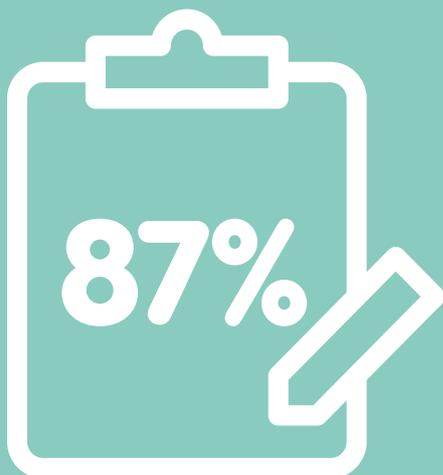
By collecting anonymised data in a registry at point of diagnosis of cancer or at point of identification, we can increase our knowledge and understanding of Lynch syndrome. This includes knowing how many people are affected and whether there are any regional differences in treatment, care and outcomes.

A national registry could also help to improve consistent management of Lynch syndrome by either feeding the data into a national programme, such as the NHS Bowel Cancer Screening Programme, or to

regional genetic services to ensure people are given the regular colonoscopy they require. Any system established must have effective recall capabilities so that people with Lynch syndrome are sent their surveillance appointments promptly. For the registry to be successful, a systematic process must be in place for adding people newly diagnosed with Lynch syndrome. Their consent to be involved would need to be mandatory. Consent should be collected at first point of diagnosis with their privacy guaranteed.

Many clinicians have come forward in support of a national registry. The Mallorca Group recommended that a regional, or national registry is needed to guarantee the continued surveillance of people with Lynch syndrome. Our survey found that 87% of our respondents would consent to be part of a registry if adopted in the UK. Reasons for this support include:

- furthering research;
- raising the profile of the condition;
- coordinating consistent care services
- wanting to help others.



87% would consent to being part of a genetics registry

“Only my sister and husband know, not my dad who it was passed on from as we didn’t want to upset him and make him feel guilty for passing it on”

“I contacted relatives on both sides since we did not know which parent passed down the gene. I still have not asked my parents to do the test as they already feel terrible”

“By creating a registry we can help improve the way screening is managed for people with Lynch syndrome. The care received currently is far too varied and we need to correct this so we all get diagnosed and receive the best screening and care.”

“In the hope that treatment and information will be the same for everyone. The data collected will help future generations with better diagnosis and maybe earlier diagnosis”

“I think it’s very important to have a register. We need national guidelines for screening etc, there shouldn’t be differences across the country”

“It’s vital that genetic teams can identify families potentially affected and for the NHS to have a clear understanding of how many people Lynch syndrome affects”

Recommendation:

A **national registry** of people identified as having Lynch syndrome must be developed. This could help increase our knowledge and understanding of Lynch syndrome, including clarifying how many people are affected by the genetic condition and the regional differences in treatment and care which need to be address to improve outcomes.

Part 3 – Managing Lynch syndrome

While there is no known cure for Lynch syndrome, there are steps that can be taken to prevent bowel cancer from developing or to detect it an early stage, when it is more treatable and the chance of survival is high.

These steps to reduce the risk of bowel cancer include:

- Regular colonoscopy
- Aspirin
- Preventative surgery

For those who do not have preventative surgery, which involves removal of the colon, it is important this risk of cancer is regularly monitored and managed consistently over the course of a person with Lynch syndrome's lifetime. Because the risk of bowel cancer in people who have Lynch syndrome is so high, it is crucial that their care is well coordinated, delivered on time and is of high quality.

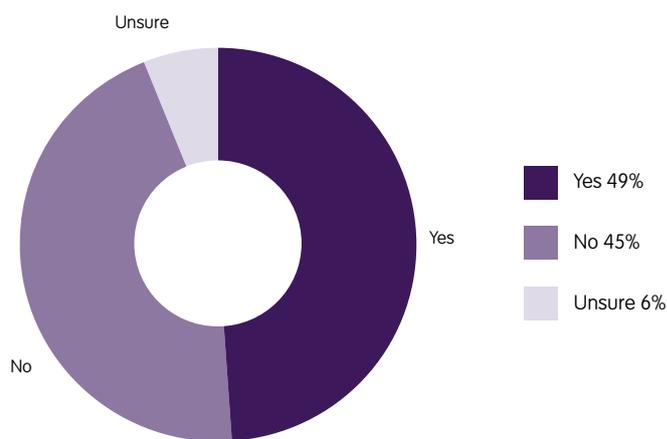
Access to regular colonoscopy

Guidelines from the BSG¹⁷ recommend that people who have Lynch syndrome are placed in a surveillance programme to receive regular colonoscopy every 18 months to two years, depending on their risk. Regular colonoscopy has been shown to reduce the chance of dying from bowel cancer by 72%¹⁸ by detecting bowel cancer early. Encouragingly 97% of our survey respondents understood why they should be having regular colonoscopy, the benefits and risk associated with this and how often they should be having them. However, 49% told us that they had experienced delays to their planned appointment date and **78% of these reported waiting more than six weeks** beyond their planned procedure date.



78% waited more than six weeks beyond their planned procedure date

Has your planned appointment for your regular colonoscopy ever been delayed i.e. beyond the planned appointment date?



There are several factors of concern that relate to why people with Lynch syndrome experience delays to their planned procedure. These are:

1. Demand on endoscopy services

Increasing demands placed upon endoscopy units have meant that many units are struggling to cope with referrals from a range of sources, including from those at high risk who require regular colonoscopic surveillance. Reports¹⁹ have estimated that nearly a million more endoscopy tests are needed year on year over the next five years. However there is a serious lack of capacity to meet this demand which is causing a detrimental impact on waiting times for these tests. In particular, the emphasis on meeting waiting time targets for those referred through the screening programme or as an urgent referral by a GP means these patients are prioritised over those at high risk due to genetic conditions, such as Lynch syndrome. As such, they are often made to wait unacceptable lengths of time before being seen. Unless those at high risk because of genetic conditions such as Lynch syndrome are placed on par with screening and symptomatic patients they will continue to be left behind.

2. An inconsistent service

The management of colonoscopic surveillance is currently carried out at a local level. However, at present there is an inconsistent approach and wide variations in the local management of bowel cancer patients with a family history of the disease. A national survey of hereditary bowel cancer services²⁰ in the UK found that:

- Many clinicians were not aware of BSG guidelines on the surveillance of people at higher risk.
- More than 1 in 5 clinicians did not think there was an adequate surveillance service for higher risk patients.
- There is no apparent formal patient pathway in 52% of centres and only 33% of centres maintain a registry of patients.
- 64% of clinicians believed that someone else should be carrying out surveillance work.

This research presents a deeply worrying picture of the management of people with Lynch syndrome at a local level. It demonstrates a system that is riddled with inconsistencies and as a result fails to provide consistent and high quality care to those at high risk of developing bowel cancer across the UK. This must be addressed urgently. An inconsistent approach to managing people at higher risk of bowel cancer will undermine efforts to save lives from this treatable disease.

“I have to do a lot of chasing and pushing for the surveillance I need. Everything is a fight, due to cost I feel I have to justify why I should have it done. One trust has a different threshold to another. Hence one hospital will do a particular screening and another I have to put up an argument and I am still not receiving the screening I require. Very frustrating”

Developing a high quality surveillance programme

The localised approach to the management of people at high risk of bowel cancer from genetic conditions, like Lynch syndrome, has contributed to the creation of an inequitable service and a postcode lottery of care. This means those at high risk who require coordinated, timely and high quality care in order to reduce and manage their risk of bowel cancer fail to receive it. To address these issues a national approach to managing the surveillance of these patients is required to significantly reduce this variation. However the lack of responsibility and leadership at a local level is also apparent at a national level. In England neither Public Health England nor NHS England are taking responsibility for improving the experience and outcome of people with Lynch syndrome.

One option for delivering a high quality national surveillance programme can be found in the service delivered for breast cancer patients. The Breast Cancer Screening Programme,²¹ which routinely tests healthy women for risk of cancer, has set a precedent for a national screening programme managing the surveillance of those with a known genetic mutation, such as BRCA 1 or 2 that increases the risk of cancer. Women who have been assessed by genetics services as being at high risk of breast cancer due to a genetic mutation are referred into the screening programme, where their surveillance is managed. Like people at high risk of bowel cancer these patients also require regular surveillance to monitor their risk. Through the Breast Cancer Screening Programme these individuals are systematically sent their appointments for their planned procedure. This reduces variation, ensures a streamlined approach to managing their surveillance and that after they are seen on time, as the programme is subject to strict waiting time targets and quality assurance protocols.

The National Bowel Cancer Screening Programmes in the UK, aimed at the general population, also provide an efficient high quality service, with their own strict waiting time targets. However, so far in England there has been reluctance from Public Health England to manage these patients as is carried out by the Breast Cancer Screening Programme. Yet it would be a relatively straight forward service for them to provide. This disparity between the screening programmes and the services received by those without symptoms and those at high risk of bowel cancer, who are up to 80% likely to be diagnosed with the disease, is unacceptable. People with Lynch syndrome should receive the same high quality and timely care provided to those who take part in population screening who overall have a much lower risk of bowel cancer.

If the National Bowel Cancer Screening Programmes in the UK were to manage the surveillance of people with Lynch syndrome, similar to those at high risk of breast cancer, this could help to reduce the vast variation in access to regular colonoscopy for this group of patients. Clearly this is not the only option available and it may be possible to deliver a system using regional services such as that delivered in the North West of England. The reality is that until there is clear local and national leadership and a firm commitment to improve the services for people at high risk of developing bowel cancer, people with Lynch syndrome will continue to fall through the gap and lives will be needlessly lost.

Aspirin: reducing the risk of bowel cancer

Clinical trials have shown that taking regular aspirin from the age of 50²² can significantly reduce the risk of bowel and other cancers in Lynch syndrome carriers by over 50%. We found that 62% of our survey respondents had been recommended aspirin as part of their treatment. However the dose at which aspirin should be taken is still unknown. Further research and guidance on the use of aspirin in this group of patients is needed.

CaPP3 trial²³

The Cancer Prevention Programme (CaPP3) is a three year trial focussed on finding the right daily dose of aspirin to reduce the risk of cancer in people with Lynch syndrome. Participants will be asked to take three tablets daily for two years – one will be a dummy tablet and at least one will contain aspirin at; 100mg, 300mg or 600mg levels. The tablet levels will be randomised so that once the trial is completed it will be known if a lower dose is sufficient to help decrease the rate of cancer.

www.capp3.org

Recommendations:

- A quality assured **national surveillance programme** for people with Lynch syndrome should be established to reduce the vast variation in access, quality of and timeliness of regular colonoscopy. A feasibility study should be undertaken to develop and model the most efficient way of delivering surveillance to people with Lynch syndrome.
- A **single lead within each colorectal multidisciplinary team (MDT)** should have oversight of service coordination and delivery within their institution, with support from regional genetic services.

Annika Perdomo, 53, from Warwickshire



I was diagnosed with bowel cancer four years ago when I had just turned 49. Being told I had bowel cancer was devastating, not only for me but all of my family too. It didn't make sense. I had surgery a few weeks later to remove a large section of my colon, which contained the 11cm long tumour. It was only after treatment that my family history of cancer was discussed and I was eventually referred to a genetics counsellor by my colorectal nurse specialist. I have a lot to thank her for. She was amazing and helped me through this whole process a lot. At this point I was tested and eventually diagnosed as having Lynch syndrome. Prior to this the possibility that my cancer might be hereditary was never discussed, no one asked me about my family history and I was never aware of having the provisional molecular screening test, at diagnosis of bowel cancer. Overall it took about a year from first being referred for testing to having my diagnosis of Lynch syndrome confirmed. Getting the results of the genetic test took 7 months alone.

While I was given some information which explained my diagnosis and outlined what care I should receive, I wasn't given a personalised written care plan and I had to supplement the information I received with my own research. Although I've been provided with a dedicated healthcare professional who manages my care, I feel I have to keep on top of it otherwise I sense I may be forgotten in the system.

Having never even heard of this before, it began to make sense why I may have had the cancer. My paternal grandmother died of bowel cancer many years ago, and my father and paternal aunt of other Lynch syndrome related cancers. Of course we were not aware before my illness of any link between these family deaths. Unfortunately three of my siblings have since tested positive with the genetic condition.

Lynch syndrome is very scary due to the fact that tumours can apparently grow more rapidly. It was for this reason that I opted for preventative surgery two years ago by having a full abdominal hysterectomy to rid the chance of me ever having an endometrial cancer. My sister also has Lynch syndrome and is about to go through the same preventative surgery at the end of this year.

The saddest thought out of all this for me is the fact that there's a 50:50 chance of my daughter having Lynch syndrome too. She will be tested when she reaches the age of maturity, as will my nephew and nieces. But the major difference for them is that they will be screened and monitored from an early age. Unlike me, any symptoms they may be experiencing however young they are at the time, their GP will at least sit up and listen.

I'm really grateful for how quickly they dealt with the rest of the family in getting them screened and tested once I had the diagnosis. If they hadn't worked so speedily, my brother's undetected cancer may have spread. But I feel my experience of being diagnosed with Lynch syndrome could have been improved if there had been a speedier referral to diagnosis process, especially as I was under 50.

Part 4 – A personalised approach to care

Increasingly the NHS is moving to a more personalised approach to treatment, which is putting patients at the heart of their care. Research shows that a personalised and patient-centred approach to the care of people with long-term conditions can improve patient experience of the healthcare system. As people with Lynch syndrome require regular management and monitoring of their cancer risk over the course of their lifetime and will come into contact with a range of healthcare professionals this type of personalised approach would be of benefit to them.

Care plan

Throughout this survey many people have informed us that they have not had access to enough personalised information on their condition and also have to explain to healthcare professionals who they come into contact with what their diagnosis means, and what care they need. A key tool to ensuring that care is personalised and streamlined is through the use of a written care plan²⁴. This should be a standard part of a person's care and should address the full range of their needs, contain relevant information on their diagnosis of Lynch syndrome, cancers they have developed and treatment received. It should also be a record of the outcome of their surveillance appointments, so their risk can be monitored. The care plan should be held by the person with Lynch syndrome, so they can share it with any healthcare professional they may come into contact with, such as their GP. However, we found that over 70% of our respondents had not been offered a written care plan, but nearly 95% would have liked one.

“Some of the professionals I have seen have been wonderful. However I am frequently not listened to and I feel I'm a hindrance or just a number to them. I would appreciate a personalised care plan – there has been nothing personalised about my care so far. It has been very upsetting and disappointing”

70%



70% have not been offered a personalised care plan

A care plan could include:

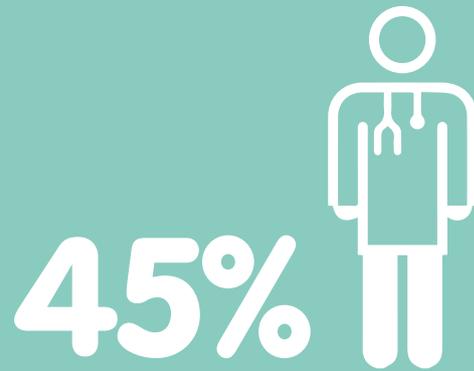
- Information about their specific Lynch syndrome gene mutation.
- Key colonoscopy dates and their frequency.
- Any treatment they may have had such as preventative surgery, radiotherapy or chemotherapy.
- Any involvement in clinical trials/alternative therapies they may be involved in, such as aspirin.

A dedicated healthcare professional

A range of healthcare professionals can be involved in the care of a person with Lynch syndrome, including a gastroenterologist, oncologist, surgeon, and geneticist. For people with Lynch syndrome the involvement of different healthcare professionals can lead to care feeling uncoordinated and not knowing who to go to for relevant information and support. A single point of contact could help them to navigate often complex pathways and could help provide continuity of care, improving the experience of what is an incredibly stressful and anxious time for Lynch syndrome families.

However, we found that 45% of our survey respondents do not have a dedicated healthcare professional to manage their surveillance and a further 15% were uncertain. A range of healthcare professionals were listed for those who had one and these ranged from an oncologist, to a nurse to a gastroenterologist. Not all people with Lynch syndrome will have had a diagnosis of cancer so whoever the healthcare professional, they must have the relevant skills required to ensure continuity of care.

“There needs to be much more of an emphasis on emotional support for both Lynch syndrome and cancer to help people deal with a diagnosis better and know where to turn for help and that their feelings are normal”



45% do not have a dedicated healthcare professional managing their care

Recommendations:

- Everyone with Lynch syndrome should have a **dedicated named healthcare professional** to manage their surveillance and offer support throughout their care.
- Everyone with Lynch syndrome should be provided with a written **personalised care plan** at diagnosis to support further involvement in making suitable treatment decisions for their care.

Part 5 – Access to information and services

Access to high quality information is essential to enabling people with Lynch syndrome to fully understand and come to terms with what their diagnosis means, what the next steps are, what treatment options are available and to make informed decisions about their care. However over a quarter (43%) of our survey respondents reported they would have wanted more relevant and useful information about Lynch syndrome.

In particular, our survey respondents would have liked further information on the following:

- 1** Information on other Lynch syndrome related cancers (such as womb, stomach and ovarian) – 63%
- 2** Tailored information relevant to them and their family's history of cancer – 60%
- 3** Their surveillance programme – 51%
- 4** Impact of Lynch syndrome on treatment options for cancer – 50%
- 5** General information on Lynch syndrome – 42%

Being diagnosed with Lynch syndrome can be incredibly overwhelming and affects not only the individual but family members too. Our findings show that there are huge information gaps on important aspects of treatment and care that need to be urgently addressed. One aspect that is particularly lacking is personalised and tailored information. It is crucial that people with Lynch syndrome are provided with information that is relevant to them, at the time they need it most. Conditions like Lynch syndrome can affect people in different ways and appropriate information could help to alleviate some of the anxiety that people experience.

Our findings show that although our survey respondents receive their information from a variety of sources, many of them rely on their own research by carrying out web searches, visiting online discussion

boards and charity websites. Although websites and online chat rooms can be an excellent source of support for patients the quality and accuracy of information can vary. There needs to be a greater provision of Information Standard²⁵ accredited information from the NHS and signposting from clinicians to the highest quality information.

Unsurprisingly most people receive their information through their genetic service but information could, and should, come from a range of healthcare professionals, including clinical nurse specialists, gastroenterologists and oncologists. As people with Lynch syndrome will come into contact with a number of healthcare professionals it is important that they all have a basic understanding of the condition and are able to sign post to other sources of information.

The primary sources of information for our respondents include:

- 1 Genetics Counsellor – 71%
- 2 Own web search – 60%
- 3 Online discussion boards and chat rooms – 35%
- 4 Charity websites e.g. Lynch syndrome UK – 33%
- 5 The Hospital – 13%

Many of our survey respondents reported that they were in fact the ones to inform their clinician about Lynch syndrome. This could be related to a lack of understanding and knowledge among many clinicians. 73% of our survey respondents told us that a knowledgeable GP could have improved their experience, but Lynch syndrome is a rare condition so it is possible that GPs in particular may not have heard of the condition. Educating all healthcare professionals to at least a basic understanding of what Lynch syndrome is, and how to manage and support people with the condition, could ultimately help to identify more people and prevent more deaths from cancer.

“I get so cross having to explain Lynch syndrome to the healthcare professionals in this area”

“The fact that so many doctors are not up to date with this and even some discarding it as scaremongering”

“GPs must be more upskilled in Lynch syndrome so that the patient is not the one having to endlessly bring other health professionals up to date”



43%

43% want more information on Lynch syndrome

Recommendations:

- **High quality and tailored information** on Lynch syndrome should be offered to people at diagnosis and made available on a number of platforms.
- All healthcare professionals involved in the diagnosis, management and care of people with Lynch syndrome should receive **an appropriate level of education and training** in order to provide adequate support to their patients.

Conclusion: Improving the experience of being diagnosed and managed for Lynch syndrome

This report shows us that far too many people with Lynch syndrome are struggling to get the right support they need from the health service. This is being seen across the entire pathway; delays in genetic testing are leaving people waiting for a diagnosis, delayed colonoscopy appointments are preventing them from taking necessary precautionary action and without access to sufficient support and information many

feel alone and overwhelmed. We asked survey respondents what could have been done to improve their experience of being diagnosed and managed for Lynch syndrome and we found that most people want better informed clinicians, many want to be more involved in their care by having a personalised treatment plan and many also felt a single point of contact could have improved their experience.

Respondents' top 5 improvements

- 1 Better informed family doctor/GP – 73%
- 2 A personalised care plan – 55%
- 3 A dedicated named healthcare professional – 49%
- 4 More Lynch syndrome specific information from cancer support organisations – 45%
- 5 A more reliable surveillance programme – 40%

This shows that substantial changes need to be made to improve the people with Lynch syndrome's experience of being diagnosed and managed for the condition.

We are calling on the NHS, commissioners and health bodies across the UK to implement the following recommendations:

Recommendations

Identifying Lynch syndrome

- The development of **comprehensive UK guidelines** that set out best practice for the clinical management of people with Lynch syndrome.
- A **national registry** of people identified as having Lynch syndrome must be developed. This must be a systematic process with patient consent obtained by a clinician at the point of diagnosis. This would increase our knowledge and understanding of Lynch syndrome, including knowing how many people are affected by the genetic condition and whether there are any regional differences in treatment, care and outcomes.
- All hospitals should ensure that molecular testing for Lynch syndrome takes place automatically, at **diagnosis**, as a **reflex** test on **all** patients diagnosed with bowel cancer and that the results of this test is communicated to patients.
- All hospitals should carry out **regular audits** to determine whether molecular testing of tumours for Lynch syndrome is taking place at diagnosis of bowel cancer and should ensure that clinical and laboratory teams are fully engaged with the task of delivering guidance on molecular testing for Lynch syndrome.
- A **follow-up appointment with a genetic counsellor** should be standard practice after a diagnosis of Lynch diagnosis has been confirmed.
- The **development of a streamlined and expedited route to diagnosis** to help prevent delays in the pathway and ensure a patient receives their diagnosis as quickly as possible.
- **Better information and support** should be made available to people with Lynch syndrome to enable them to feel confident and prepared when explaining and sharing their diagnosis with family members.

Treatment and care

- A quality assured **national surveillance programme** for people with Lynch syndrome should be established to reduce the vast variation in access, quality of and timeliness of regular colonoscopy. A feasibility study should be undertaken to develop and model the most efficient way of delivering surveillance to people with Lynch syndrome.
- **A single lead within each colorectal multidisciplinary team (MDT)** should have oversight of service coordination and delivery within their institution, with support from regional genetic services.
- Everyone with Lynch syndrome should have a **dedicated named healthcare professional** to managing their surveillance and offer support throughout their care.
- Everyone with Lynch syndrome should be provided with a written **personalised care plan** at diagnosis to support further involvement in making suitable treatment decisions for their care.

Access to information

- **High quality and tailored** information on Lynch syndrome should be offered to people at diagnosis and made available on a number of platforms.
- All healthcare professionals involved in the diagnosis, management and care of people with Lynch syndrome should receive **an appropriate level of education and training** in order to provide adequate support to their patients.

Appendix A: References

- 1 'Molecular testing strategies for Lynch syndrome in people with colorectal cancer' National Institute for Health and Care Excellence, Diagnostics consultation document, 2016 <https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- 2 UK Colorectal cancer patients are inadequately assessed for Lynch syndrome, Adelson et al, Frontline Gastroenterology (2013) <http://fg.bmj.com/content/early/2013/08/09/flagstro-2013-100345>
- 3 'Molecular testing strategies for Lynch syndrome in people with colorectal cancer' National Institute for Health and Care Excellence, Diagnostics consultation document, 2016 <https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- 4 A national survey of hereditary colorectal cancer services in the UK – Monahan K – <http://fg.bmj.com/content/early/2013/09/16/flagstro-2013-100362>
- 5 'Revised guidelines for the clinical management for Lynch syndrome (HNPCC): recommendations by a group of European experts' The Mallorca Group, GUT 2013 62 (812-823) <https://www.ncbi.nlm.nih.gov/pubmed/23408351>
- 6 Amsterdam criteria set by the International Collaborative Group on Hereditary Non-Polyposis Colorectal Cancer [http://www.nchpeg.org/documents/crc/11-0456%20Fact%20sheets%20\(Amsterdam%2011%20criterial.pdf](http://www.nchpeg.org/documents/crc/11-0456%20Fact%20sheets%20(Amsterdam%2011%20criterial.pdf)
- 7 A systemic review and economic evaluation of diagnostic strategies for Lynch syndrome, Snowsill et al, 2014, NHS National Institute for Health Research http://www.journalslibrary.nihr.ac.uk/___data/assets/pdf_file/0005/125978/FullReport-hta18580.pdf
- 8 'Molecular testing strategies for Lynch syndrome in people with colorectal cancer' National Institute for Health and Care Excellence, Diagnostics consultation document, 2016 <https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- 9 Dataset for colorectal cancer histopathology reports, Royal College of Pathologists (2014) <https://www.rcpath.org/resourceLibrary/dataset-for-colorectal-cancer-histopathology-reports--3rd-edition-.html>
- 10 Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups British Society of Gastroenterology (update from 2002) http://www.bsg.org.uk/images/stories/docs/clinical/guidelines/endoscopy/ccs_10.pdf
- 11 2016 Data Briefing: Reflex testing for Lynch syndrome in people diagnosed with bowel cancer under the age of 50: 2016, Bowel cancer UK <https://bowelcancerorguk.s3.amazonaws.com/Final2016DataBriefingLynchsyndrome.pdf>
- 12 Universal tumour screening for Lynch syndrome: Assessment of the perspectives of patients with colorectal cancer regarding benefits and barriers, Hunter et al, Cancer, 2015 121(18):3281-9 <https://www.ncbi.nlm.nih.gov/pubmed/26036338>
- 13 'Molecular testing strategies for Lynch syndrome in people with colorectal cancer' National Institute for Health and Care Excellence, Diagnostics consultation document, 2016 <https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- 14 A systemic review and economic evaluation of diagnostic strategies for Lynch syndrome, Snowsill et al, 2014, NHS National Institute for Health Research http://www.journalslibrary.nihr.ac.uk/___data/assets/pdf_file/0005/125978/FullReport-hta18580.pdf
- 15 'Molecular testing strategies for Lynch syndrome in people with colorectal cancer' National Institute for Health and Care Excellence, Diagnostics consultation document, 2016 <https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- 16 NHS – Your rights in the NHS: guides to waiting times <http://www.nhs.uk/choiceintheNHS/Rightsandpledges/Waitingtimes/Pages/Guide%20to%20waiting%20times.aspx>
- 17 Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups British Society of Gastroenterology (update from 2002) http://www.bsg.org.uk/images/stories/docs/clinical/guidelines/endoscopy/ccs_10.pdf
- 18 Prevention of colorectal cancer by colonoscopic surveillance in individuals with a family history of colorectal cancer: 16 year, prospective, follow up study, Isis Dove Edwin et al (2005) BMJ
- 19 Scoping the future: An evaluation of endoscopy capacity across the NHS in England, Cancer Research UK – https://www.cancerresearchuk.org/sites/default/files/scoping_the_future_-_final.pdf
- 20 A national survey of hereditary colorectal cancer services in the UK – Monahan K – <http://fg.bmj.com/content/early/2013/09/16/flagstro-2013-100362>
- 21 National Breast Cancer Screening Programme <https://www.gov.uk/government/publications/breast-screening-screening-of-higher-risk-women>
- 22 The Family History of Bowel Cancer – familyhistorybowelcancer.wordpress.com/tag/kevin-monahan/
- 23 Cancer Prevention Programme – www.capp3.org
- 24 'Delivering better services for people with long-term conditions: Building the house of care' Coulter et al (2013) The King's Fund https://www.kingsfund.org.uk/sites/files/kf/field/field_publication_file/delivering-better-services-for-people-with-long-term-conditions.pdf
- 25 The Information Standard – NHS England – <https://www.england.nhs.uk/tis/>

Bowel Cancer UK is the UK's leading bowel cancer research charity.

We are determined to save lives and improve the quality of life for all those affected by bowel cancer.

We support and enable research, educate patients, public and professionals about bowel cancer and campaign for early diagnosis and best treatment and care for all those affected.

Find out more at [bowelcanceruk.org.uk](https://www.bowelcanceruk.org.uk)



/charitybcuk



@Bowel_Cancer_UK

