NHS
South East
Genomic Medicine Service

HOW CAN WE IMPROVE THE EXPERIENCES OF PEOPLE WHO NEED TO BE SCREENED FOR LYNCH SYNDROME?

February 2025

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EXECUTIVE SUMMARY

To evaluate the achievements of the National Lynch syndrome Programme and ongoing work across the South East region, we wanted to understand the experiences of patients who have been screened for Lynch syndrome. To achieve this, we gathered feedback with people who had been screened for Lynch syndrome to understand and explore their experiences.

Analysis of their feedback was performed to understand the demographics of who took part in the survey, how they rated their overall experience of screening, and the key factors that contributed to this.

Who participated?

A total of 122 people shared their experiences. Of these, 7 people had not been screened for Lynch syndrome, so were not eligible to participate. A further 7 people received their Lynch syndrome care from outside of England. This resulted in 108 participants that we were able to include within the scope of this report. Nationally, survey data shows that most responses were from the South East, with 45 individual participants. The majority of participants were female, representing 84% of the cohort nationally. The predominant ethnicity was White British, with the most common age group being 50-59 years old, although a wide range of age groups was represented from 20 – 29 to 70 years old and above.

Overall Experience

Overall experience of screening varied across regions. National responses are explored in more detail within the survey results section, although the primary focus of this report is patient experience across the South East.

Respondents who received their care through a mainstreamed pathway within the South East, reported an overall positive experience. Participants that did not rate their overall experience as positive described that their poor experiences were often the result of long waits for clinical genetics appointments, and a lack of an accessible and knowledgeable point of contact to help them through their diagnosis.

Thematic analysis was used to investigate these experiences further by categorising qualitative responses and patient quotes into areas of success and opportunities for improvement. From each theme, a set of actionable recommendations have been developed that can be implemented by hospital services, clinical genetics departments, cancer alliances and Genomic Medicine Services nationwide.

LACK OF INTEGRATED CARE

People talked to us about the lack of integration between cancer services, clinical genetics and their GP. This meant they often had to have repeated conversations about difficult topics such as a cancer diagnosis, or diagnoses of family members.

Respondents also shared their concerns with us about facing long waiting times for a clinical genetics appointment with some getting lost in the system and having to repeatedly follow up. This caused anxiety and distress, leaving patients feeling alone and dismissed.

EXPERIENCES WITH CLINICAL GENETICS SERVICE AND HOSPITAL CARE

Many respondents referred to their clinicians by name and told us that they had developed a positive relationship with them after being helped through a difficult diagnosis. This continuity of care was important to patients, with many sharing having a dedicated person they can contact within their oncology team made a positive difference to their overall experience.

'KNOWLEDGE IS POWER'

Patients who received a diagnosis of Lynch syndrome said that they were thankful that they were able to make positive changes as a result, with a number of people describing their diagnosis as lifesaving.

With this, some respondents wished that they and their family members had been diagnosed earlier. This identified a need for increased awareness amongst clinicians about Lynch syndrome, and better tools to provide patients with appropriate and standardised information about screening.

SHARED EXPERIENCES WITHIN FAMILIES

Participants' experiences were often strongly associated with that of their family members. For instance, positive experiences were overshadowed if family members had a poor experience of cascade testing.

The potential impact upon family members significantly influenced patients' decisions around testing and onward management. This highlights that family members should be considered when guiding patients through the screening process,, as this is often one of their primary concerns.

PSYCHOLOGICAL AND EMOTIONAL SUPPORT

Receiving a genetic diagnosis can be extremely emotional, particularly if the patient is simultaneously undergoing cancer care or treatment. This can be exacerbated by long waits for clinical genetics appointments, leaving patients feeling lonely and anxious due to the lack of support available. This highlighted the need for additional psychological and emotional support though the entirety of the pathway.

PROVISION OF AN INCLUSIVE AND ACCESSIBLE EXPERIENCE

The diverse range of people that access genetic testing and oncology services should be considered when aiming to deliver compassionate and personalised care. It is crucial to respect individual preferences and accessibility needs to prevent additional barriers to accessing care or attending appointments.

This report is based on the responses received from those who received their care in the South East region, though similar themes were echoed through responses nationally. We aim to share these patient-driven recommendations, so that they can be heard, implemented, and acted upon across the South East and nationwide to improve experiences of screening for patients in the future.

BACKGROUND & CONTEXT

Lynch syndrome is a common condition that can run in families and can lead to a higher risk of developing cancer at a younger age.

The most common types of cancers associated with Lynch syndrome are colorectal (bowel) cancer and endometrial (womb) cancer. Lynch syndrome is responsible for 3% of all bowel and womb cancer diagnoses. Other cancers related with Lynch syndrome are ovarian, prostate, stomach, small bowel, upper GI, bile duct/gallbladder, pancreas, bladder, brain, ureter and sebaceous skin cancer.

We previously co-led a national project to ensure that more people are screened for Lynch syndrome across the UK, meaning more cancers will be detected earlier.

As a result, 95% of people with bowel or womb cancers in England now get screened for Lynch syndrome, compared to 50% before our work began.

The South East Genomic Medicine Service supported NHS hospitals to offer standardised and equitable screening for Lynch syndrome for colorectal and endometrial cancer patients through the National Lynch syndrome Programme. The key aim was to embed screening for Lynch syndrome within pathology, as recommended in NICE guidance.

A secondary aim was to enable oncology teams to offer diagnostic testing for Lynch syndrome in patients detected through the initial screening. This shift to offering this diagnostic genetic testing by local oncology teams, as opposed to referring to regional genetics services, is called ‘mainstreaming’. Mainstreaming is thought to be beneficial to patients as it offers quicker access to genetic testing, delivered by a familiar clinician, and in their locality.

We’re monitoring and addressing any geographical variations in testing and supporting NHS Trusts to set up their own processes for screening and supporting their patient

AIMS & OBJECTIVES

We wanted to understand how people experience Lynch syndrome screening, and explore how we could make improvements for the future.

Hear from a range of voices about their experience

Improve the pathway to improve peoples' experiences

OUR APPROACH

At the South East Genomic Medicine Service, we have fostered a strong culture of involvement, which sees people and patients being involved in every aspect of our work, from attending board meetings, being equal voices in team meetings and having an active involvement in our projects.

We regularly hear from a number of people about their experience of being screened for Lynch syndrome, but we wanted to hear from more voices, and to delve deeper into the issues that they face.

Working together with our patient representatives, we designed this project to enable us to hear from as many people as possible. Equally, our approach enabled us to have in-depth conversations with individual people to better understand their experiences, and how we could make improvements based on this.

This report details how we went about it, what we heard and what we are recommending as a result.

The Headlines

- 120 people shared their story with us
- Experience of Lynch Syndrome varies hugely across regions and hospital sites
- Lack of knowledge about Lynch and the pathways amongst professionals
- A dedicated Lynch specialist is transformational to someone's experience
- Everyone we spoke to felt that the knowledge genetic testing gave them was powerful

METHODOLOGY

Our aim was to hear from as many people as possible. This piece was focused on the South East region in particular, but we opened up the survey nationally.

To achieve this, we created an online survey to gather quantitative data, and combined this with in-depth personalised conversations with individual people.

Online Survey

An online survey was live for 11 weeks, from 1st August to 18th October 2024. All responses were anonymised.

Patients were involved in the curation of the survey to ensure that all questions were compassionate and inclusive. To capture both quantitative and qualitative insights, the survey included a range of question types such as Likert-scale questions, as well as both open and closed ended questions.

A copy of the survey questions can be found in Appendix 1.

Distribution

The survey was distributed nationally through a range of channels to ensure a broad and diverse reach including:

- **Direct patient outreach** via Lynch Champions and clinicians. The survey was also distributed via patient registries.
- **Charities** including Lynch syndrome UK, Bowel Cancer UK, Macmillan, The Bowel Movement, Peaches Trust, The Eve Appeal and Bowel Research UK shared the survey with their members and via their social media channels.
- **Newsletters & social media.** We shared the survey link in all SE GMSA newsletters, our website, LinkedIn and X. Our patient representative also shared the survey with Lynch syndrome support Facebook groups.

Individual Conversations

As part of the survey, participants were invited to take part in an individual conversation about their experience. Within these one-to-one conversations, patients spoke in depth about their experience, and what could be improved.

22 people took part in individual conversations, with each session following an agreed conversation framework. All participants were provided with a consent and pre-session information sheet explaining the purpose of the conversations. Support was also available from our Lynch syndrome Specialist Nurse should they need it.

Data Analysis

Quantitative responses from the survey were analysed by using Excel software. Findings are described in the results section of this report.

Using the data collected through qualitative questions and patient interviews, responses were thematically analysed to categorise feedback into six key themes. This analysis can be found in the 'what did we hear?' section of this report.

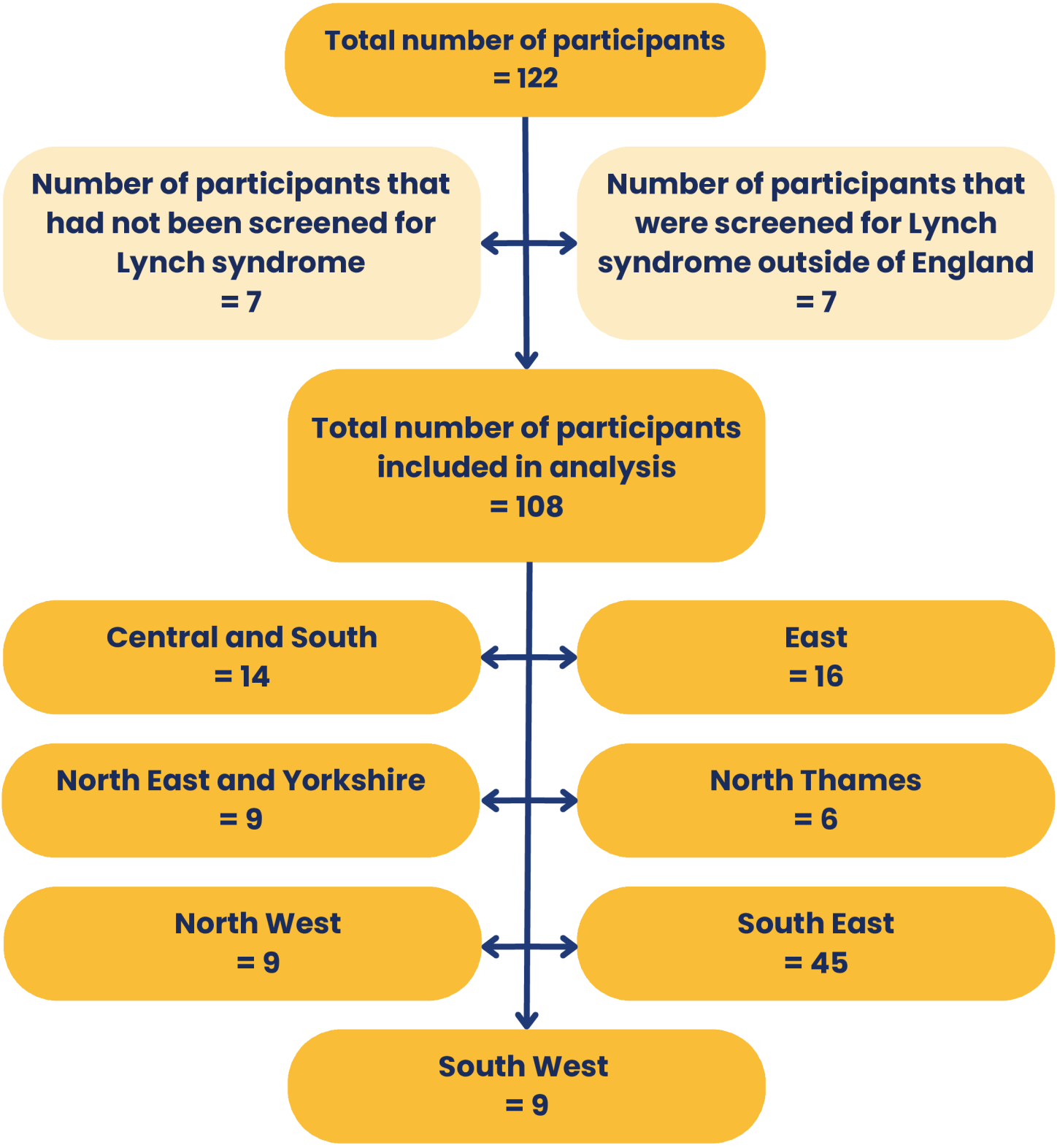
Confidentiality

All responses from both the survey and interviews have been anonymised and stored securely in accordance with Guy's and St Thomas' NHS Foundation Trust information governance policies.

SURVEY RESULTS

The survey received 122 individual responses. Response data from participants who had not been screened for Lynch syndrome, along with those who were screened outside of England were omitted from this analysis. The final dataset resulted in a total of 108 responses, 45 of which were from patients who received Lynch syndrome screening in the South East region.

Who responded to the survey?



Demographics

Of the 108 responses from England, 84% were female and 16% were male. Most participants were of a White British origin, with 92% self-identifying as English, Welsh, Scottish or Northern Irish. 2% of respondents were White and Black Caribbean, and a further 2% were Irish. The remaining 5% accounts for Chinese, Indian, Greek, White American and White European groups, each making up 1% of the cohort.

Survey participants represented a wide range of age groups. 78% of respondents were above the age of 50, with the most common age group being 50-59 years old.

	Group	No. of Participants	Percentage of Participants
Sex	Female	91	84%
	Male	17	16%
Ethnicity	Asian or Asian British - Chinese	1	1%
	Asian or Asian British - Indian	1	1%
	Greek	1	1%
	Mixed/Multiple Ethnic Groups - White and Black Caribbean	2	2%
	White - English / Welsh / Scottish / Northern Irish / British	99	92%
	White - Irish	2	2%
	White American multiple white- Irish, Italian	1	1%
	White European	1	1%
Age	20-29 years old	4	4%
	30-39 years old	7	6%
	40-49 years old	13	12%
	50-59 years old	41	38%
	60-69 years old	27	25%
	70 years old or above	16	15%
Region	Central and South	14	13%
	East	16	15%
	North East and Yorkshire	9	8%
	North Thames	6	6%
	North West	9	8%
	South East	45	42%
	South West	9	8%
Reason for Screening	Screened following a cancer diagnosis	64	59%
	Screened due to their family history	43	40%
	I'm not sure	1	1%
Diagnosis	Diagnosed with Lynch syndrome	76	70%
	Not diagnosed with Lynch syndrome	32	30%

NATIONAL KEY FINDINGS

National Results

Nationally, most patients who received care through a mainstreamed pathway or through a traditional pathway via clinical genetics had a positive overall experience of screening.

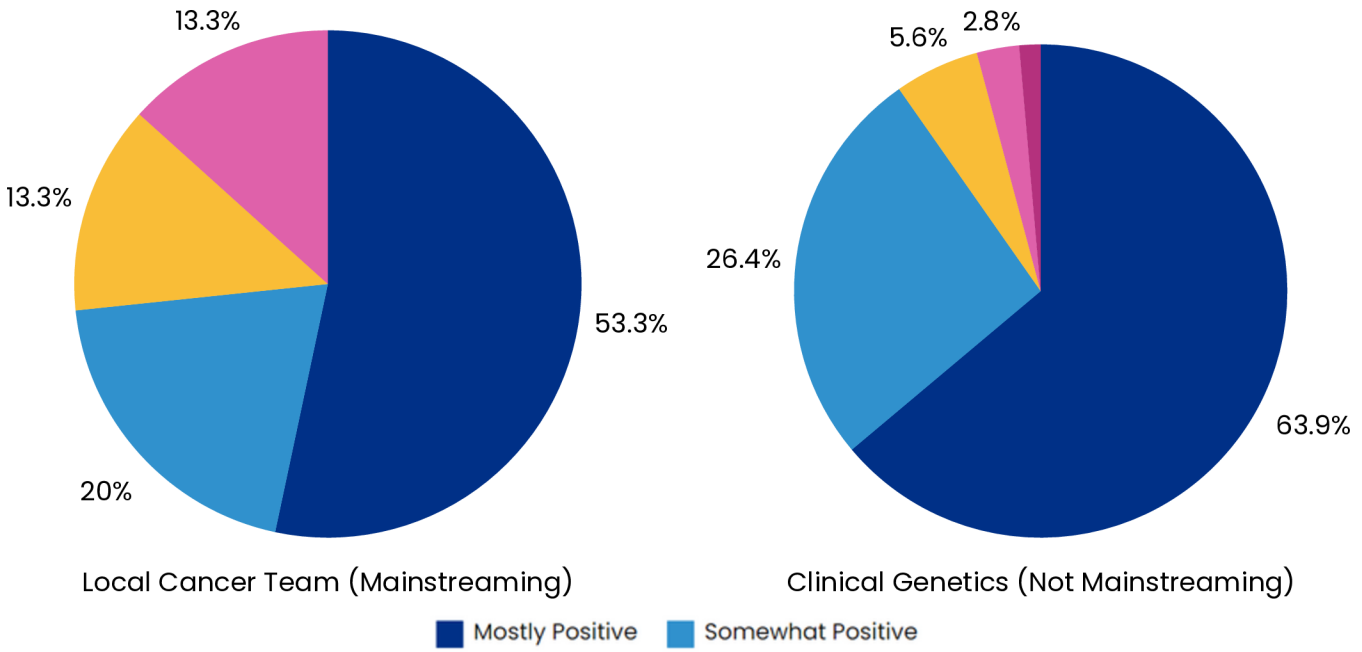
Within the cohort that did not receive care through a mainstreamed pathway, negative experiences were due to long waits for a clinical genetics appointment, and the anxiety that this caused. Poor experiences were also found to be due to a disconnect between the referrer, whether GP or local cancer teams, and clinical genetics, leaving patients feeling lost with no one to contact.

No patient that experienced a mainstreamed pathway had a mostly negative experience of Lynch syndrome testing.

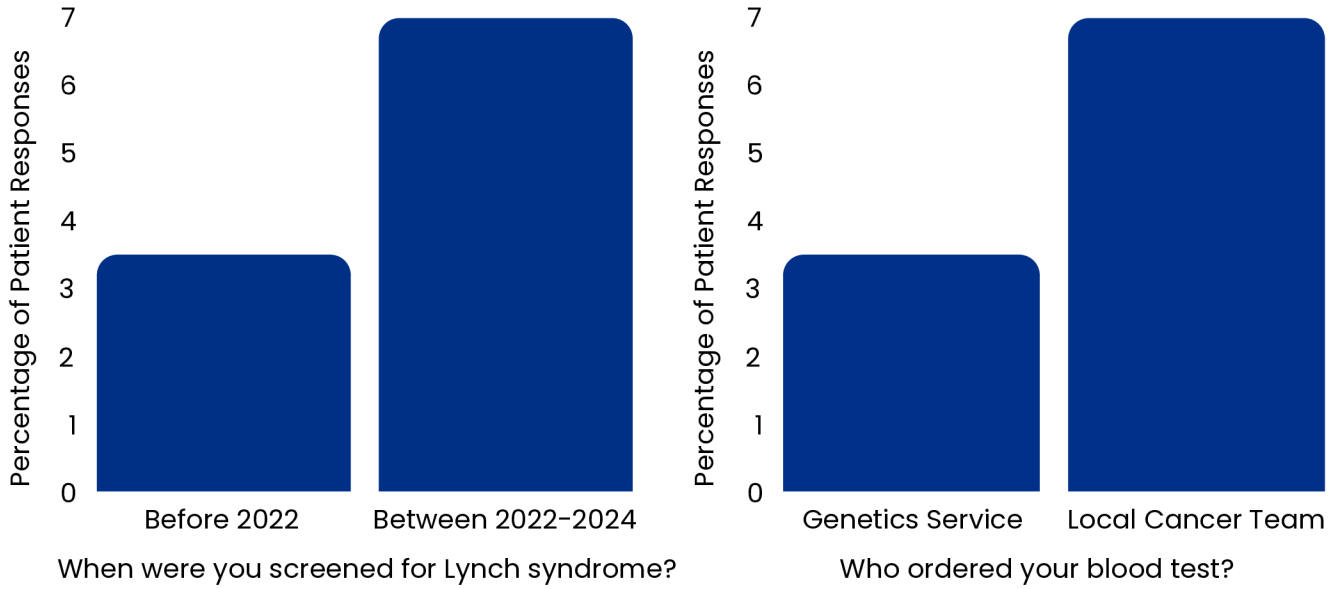
Somewhat negative experiences observed in patients who self-identified as having their blood test ordered by their local cancer team were due to a lack of explanation of the implications of a positive result. Patients stated that they were made aware that the mainstreamed clinic was newly established, and assumed that this missing information was due to a lack of training.

However, many patients who experienced a mainstreamed model of care shared that they had an excellent experience, and that they couldn't suggest any possible improvements to their care. The patients had a mostly positive overall experience of mainstreaming stated that this was because of the ease of their care being delivered by their local cancer teams as part of their cancer treatment with minimal additional appointments.

Overall Experience of Lynch Syndrome Testing Across England by Team Who Ordered Blood Test



Percentage of Patients Across England Who Waited 1 Month or Less for Their Lynch Syndrome Test



The concerns and dissatisfaction regarding long waiting times are reflected in the time taken to receive a Lynch syndrome test in different groups. Those who received their Lynch syndrome test between 2022 and 2024, and those who received their care though a mainstreamed pathway had a higher chance of being tested within the first month.

Screening Pathway

When were you tested?

To better understand the pathway experienced by our respondents, we asked questions to deduce the model of care that they experienced. We asked when participants were screened to understand if they had received their care before or after the improvements made through the National Lynch syndrome Programme, which commenced in 2021. Responses were evenly split, with 51% being screened before 2022, and 49% being screened between 2022 and 2024.

Who ordered your blood test?

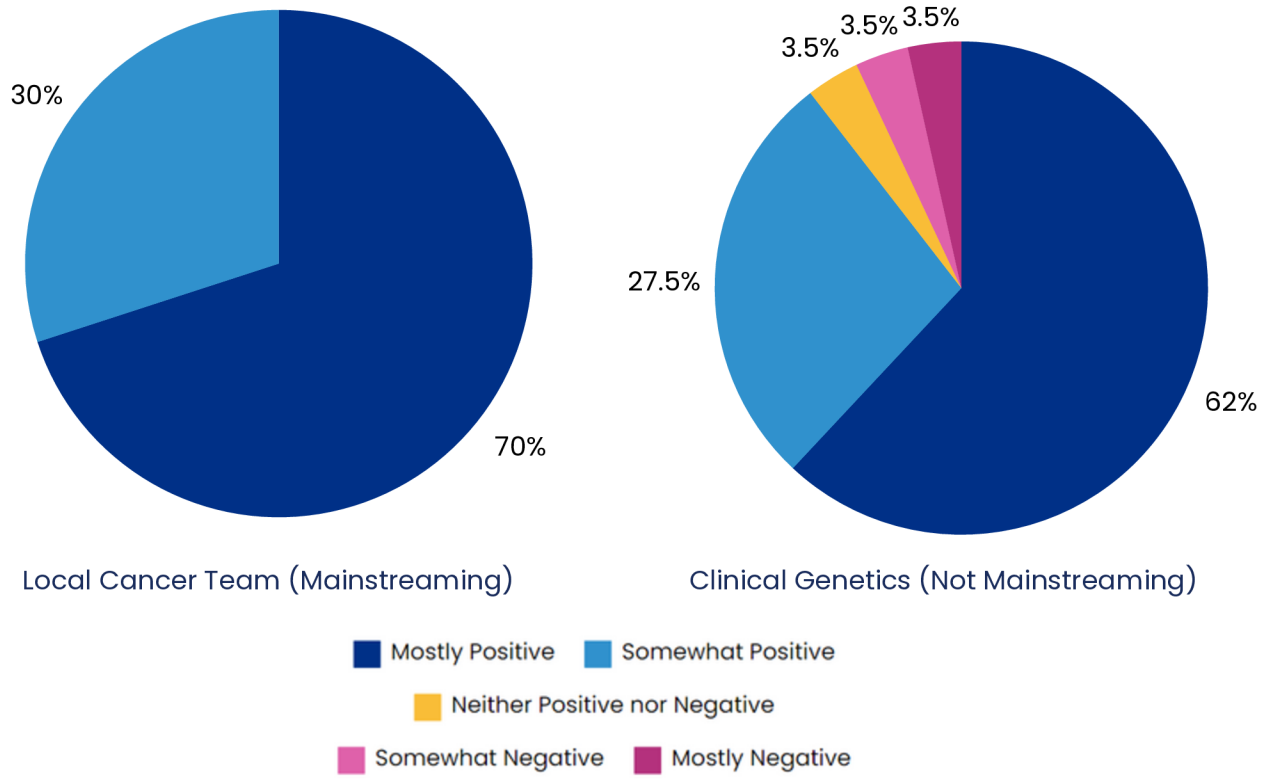
To identify patients who had received their care through a mainstreamed clinic, we asked who ordered their Lynch syndrome test. Nationally, 22% of participants had their blood test ordered by their local cancer team, indicating they their care has been mainstreamed. 67% had their blood test ordered by clinical genetics, suggesting that their care was delivered through the traditional pathway. The remaining 11% were not sure who ordered their blood test.

SOUTH EAST KEY FINDINGS

Overall Experience

100% of patients defined their experience of the Lynch syndrome screening pathway as mostly positive or somewhat positive within the mainstreaming model of care. This was broadly equivalent to patients received their screening through clinical genetics. This demonstrates that care delivered by through a mainstreamed pathway perceived as similar or better than that delivered by a traditional non-mainstreamed pathway provided entirely by clinical genetics.

Overall Experience of Lynch Syndrome Testing Across the South East by Team Who Ordered Blood Test



Faster Access to Testing

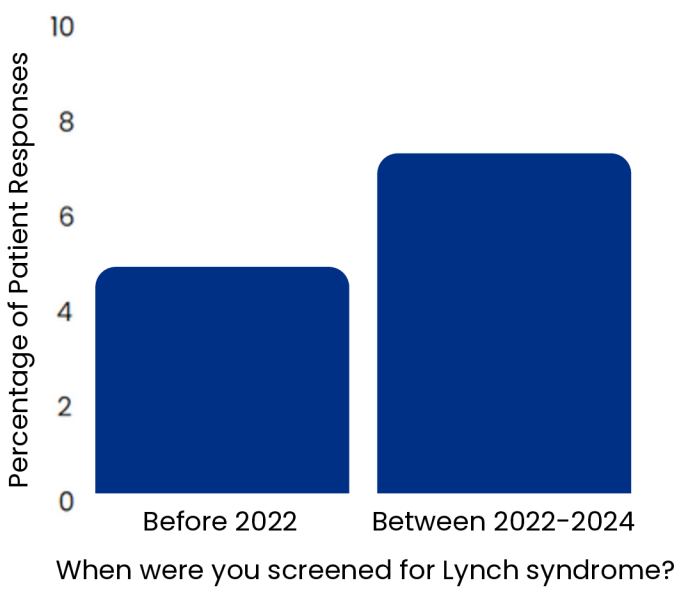
Those highlighting their experiences in clinical genetics (the previous pathway) as either somewhat negative, mostly negative or neither positive or negative, highlighted to barriers to accessing clinical genetics, delays in appointment scheduling, and long waits to receive results. These factors were not present within the new model of care, through mainstreaming.

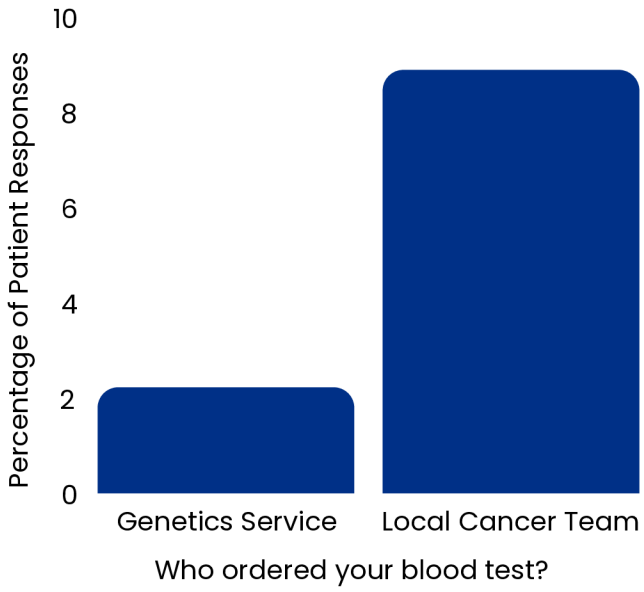
Many patients within the South East who received their care through a traditional non-mainstreamed model shared that the most difficult aspect of the screening process was waiting to be tested, and for subsequent appointments. A patient from the South East who was screened before mainstreaming began in 2022 shared that their experience would've been improved with 'faster access to testing, so that people are not left in limbo for too long'. A patient from South West London expressed a desire for 'Lynch syndrome testing to be initiated immediately after a cancer diagnosis is shared', as it took over 6 months to be offered a test by clinical genetics.

A patient who was not mainstreamed shared that their anxiety would have been helped by 'a quicker appointment to discuss the diagnosis and what happens next'. Other patients described the wait to be seen by clinical genetics as 'a time bomb' and 'torturous'.

As a result of mainstreaming and support from the Regional Lynch syndrome MDT, time taken for a Lynch syndrome test to be ordered is shorter when provided by local oncology services. Patient experience survey data shows that patients who have their Lynch syndrome test ordered through a mainstreamed clinic will have their test ordered within 1 month from cancer diagnosis, whereas patients who have their test ordered by clinical genetics will often have to wait 4-6 months for a test to be ordered, prolonging time to diagnosis

Percentage of Patients Across England Who Waited 1 Month or Less for Their Lynch Syndrome Test





WHAT DID WE HEAR?

Thematic analysis of qualitative survey responses and interview conversations has identified six key themes that describe patients' experiences, needs and perspectives. These themes are explored in detail to highlight key areas of success and improvement throughout the patient journey.

Click on each theme to view:

- Lack of Integrated Care
- Shared Experiences within Families
- Experiences with Clinical Genetics Services and Hospital Care
- Psychological and Emotional Support
- 'Knowledge is Power'
- Providing an Inclusive and Accessible Experience

Based on these themes, targeted recommendations have been proposed to improve patient care and experience, and to address the specific challenges identified through analysis of responses.

LACK OF INTEGRATED CARE

Respondents shared that their experiences with primary care impacted upon their overall experience, indicating a clear need for improvement. We also heard about the lack of integration between their GP, hospital and cancer and genetic services.

Multiple patients expressed a worry that their family members or other patients would "slip through the net" as a result.

Patients discussed feeling dismissed, "belittled" and "fobbed off" by their GP, with their concerns being "written off" because of their age or gender, for being an "anxious patient", or due to having active cancer. Some felt as though they were not listened to when sharing their family history. Many put this down to lack of knowledge or education amongst primary care providers. For many, this was the first hurdle to getting referred for further testing. One patient commented, after a letter had been sent from the Clinical Genetics service to the GP to refer her for genetic testing:

> *I went in for something else and asked them about the letter … they had filed the letter because they thought they didn't have to do anything with it*

Other people had similar experiences across both primary care and oncology services:

> *"The GP surgery where I had the test were very uninformed about the process and did not think they could do the test."*

> *"It would have been better for the GP to have known about the condition, its local pathway(s) and to have correctly referred and prescribed for my history (or to have sought specialist guidance to correctly done the above). This didn't happen. To have had meaningful and timely action/ outcomes around existing risk symptoms - rather than to be 'signed off' due to a lack of an active cancer."*

"Oncologists should have more knowledge as they are the first point of contact for the patient. For me it was quite a worrying few months before I saw the geneticist and the oncologist I saw knew very little about the screening process"

"I am sure that testing and aftercare is much better now. It is quite surprising that so few medical people have a handle on Lynch even now."

"Being informed that you were being tested for Lynch Syndrome and the outcome if there is a positive test. I was only informed that it was negative and didn't even know I was being tested and had never heard of it."

It is important to understand the emotional impact that this has on patients. Several respondents shared that when the first clinician they spoke to was not understanding or knowledgeable about Lynch syndrome, it felt like an additional barrier to overcome.

Equally, we heard about the anxiety and frustration people felt throughout the pathway as a whole. One patient was relieved to have a diagnosis, because for her it meant that she hadn't been 'crazy' all along. Conversely, patients shared that they had a positive experience when their primary care provider was knowledgeable, or who sought knowledge in advance of their appointment.

"Once it was explained to the GP they were better and more interested and understanding"

"Personally, my experience has been well streamlined from start to finish. In part because I had cancer I have been well looked after and my GP has taken a keen interest in Lynch Syndrome. There should be ongoing education and awareness being raised to all health professionals"

However, it should be noted that some experienced poor care, even when this was provided, and that primary care experiences varies across the region. For example, some GPs were happy to follow recommendations from consultants regarding prescriptions, but some would not approve prescriptions, particularly for aspirin.

Another source of frustration amongst our respondents was the lack of integrated care. We heard examples of missing information and records between services, a lack of communication, and stark differences in care across services. This was a source of anxiety for patients. When asked how the service could be improved, patients responded:

> *"I would want better communication between GPs and consultants and all the people who need to know stuff … it makes me worried about patients slipping through the net"*
>
> *"More joined up parts of the NHS system so that I wasn't the one having to chase up the hospital to conduct tests on the tumour and I didn't have to arrange the blood test at the GP surgery and explain why it was necessary and what was needed. While I am capable of doing this it put an extra strain on me in the circumstances and not everyone would persevere."*

For some patients, the diagnosis of Lynch syndrome and the "letter from genetics" has improved their care and the integration of their care:

> *"Since the Lynch genetic letter hit them things have moved quicker"*

We heard from a number of people that a person centred approach, whereby clinicians took time to understand other things happening in a patients' health or life was much preferred.

At the time of screening for Lynch syndrome, people told us they were also dealing with cancer diagnoses, diagnoses and care of family members, and thinking about the future of their children, or potentially not having children. For example, one young woman from the South East talked to us about how the medications affected her birth control decisions. Another discussed how they were not tested for Lynch syndrome until the end of their pregnancy, as it could cause emotional distress and it was difficult to make decisions. These people felt that it was important for clinicians to remember that screening for Lynch syndrome doesn't happen in isolation.

Recommendations

1

Training for GPs & oncologists in Lynch syndrome needs to be a priority.
The majority participants were generally accepting of genetic testing and preventative screening, and recognised that these steps were beneficial to themselves and their family, but more information is required.

2

Improved communication between services about the Lynch syndrome pathway
Patients should not be made to feel the need for chasing up appointments or results.

3

Taking a person-centred approach to Lynch syndrome.
Recognising that there will be multiple factors affecting someone's health decisions, and that everyone needs personalised health plans.

EXPERIENCES WITH CLINICAL GENETICS SERVICES AND HOSPITAL CARE

Many people who shared their experiences with us spoke about their genetic counsellors by name, and the positive relationship they had with them. They talked to us positively about how their genetic councillors explained Lynch syndrome in the appropriate amount of detail, whilst remaining compassionate. This was achieved by genetic councillors showing patience, and taking the time to explain the meaning and impact of results thoroughly.

Many respondents had a positive experience of clinical genetics, as the 'door was left open', with a named contact to speak to if ever in need. Although only a few patients had ever actioned this offer, knowing that the option was there for them and their families was extremely reassuring. For example:

> *"I didn't feel frightened due to the support for me and my daughter"*
>
> *"The Genetics team checked in with me and left the door open, it is nice to know that someone was there. I wanted to do this survey because I'd had a really good experience, it was easy and all the services ran well together"*
>
> *"Having Genetics [professionals] that had excellent knowledge. The scope of his knowledge and support have made a huge difference … my hospitals have been brilliant excellent care and experience for something so horrible."*

Clinical Genetics services were praised for being compassionate and understanding, and not just treating patients as Lynch syndrome case:

> *"The Genetics unit is one of the best bits of the NHS – they dealt with patients not like a piece of meat, and you never felt that they were being dismissive"*

The importance of a named or known contact also extends into other services; for some this was dedicated cancer clinical nurse specialists, gynaecologists, or other consultants delivering mainstreamed patient appointments.

Their knowledge and understanding made them stand out positively to their patients. Continuous and consistent care was also important to people, which could be provided when care was delivered by local cancer teams.

> *"It was the same person who operated on my mum. They explained the surgery option and made it clear that it was my decision … it helped to speak to someone that knows and is not pressurising"*

However, this was not the case across everyone we heard from.

We heard examples of clinical genetics teams that had not shown compassion. Some patients were worried that they had been "forgotten" by the clinical genetics service, and that they wouldn't receive an appointment. Similarly, others did not experience the "open door", and felt that there was no one to contact following their diagnosis.

When asked what could be different or improved, multiple people expressed a desire for a dedicated clinic or specialist clinician in a single service where this had not been provided. For example:

> *"If the gynae oncology nurses had been able to do the testing and counselling themselves instead of me having to wait for 9 months for formal confirmation of the diagnosis and genetic counselling."*
>
> *"A Lynch clinic for annual one stop screenings with people who know their stuff. Access to research trials locally."*
>
> *"Dedicated Lynch screening centres with good relationships with and pathways to cancer centres; with dedicated diagnostics and screening equipment"*

These responses further evidence the need for mainstreaming in order to make care more equitable, and to reduce geographical variation across the region. A few people expressed that they would want to access care elsewhere due to the quality of the service.

> *"The national variation for non-bowel cancer prevention really worries me, it seems to be a lottery as to what you are offered. As an NHS employee, the fact that I am considering paying myself to get a private gynaecology consultation is heart-breaking, I feel really let down."*

When asking patients what could have been improved or done differently, multiple said that having a named, familiar person, or dedicated Lynch clinic, would make a positive difference. This matches the positive experiences of people who did receive co-ordinated care.

Recommendations

1. **Every patient should have a named contact in a mainstreamed clinic**
Standardising and scaling the 'open door' approach to enable patient to contact their named person for support through the screening process. This is the singular biggest change needed to positively impact on someone's personal experience.

2. **All clinicians providing mainstreamed clinics should have access to appropriate guidance. This knowledge can then be shared with patients at the point of care.**
The RMP Beginners Guide to Lynch syndrome is recommended.

3. Where possible, the same named contact should extend across family members to provide consistency and improve person-centred care.

4. **Reduce pathway variation and ensure geographical equity of care.**
There is vast variation in quality of care, creating a healthcare 'lottery'. This means that a family may experience disjointed care when cascade testing.

5. **Dedicated, specialist clinicians trained in Lynch syndrome mainstreaming** should be provided for all patients.

‘KNOWLEDGE IS POWER’

Many respondents emphasised that “knowledge is power.” Receiving their results gave them a greater sense of control, understanding and peace of mind. This new-found clarity helped them make sense of their family history. Two patients reflected:

“It explained why so many in my mother’s family died young.”

“It’s encouraging to see progress. When my mother had her endometrial cancer, she was told it wasn’t hereditary, but now we know it is and can test for it.”

Beyond providing answers, many participants pointed to the positive impact of being able to take proactive steps, such as increased monitoring and surveillance. Many described their diagnosis as “life-saving”, as it allowed them to pursue proactive measures such as regular colonoscopies, aspirin use, and risk-reducing surgeries. Access to genetic counselling and clinical support further empowered patients with both early detection and clear action steps, helping them to feel more confident about managing their future health:

“The diagnosis led to a colonoscopy and endoscopy, where a 5cm benign tumour was found. I had Whipple surgery, which saved my life.”

“I am grateful my womb cancer was tested for Lynch or I would not have known. This way I have access to potentially life-saving information to take aspirin and to have regular colonoscopies.”

The benefits of this new knowledge extended to their families too. Some respondents felt reassured about the ability to begin the process of cascade testing, offering relatives the chance to detect Lynch syndrome early on:

“I am glad I was tested, it led to my brother being tested who had a positive result so he knows he needs more monitoring in the future.”

Despite these positives, many people also expressed regret about not knowing their diagnosis sooner, believing it could have significantly changed the course of their illness:

> *"I wish I knew earlier, maybe I would have been Stage 1 instead of Stage 3b."*
>
> *"If I had known I had Lynch Syndrome earlier, I would have been monitored and might not have incurable endometrial cancer now."*

Additionally, some participants reported having a family history indicative of Lynch syndrome but were unaware that it existed. This highlights a need for greater awareness and the importance of educating patients and professionals about Lynch syndrome.

Others shared frustrations around the lack of information available, with one person stating that they found all information themselves whilst waiting for a genetic counselling appointment. Another shared that they were supporting their nephew in preparation for his predictive test, as he has not received any information directly.

Many patients expressed a need for clear, accessible information or e-resources about Lynch syndrome, covering fundamental knowledge such as what it is, its link to cancers, and the benefits of testing. Numerous patients also mentioned that they would like to have more information about enhanced screening and surveillance pathways, preventative treatments, how to discuss the condition with relatives, and how it may affect relationships and family planning. A number of respondents shared that there were inconsistencies in existing information. A review of this information would help individuals to manage their care independently, and to understand more about living with Lynch syndrome.

One patient felt that the tone of this information should be approachable and "light-hearted," whilst another suggested that:

> *"Puns and clever wording will grab the attention of young people in a sea of other information."*

One participant spoke about the "fear" associated with the statistics used to raise awareness about cancer, but these figures can actually prevent people from accessing screening. Some participants told us that they received contradicting information from various hospitals,

highlighting the importance of providing consistent, factual and reassuring information e.g. 'screening reduces risk but does not negate it', '[Lynch syndrome] is not a death sentence', and that 'aspirin reduces risk but does not eliminate it'.

Support tools, such as the Lynch syndrome app by East Genomics, have been highlighted as valuable resources. Participants found that such tools aided focused conversations around approved guidelines, and assisted interactions with pharmacies and GPs when explaining the effect of certain variants on medication. Additionally, patients stated that patient information days hosted by Lynch syndrome UK provided a valuable forum for patients and families to learn more about the condition.

This feedback evidences the value of support networks and charities such as Lynch syndrome UK, especially when managing the emotional aspects of Lynch syndrome screening, diagnosis and management.

Recommendations

1. **Standardised patient information should be created and utilised across all regions and NHS providers**
Targeted and accessible information for those with a family history of relevant cancers is also required, underscoring the importance of early screening and how it can change the course of a diagnosis.

2. **Lynch Syndrome App should be widely promoted** among healthcare providers to help increase awareness of the condition.

3. **Everyone diagnosed with Lynch Syndrome should be given information about Lynch Syndrome UK**, online support forums, informational webinars, and patient days to offer a channel for peer support.

4. **Standardised information about 'Living with Lynch',** talking to relatives, family planning, and mental health support should be created.

SHARED EXPERIENCES WITHIN FAMILIES

Many respondents' experiences of Lynch syndrome extend to their family, as a diagnosis also impacts upon their relatives. Many people talked to us about their own experience being linked inextricably with that of their family. Patient's experiences often become shared with their family. For instance, even if one person's experience of testing was positive, their overall experience may be negative if their family members experience of cascade testing was poor. For example:

> *"There is a full history of cancer in my family. Lost both parents, sister had it twice and survived and now I have had it. So this type of genetic testing is important to me especially having 3 teenagers who are concerned and are aware of my family background. Now I am three years out I don't feel I have anywhere to go and ask without feeling an inconvenience. I just want my children to be informed for their future."*

For example, one couple that we spoke to told us that they "discuss it as a family and keep their medical notes together", whereas others rely on their family to work through the screening process together.

> *"I was lucky to have a daughter that handled everything with much determination. As a family we were able to discover that after testing I was the carrier of the MSH6 gene."*

Many told us that they had different experiences to family members based on their age or location, or just by chance. This can heavily impact overall patient experience, as if someone personally had a smooth experience, their experience may be negatively affected if their family member does not. This further evidences the importance of creating an equitable service.

As well as a shared experience of Lynch syndrome testing, people often come from a family with history of cancer, whom they may be supporting or grieving.

> *"If you go back, we've all had cancer, it's horrendous … the family history is frightening"*

One person discussed how receiving a diagnosis of womb cancer made them feel like they'd "hit the jackpot", as one of their family members had died of bowel cancer which had spread.

They were very thankful that their cancer had been diagnosed at an early stage, given that they were worried that the same would happen to them. For another patient:

> *"I think that's the most stressful part, finding out if you've passed it onto your children"*

When making decisions regarding Lynch syndrome testing and onward management, consequences for family members is often considered as a priority for many patients. Therefore, the potential impact upon family members should be considered by healthcare professionals when guiding patients through this process. This can be achieved through mainstreamed patient appointments delivered by familiar clinicians from their existing cancer care team. Many patients told us that the impacts on future generations was one of the most important factors when considering next steps:

> *"I took the hysterectomy because I would feel guilty if my child had cancer."*

A number of respondents shared that one of the most difficult aspects of the screening process was gathering family history paperwork and gaining consent from those concerned. This can be particularly hard if the family is disconnected. To mitigate this, participants shared that their relatives were invited to send family history information directly to clinical genetics services. We also heard a clear need for support in communicating a Lynch syndrome diagnosis to family members. Patients also asked for additional support during cascade testing, as long waiting times can cause anxiety and feelings of guilt. One patient shared that they were able to bring their family members along to a Lynch information session, which was extremely beneficial. They reminded us that it can be just as frightening and confusing for the family as it can for the patient themselves, and that obtaining information and support is crucial. This demonstrates the requirement for additional psychological support and resources, in which family and relationships should be considered.

Recommendations

1. **New processes around cascade testing** should be considered to improve support and information provided for the patient and their families.

2. **Consider concerns of/about family members**
As part of person-centred care delivery, it should be considered that a patient's fears and decisions may be influenced by the experiences the experiences of their family members. As such, discussions of family history, even when it is not deemed clinically necessary, may be beneficial.

PSYCOLOGICAL AND EMOTIONAL SUPPORT

A strong support system is essential for Lynch syndrome patients, as the journey from cancer diagnosis to receiving genetic results can be emotional. It is imperative to recognise the multifaceted experience that patients go through. For instance, patients could be dealing with cancer treatment simultaneously with discovering that they have an inherited genetic condition, and that this may have been passed on to their children. As one person voiced, it was "a bit of information overload as I was going through chemo and stoma reversal, so was not fully in possession of my physical and mental strength". Others may have experienced a loss within their family, which may have been caused by a Lynch syndrome related cancer, with a patient sharing that they 'had just lost my dad through bowel cancer and then I got diagnosed."

Many patients highlighted the long wait for results and appointments. One person described this experience as "lonely." A number of these people told us this period of uncertainty was their main source of anxiety, regardless of the result.

> *"I was not happy about having to wait so long to deal with the emotional burden of knowing that I could pass this gene mutation on to my children and grandchildren."*

> *"Being handed leaflets at the hospital and doing own research was isolating. Then the 5 month wait to see genetics, only to be told what I'd found out myself was disappointing. I'd expected more support in the form of appointments or a specialist. I feel that unlike other illnesses I'm on my own with this one."*

Some people recognised that faster diagnosis may not be possible due to current pressures within NHS, and suggested that in order to alleviate concerns the service should provide reassurance through updates on waiting times. It was also suggested that dedicated point of contact for inquiries from people who hadn't been contacted with an agreed time frame would have been appreciated.

Many patients commented that whilst they had strong network of friends and family to support them, this was not the case for everyone and "the state of being in limbo could affect [those who were processing the diagnosis alone]' and that results could be "damaging to their mental health." This further evidences the need for emotional support networks.

To address these challenges, patients recommended that increased psychological and emotional support should be provided throughout the entire screening process. One patient mentioned the "lack of psychological support, especially at the start" whilst another mentioned the importance of "preparing people for the emotional impact" of a diagnosis. This support is also necessary post-diagnosis, with patients suggesting that the service should provide mental-health support through a variety of methods. Suggestions included appropriate sign-posting to information, emotional support services, a dedicated helpline, annual follow-up calls and consultations with trained mental health counsellors or therapists. One person likened this to the role of a Macmillan nurse, who could offer compassionate support throughout the journey.

Crucially, some patients may wish to access support at different time points along the pathway, and it is important that access to this support is available when required.

> *"Sometimes in my cancer and CMMRD experiences people have asked if I wanted to talk to someone or they have given me a leaflet for counselling but it's been too soon when I didn't feel I needed or wanted it. Actually it was only a while afterwards when everything hit and it would have helped to have some counselling, but by then you're sort of forgotten about."*

Patient networks were recognised as an extremely valuable support system. A number of people told us that they would like to be connected with others who have been diagnosed with Lynch syndrome, whilst several commented on how the charitable organisation Lynch syndrome UK has provided a "sense of not being alone" via their Facebook group and in-person events. However, a few individuals found the group difficult to engage with as it was "all about cancer." One male respondent mentioned that it would be beneficial to create specific spaces for men to connect about their experience with Lynch syndrome, as existing groups were mainly populated with women.

The variety of feedback and experiences evidences the need for a range of varied support to meet a multitude of different needs. A one size fits all approach is not appropriate.

Recommendations

1. **Regular, clear and consistent communication for patients whilst awaiting appointments or results is necessary.** Suggestions include updating patients about expected wait times, information to read during this waiting period, or creating a contact helpline to ensure that patients do not feel like they have "slipped through the system."

2. **Increased psychological support.**
Processes should be in place to refer patients for further psychological support, with a range of flexible options.

3. **Everyone diagnosed with Lynch syndrome should be given information about Lynch syndrome UK**, online support forums, informational webinars, and patient information days to offer a channel for peer support.

PROVIDING AN INCLUSIVE AND ACCESSIBLE EXPERIENCE

Providing an inclusive and accessible experience for all patients is essential to delivering compassionate and personalised care for the diverse range of people that access genetic and oncology services.

Some patients, particularly those with accessibility needs, told us that they had a preference for remote appointments. For example, one patient who identified as neuro-diverse with ADHD told us that they found travelling to cities overwhelming, which would've introduced additional difficulties when attending appointments. However, others who had remote consultations shared that a face-to-face session with genetic counsellors would have promoted a more personal connection, especially following a Lynch syndrome diagnosis. This highlights the importance of offering flexible options to allow patients to choose the environment that best suits their needs.

One person commented on the importance of genetic counsellors tailoring the approach of their conversation, dependent on the patient's emotional reaction. People will react differently to a diagnosis, and will have varied expectations of the service.

Another key aspect of providing a service that is open to all is ensuring gender inclusivity and respecting patients' individual identities. One patient discussed his experience as a male of trans history:

> *"As a trans person, I had the symbol associated with my birth sex on my chart (a circle, as opposed to the square for men). I understood why, but that wasn't comfortable. However, when I actually went for my face to face meeting, the person I spoke to said I should have a hexagon as that's what's 'on offer' for trans people. It would have been good to know about this [and] for that have to been made clear in advance."*

In further conversation, this person reflected that it is important to ensure that clinicians are aware of symbols and terminology that are used to identify patients. They highlighted that a similar experience could be very difficult for those who may not be as open about their trans history.

Recommendation

1. **Training given to all clinicians** (not just those working with Lynch) about gender inclusivity and how to ensure that patients feel welcomed and respected within any NHS service.

CONCLUSION

Our aim was to hear a range of voices about their experience of being screened for Lynch syndrome.

We've heard six key themes from these voices, with both positive and negative experiences. The majority participants were generally accepting of genetic testing and preventative screening, and recognised that these steps were beneficial to themselves and their family. Although, we heard that there is ongoing **frustration with healthcare providers who do not have a working knowledge of Lynch syndrome**. This can greatly affect access to testing and referrals, as well as overall experience of care and emotional support.

We hope that this report will encourage continued action to **increase education** for healthcare providers. There was a strong desire for a **knowledgeable and accessible point of contact**, both during testing, and after the diagnosis of Lynch syndrome is made. Patients longed for a **dedicated named person** or specialist Lynch syndrome clinic, of whom they could ask questions, knowing that would be understood and treated with compassion and patience, and that their concerns would be taken seriously.

This report has clarified that there is a great need to **reduce inequity** and to **increase integration of care**. The need for **increased psychological support** for patients before, during and after diagnosis was also clear, particularly for those who are supporting family members through cascade testing with a long wait for results.

Furthermore, this report has revealed that a patient's experience is not only based on their personal journey through screening, but is heavily based on that of their family. This is particularly important when evaluating services, highlighting the need for ongoing patient experience analysis such as this.

We urge our partners to review the recommendations of this report and continue to work towards an improved and more equitable service for everyone who is screened for Lynch syndrome.

LIMITATIONS

We would like to recognise the following limitations of this report:

- The distribution of the survey was not the same across all areas of the South East region, and the patient sample for interview and survey response was self-selecting. We therefore are unable to deduce if this sample is representative of the wider patient population. A more representative cross-section of respondents would be important for future research.
- Due to limited resources, we were unable to interview every person who offered their consent. We hope to provide opportunities for these people to share their feedback in the future.
- The survey and interviews were available in English only, and survey itself was accessible online. This may have excluded certain groups from participating and will be changed in future approaches
- Thematic analysis was completed manually, and although thorough, could be open to unconscious bias.

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