



**Lynch
Syndrome**
Australia

Many families. Many cancers.

One common cause.



**Misdiagnosed, misunderstood
and missing out:**
Lynch Syndrome
Australia's untold health story

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A report by Lynch Syndrome Australia - 2017



About Lynch Syndrome Australia

Lynch Syndrome Australia (LSA) established in 2013 is a registered charity and an all-volunteer survivor-led organisation that supports the many thousands of Australian families that carry the Lynch syndrome gene mutation.

Whilst other cancer organisations and charities concentrate on specific cancers, LSA concentrates on a genetic condition where people affected have a lifetime risk of many different cancers and sometimes more than one primary cancer at once.

LSA does this by:

- raising awareness of Lynch syndrome amongst health professionals and the public
- hosting *Living with Lynch syndrome* events, seminars and conferences
- developing tools and resources for families and health professionals to help those affected to manage their medical care and to reduce their cancer risks and
- actively engaging in and contributing to the health care reform process, both as a peak body and in collaboration with other consumer organisations to ensure the unique needs and concerns of families with Lynch syndrome are recognised, understood and given due consideration.

LSA is governed by a Board of Directors and supported by a Scientific Advisory Committee.



Lynch syndrome is believed to be Australia's most common inherited cancer risk
One in 280 Australians is believed to carry the mismatch repair gene fault
Approx 85,000 people are at risk of developing one or more primary cancers
Yet, only 5% of Australians with Lynch syndrome have been diagnosed

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Just another Lynch syndrome story

I grew up surrounded by cancer. My grandma, two aunties and one great uncle all died from cancer when I was a girl. Then it was my mum's turn. Cancer. Again. More dreadful treatment but still the same sad outcome. My brother, sister and I become adults, expecting the worst. When I was 29, the worst happened. By then, my cancer was really no surprise. What was a surprise is that, this time, they had a name for it: Lynch syndrome. A gift from the past.

It was awful going through the genetic testing process with my brother and sister. My brother and I tested positive but not my sister. She felt so guilty, she just stepped away from us. Survivor guilt, I think they call it. Two cousins even refused the test. They don't want to know if they are at risk. It's sad because they have three children between them who could lose a parent due to a preventable cancer. Meanwhile, we face having check-ups for the rest of our lives - if we don't want to wait for cancer to rear its ugly head again. The wickedly unbearable colonoscopy prep will now be part of my life every year from now on. I also have to watch out for the vague symptoms of all the other Lynch syndrome cancers that have plagued the family. Tricky, because no-one can agree about how this should be done.

Somehow, I always assumed I'd be a mum. Now I wonder whether I will ever have children. My risk of endometrial cancer is about 50%. My cousin died of ovarian cancer at 34 years of age. My doctor advised me to have hysterectomy, soon. I am 33 years old. It feels so young. But there again, I sometimes wonder what right do I have to give this gene to a child, anyway? There are other options, I know, but it all seems so overwhelming.

At least I have family members who understand what I am going through, because they face it too. And I am lucky - I have doctors and nurses who know about Lynch syndrome and they help me find my way through the system. My brother lives in a regional town. He's not so lucky. His GP doesn't understand Lynch syndrome and getting a colonoscopy every year is a real fight. No-one understands how important it is that screening happens on time. I am worried he'll just give up arguing and go without.

Yes, the future is uncertain. Not just for my health but financially, too. I can't get life insurance or mortgage insurance. My marriage didn't outlive my diagnosis. I don't really blame him - I can hardly face the worry myself, some days. I worry I can't find the gap fees to see my gynaecologist, or other specialists. But I can't afford to risk the delays in the public system.

This is just my Lynch syndrome story. Worse than some, but not half as bad as many others I know. There are plenty of challenges but I am grateful, too. I'm already better off than earlier generations. My first Lynch syndrome cancer didn't kill me, like it did them.

Lynch syndrome is my life's challenge, not my death sentence. There are 85,000 other Australians facing the same challenge. Same but different.

And there are changes we all need to see; changes to help us meet these challenges, together.

Misdiagnosed, misunderstood and missing out

Lynch syndrome is believed to be Australia's most common inherited cancer risk¹. One in 280 Australians is believed to carry the gene fault responsible for Lynch syndrome; approximately 85,000 people who are at risk of developing one or more, often-aggressive, primary cancers² in their lifetime. Yet only a few thousand of them actually know it because only around 5% of Australians with Lynch syndrome have been diagnosed.

Lynch syndrome can be a lonely experience. It is poorly understood by clinicians and virtually unheard of by the general public. There is no universal testing, no agreement about surveillance, no Lynch syndrome centre of excellence. Support services that understand the needs of people with this life-long, multi-cancer, multi-generational syndrome are rare. There is no public information campaign or health funding for Lynch syndrome and none dedicated to the complexity of medical, emotional and financial challenges that Lynch syndrome presents for lifelong care and support. Many people feel isolated and frightened by their diagnosis. Some feel angry that they are not properly informed, advised and cared for. If you have Lynch syndrome, you just don't seem to belong.

To guide our advocacy work and identify issues for priority attention, Lynch Syndrome Australia (LSA) set out to better understand the personal experiences of families living with Lynch syndrome. In 2015, LSA conducted a world-first, global survey of 461 people from Lynch syndrome-affected families. The survey was developed over eight weeks, piloted internally and externally, supported by members of LSA's Scientific Advisory Committee and endorsed by Dr Henry Lynch, the American clinician and 'father of cancer genetics', after whom Lynch syndrome is named. The survey's 50 questions covered topics such as: demographics; the experience of diagnosis; cancer risk management; individual experiences with cancer and treatment; Lynch family cancer history; navigating the health system and the psycho-social impact of living with Lynch syndrome.

Of the 461 participants, 251 were from Australian families and their responses shape the recommendations in this report. Survey results have been presented at clinical conferences and discussed with leading Australian clinicians and researchers. LSA also commissioned researchers from Macquarie University to undertake further analysis of the data.

We have learned much from the survey findings. As a result, LSA is launching a campaign for changes to clinical practice, cancer health policy and service provision. These aim to improve the identification, risk management and support for people affected by Lynch syndrome because it is now crystal clear that so many Australians with this condition are **misdiagnosed, misunderstood** and are **missing out** on essential and appropriate care and support.

And it's time for that to change.

Lynch syndrome is poorly understood by clinicians and virtually unheard of by the general public.



Summary of recommendations

Lynch Syndrome Australia is advocating for:

The Misdiagnosed

- Universal tumour testing of all bowel and endometrial cancers, regardless of age
- Referral pathway protocols between hospitals, the community and genetic services so no-one falls through the cracks
- GPs to take a family cancer history for every patient to detect families at risk
- No genetic testing without genetic counselling for people suspected of having Lynch syndrome, so that everyone gets the right support and advice
- Mandatory inclusion and communication of the exact mutation in test results so recipients can better understand the risk relating to their particular gene fault

The Misunderstood

- A national register for individuals with Lynch syndrome so that everyone with Lynch syndrome counts and can be counted.
- A National Clinical Care Standard for individuals with Lynch syndrome so that GPs, clinicians and patients know what they need to do to stay well
- Universal access to treatment that meets the clinical care standard so that a person's postcode, wallet or understanding of the condition will no longer affect how their Lynch syndrome cancer risk is managed
- Information campaigns to ensure people with Lynch syndrome and those who care for them understand:
 - that the cancer risk -reducing potential of healthy lifestyle and behaviour is even more important for those with Lynch syndrome
 - the differences in risk associated with different genes to help inform decisions relating to treatment and preventative surgery

Those Missing Out

- A care coordinator so no-one has to navigate the health system alone
- Simplify complex information so everyone can make the best choices

Lynch syndrome in Australia - A brief overview

What is Lynch syndrome?

The basics

- Lynch syndrome is a mistake (mutation) in a gene that should be repairing new cells
- Each child has a 50/50 chance of inheriting the mistake from a parent who has it
- The gene that causes Lynch syndrome does not skip generations, so if a parent has not inherited it then none of their children is at risk
- The gene mistake results in a high risk of getting cancer, starting at a younger age than normal
- Someone with Lynch syndrome often has a strong family history of cancer; often different types of cancers that don't seem to be connected
- The average lifetime risk of having a Lynch syndrome cancer is around 70%, although not everyone who inherits the gene mistake will develop a Lynch syndrome-related cancer
- People who have one Lynch syndrome-related cancer, have a much higher risk of a second than someone who has not had cancer
- Once the Lynch syndrome gene mistake has been identified in a family, a simple blood test can check whether other family members have inherited it

The science

- Lynch syndrome (formerly known as HNPCC) is characterised by a mutation in one of four genes that repair mistakes in cells when they divide (i.e., mismatch repair (MMR) genes)
- The MMR genes affected are MLH1, MSH2, MSH6, PMS2
- Everyone has two copies of each gene, one inherited from their mother, the other from their father
- If one of these copies is faulty, as in Lynch syndrome, the person carrying the gene mutation has a higher risk of developing one or more of up to twelve different cancers
- The average age that people with Lynch syndrome develop their first cancer is about 40 years of age but cases of teenage cancer are also seen, compared with 60 years in the general population
- Cancers that are considered to be associated with Lynch syndrome are located in the following organ/systems:
 - Colon/rectum (also known as bowel cancer)
 - Endometrium (sometimes described as womb cancer)
 - Ovaries/fallopian tubes, stomach/duodenum
 - Biliary tract/gallbladder, pancreas
 - Urinary tract, kidney/ureter
 - Skin, brain, breast, prostate

The history

- Lynch syndrome is named after an American doctor, Dr Henry T. Lynch, who has worked tirelessly from the 1960's until today to establish the hereditary link for some cancers
- Dr Lynch is an enthusiastic supporter of consumer advocacy and has been a loyal ally of Lynch Syndrome Australia from its beginning in 2013

What we know (about Lynch syndrome in Australia)

It's not rare

- 85,000: Number of people in Australia with Lynch syndrome; approximately 1:280 people³
- 5%: Estimated percentage of those with Lynch syndrome who have a confirmed diagnosis

It's underdiagnosed

- 80,750: The estimated number of people with Lynch syndrome who are not yet diagnosed
- 1,304: The number of people confirmed as currently registered on state-based hereditary cancer registries⁴
- 3: Number of years it takes for a person who knows they may be at risk to actually decide to get tested and then take the test⁵

It's a lifelong cancer risk

- 75%: A woman with Lynch syndrome's lifetime risk of developing cancer, to 70 years⁶
- 72%: The cumulative lifetime risk of any LS cancer for people who have two of the most common gene mistakes in LS (i.e. MLH1, MSH2)⁷

Lynch cancers happen at a younger age

- Under 50: Age at which half of those with Lynch syndrome who develop cancer are diagnosed compared with 10% in the general population.
- 40 - 50 years old: Average age range at diagnosis of first Lynch syndrome cancer⁸
- 50 years: average age of endometrial cancer diagnosis (versus 72 years in the general population)

Cancers develop much more quickly

- 30 months: Average time it takes for a Lynch syndrome associated bowel cancer to develop⁹ from a polyp
- 10 years: Average time it takes for a sporadic adult cancer to develop

More than one cancer

- 2: Average number of cancers for those who develop cancer due to Lynch syndrome
- 1 in 16: Chance of developing more than one Lynch syndrome cancer at the same time¹⁰
- 12: Number of different cancers already confirmed as associated with Lynch syndrome

It's isolating

- 1: Number of peer support counsellors trained specifically to support those with Lynch syndrome
- 0: Number of specialist support, survivorship or wellness programs designed for those with Lynch syndrome

What we don't know (about Lynch syndrome in Australia)

Misdiagnosed

- Who are the 95% of Australians with Lynch syndrome yet to be diagnosed?
- How many opportunities to diagnose Lynch syndrome in families have been missed by failing to conduct a tumour test?
- How many people might have been misdiagnosed?
- How many doctors follow best practice and ask about family cancer history?
- Since no central records are kept, the number of Australians offered genetic testing is unknown.
- How many have had genetic testing?
- How many turned down the opportunity for genetic testing? Why have they done so?
- How many diagnosed people have not informed their blood relatives of their diagnosis?

Misunderstood

- How many Lynch syndrome-associated cancers occur each year in Australia?
- Why is there such discrepancy between current surveillance and prevention protocols and the actual measures people are taking to protect themselves?
- Why do these protocols differ so markedly from country to country?
- How many cancers could be prevented or detected earlier with greater investment in early screening, detection, surveillance and survivorship?

Missing out

- With tens of thousands of people at risk, why are there no campaigns to encourage awareness of patterns of family cancer?
- Why is so little information about Lynch syndrome available to health consumers and health professionals?
- Why is there so little help for people to manage this complex condition?
- Why do people report problems accessing essential medical surveillance on a timely basis?
- Why do 1 in 5 people with Lynch syndrome report problems obtaining life insurance?



1

Misdiagnosed

People usually find out that they have Lynch syndrome in one of three ways:

- after a cancer has been detected and testing of the tumour raises the possibility; or
- when another family member tests positive for the genetic mutation and notifies them that they might also be at risk, or
- through their own investigations into family history after observing an unusually high rate of cancer in other family members.

Health professionals also follow these same pathways. They look at the four proteins made by the Lynch syndrome genes and if the stain is absent the gene associated with that protein may not be working. They consider the family's cancer history, or the diagnosis of a specific family member. Where this indicates possible Lynch syndrome, the patient should be referred to a Family Cancer Clinic for genetic counselling and, where relevant, genetic testing.

1.1 Tumour testing

Tumour testing involves subjecting a sample of tumour tissue to Immunohistochemistry (IHC)/ microsatellite instability (MSI). This test looks for loss of proteins that indicate whether any of the four MMR genes are not working in the tumour¹¹. The result of the testing determines whether the patient should be referred to a Family Cancer Clinic for further genetic sequencing to determine whether the mutation is in the germline (i.e. inherited, as in Lynch syndrome) and, if so, to describe the specific location of the mutation. If the IHC testing indicates a negative staining (loss of protein) result for the MLH1 gene, then the sample needs to be checked to see if the mistake is likely to be just present in the tumour: a BRAF V600E. This is a relatively inexpensive test can distinguish broadly between the germline mutations that occur in the MLH1 gene (e.g. those in Lynch syndrome carriers which can be identified through genetic sequencing) and the far more prevalent somatic changes (e.g. due to environmental influences) that randomly occur in genes over time. Once formally diagnosed, a person with Lynch syndrome can start taking steps to protect him/herself and their family.

Tumour testing can be performed on tissue samples immediately after surgery, or on the small samples stored by pathology laboratories for many years after surgery was conducted. This is helpful when investigating family cancer history.

Unlike the UK, Australia does not currently advocate universal testing of *all bowel cancers* for MMR faults. Further, while some Australian health services do require the routine testing of bowel cancer tissue (and in some cases, endometrial cancer tissue) from tumours occurring in younger patients (i.e. patients under 50 years) a translational research team working with two large Australian hospitals to improve the number of people being diagnosed with Lynch syndrome, discovered that fewer than half (40% and 13% respectively) of high risk tumours were being referred to the genetic testing services for diagnosis¹².

The consumers' experience

Our survey asked participants about the process of diagnosis for themselves and other family members. Questions included:

*At what age were you diagnosed?
How were you diagnosed with Lynch syndrome?
When were you tested? Prior to or following a cancer diagnosis?
Did you have any difficulty getting tested for Lynch syndrome?*

The findings revealed that:

- approximately half the sample (47%) came to be tested for Lynch syndrome as a result of investigations into family history
- for more than 2 in 5 (44%) testing only came after their own diagnosis with cancer.

Our survey reflected the reluctance of some people to pursue genetic testing or share their test results,

"It took 18 months for the experts to decide whether to test my blood samples...this resulted in my brother and sister having been diagnosed with bowel and endometrial cancer... within months of each other"

"I have not been tested but my son and daughter both have the gene"

"3 of my cousins who are now undergoing cancer treatment chose not to be tested"

and highlighted the way this, in turn, was believed by patients to affect the ability of other relatives to determine their own cancer risk, even though this is not correct.

"One of my sister's...refuses to get the easy blood test...the kids are unaware this genetic mutation is even in the family"

Recommendations

- 1: Implement a nationwide system of universal tumour testing to test all colorectal and endometrial cancers for deficient DNA mismatch repair at the time of biopsy/surgery.
- 2: Establish referral pathways between hospitals and genetics services that ensure all patients identified as potential Lynch syndrome carriers are referred to Familial Cancer Clinic for follow-up; and that address right to know of other family members who may be at risk. And support existing and future studies to improve behaviours and compliance
 - 2.1: Expert health consumers, including a representative from LSA, should be included in processes for developing relevant pathways, protocols or clinical guidelines.

1.2 Family cancer history

Current guidelines advise sending a patient for genetic counselling and testing if they have three or more Lynch syndrome related cancers diagnosed in the family over two or more generations, with at least one of these occurring under the age of 50 yrs^{13, 14}. Anyone who has had a Lynch syndrome-related cancer under the age of 50 should also be sent for genetic counselling and testing.

However, family cancer history can be difficult to determine. Sometimes it is not possible to find out which cancers family members have had; relatives can be reluctant to discuss their personal medical history and death certificates usually record only the immediate cause of death. It is best practice for GPs to take a family health history for each patient with attention given to heart health, diabetes and cancer history¹⁵.

If universally implemented, taking a family cancer history would contribute significantly to identifying people with Lynch syndrome before a cancer diagnosis and subsequently improve their quality of life through prevention or early diagnosis. However, research has shown that 'not only do healthcare professionals rarely ask about a family history of cancer, they struggle with recognising Lynch syndrome and referring cases to clinical genetics¹⁶. In particular, many medical practitioners lack adequate knowledge of the various tumour types that are associated with Lynch syndrome and there is little awareness, in general, that so many seemingly unrelated cancers can be connected by one faulty gene¹⁷.

If universally implemented, taking a family cancer history would contribute significantly to identifying people with Lynch syndrome before a cancer diagnosis



The consumers' experience

Our survey asked questions about the family cancer history and genetic testing history of the respondent and various first and second-degree relatives. As well as the opportunity to provide free text responses, survey participants were also asked:

*How did you first come to know about Lynch syndrome?
Have members of your family been tested for Lynch syndrome?*

A number of concerns identified above were reflected strongly in our survey findings. They confirmed the value of a detailed family cancer history in indicating Lynch syndrome,

"Genetic testing was not around when my great grandfather, grandmother, mother and her three brothers were diagnosed with bowel cancer, and died within 3 years of the diagnosis"

"There is a strong history of colon cancer in my mother's family, and when she died of cancer in 1963 familial cancer was unknown. It wasn't till after my second cancer in 1995 that I was referred to genetic counselling."

"At least 4 previous generations had lynch tumours but I was the first to be tested."

The difficulty in constructing a family history for some families was also highlighted,

"Combination of family not interested and very difficult obtaining deceased relation's genetic test results even though next of kin gave permission. Large testing companies did not want to part with their information."

"Even though in my family, we had [had] over 20 cases of cancer... a tumour sample was needed before testing could be done." [This practice has been updated in recent months]

"I could not obtain genetic test results from deceased relations."

"[Had] trouble accessing my grandfather's family history as they live overseas and could not be contacted."

Importantly, the findings indicated that many General Practitioners (GPs) were failing to take or use a family cancer history to identify those individuals who may be at risk.

- Less than half (49%) the survey respondents were diagnosed based on family history
- 43% were diagnosed after a cancer diagnosis and 8% based on their own research etc.
- The majority (56%) of survey respondents were only tested for Lynch syndrome *after* they had received a cancer diagnosis

"My doctor told me that there is no such as inherited cancer of the colon"

"GP was useless [even though] I had more information based on previous family testing. Difficult to get a referral because GP had to google the syndrome. Then couldn't find a specialist. Terrible!"

Recommendation

3: Educate GPs and all health professionals on the importance of taking a comprehensive family cancer history and its role in identifying hereditary cancer predispositions such as Lynch syndrome

3.1: GPs and all health professionals must have access to clear and up to date information about Lynch syndrome, its risks and management protocols.

1.3 Genetic counselling and testing

The genetic sequencing (testing) process for Lynch syndrome is arranged by a genetic counsellor through a Family Cancer Clinic. Some people access genetic testing services directly, usually from an overseas provider, although this approach is not recommended for people with Lynch syndrome for two reasons.

- First, the National Health and Medical Research Council (NHMRC) reports that direct-to-consumer genetic testing companies have sometimes been found to return different interpretations of the same DNA sample, due to different analytical or post-analytical approaches.
- Second, regardless whether a patient is identified from a tumour sample, cancer family history or the diagnosis of another family member, s/he should always be offered timely and appropriate genetic counselling before and after the genetic testing process¹⁸.

The initial genetic testing (called a mutation search) must be done for someone in the family who has developed a Lynch syndrome cancer. The test (usually through a blood test) usually takes 6 - 8 weeks¹⁹. Once a genetic mutation is identified, other family members who are at risk of carrying the mutation can then be tested. Those tests are called predictive tests (either through a blood test or saliva sample). It is important to note that not all pathology laboratories provide these services.

There is no Medical Benefit Scheme Item number allocated for genetic testing for Lynch syndrome. However, if a patient is referred for testing by a Family Cancer Clinic, the cost of the testing can be covered through the public health system and there is no cost to the patient.

Genetic testing may also be initiated by private genetic services. In this instance, there will be a cost to the patient, however these costs may vary greatly. The testing costs are not covered by Medicare or Private Health Insurance. The cost for the appointment with the geneticist may be covered by Medicare.



The consumers' experience

Our survey inquired into the experience that individuals had with genetic counselling and testing. Questions included:

What is your Lynch status? Diagnosed clinically, genetically after counselling, or genetically after testing only?

Did you have problems getting tested for Lynch syndrome?

Did you have problems accessing genetic testing?

A surprising number of respondents reported significant barriers in getting the initial referral to a genetic counselling service, particularly from their GPs – many of whom were reportedly unaware of Lynch syndrome. Others cited long waiting periods for an appointment at Family Cancer Clinic²⁰. Free text responses illustrate these challenges,

"My GP had no idea what Lynch syndrome was and was very reluctant to send me for testing"

"GP not willing at first to refer for testing until I persisted."

"My doctor was unsure of the process involved (in organising testing)"

"There is no geneticist in Darwin. The NT government stopped the funding for a visiting genetics service some years ago. Until then I had great care from the SA Genetics with [Dr X]. After that Queensland assumed care but didn't visit and for some time didn't accept referrals. Now we need to make an appointment and travel to Queensland to access genetic care and counselling."

"[H]aving to educate medical practitioners as to what Lynch Syndrome is & delays getting into a cancer clinic for counselling & testing, then such a long wait for results. All very stressful & emotional."

"There are long waiting times for counselling and result appointments"

"Have been waiting for over 12 months to have genetic testing in public system"

"No difficulty getting the testing done through genetic health Queensland, but it took 5 years for diagnosis!"

The consequences of a diagnosis of Lynch syndrome are complex, lifelong and affect not only the individual but for the whole family. Impacts are felt physically, psychologically, financially and socially. Together this highlights the importance of access to appropriate counselling services.

"I felt like something would eventually happen as my mother and her 3 siblings have all had cancer diagnosed in their 40s."

"It is not possible to adequately describe in words the stressful journey my siblings and myself have been on since we all 3/3 have been diagnosed"

"Sometimes it feels like a ticking time bomb, but on the other hand knowing that you are having regular check-ups gives you piece of mind"

The results revealed that once people are identified for further investigation, appropriate genetic counselling is generally provided before genetic testing.

- 77% of survey respondents had genetic counselling then genetic testing
- 10% received genetic testing without counselling

Despite this, one in 10 survey respondents indicated they did not receive counselling and support.

"I wasn't offered counselling before testing. I was referred by my GP straight to a geneticist. I have since been to a counsellor and in hindsight I would've preferred going public and seeing a counsellor only. The geneticist was a huge expense and the geneticist did not provide adequate or comprehensive information."

"I have never had any counselling or really any offer of counselling".

However, those who did receive counselling spoke positively of the experience. For example,

"I have found the [genetic counsellor's] support and information provided invaluable. I don't know where I would be without this"

"My family have a very young onset age. Finding appropriate information and support for my teenagers has been difficult and I am very grateful for Dr X who recently met with my boys to explain and discuss the situation and implications with them."

Recommendation

- 4: All genetic testing arising from a cancer family history or the diagnosis of another family member must be accompanied by genetic counselling before, during and after the testing process.
If the test results arise from a tumour sample and have an immediate impact on decisions about cancer treatment decisions, genetic counselling should be provided at the earliest opportunity.
- 4.1: Health professionals need to recognise the psychological impact of a Lynch syndrome diagnosis and consider referral to appropriate support services.



1.4 Genes Affected

Once diagnosed with Lynch syndrome, it is important to know exactly which MMR gene (MLH1, MSH2, MSH6 or PMS2)²¹ is affected because mutations in different genes are associated with very different risk profiles; both in terms of the types of cancers and their occurrence²².

The Iscarisk calculator provides insight into the effect of various combinations of risk factors on cancer risk for people with Lynch syndrome (www.iscarisk.com). For example, Table 1 summarises the cumulative lifetime risk (to age 70) of a first cancer for a person now aged 30 years²³.

Table 1: Cumulative Cancer Risk for a 30 year old to age 70

Cumulative cancer risk for a 30 year-old – to age 70	MLH1		MSH2		MSH6		PMS2*	
	Male	Female	Male	Female	Male	Female	Male	Female
Any cancer type	58%	80%	71%	74%	31%	71%	0%	24%
Colorectal cancer	45%	45%	37%	31%	14%	26%	0%	0%
Ovarian cancer		12%		15%		0%		0%
Endometrial cancer		34%		51%		49%		24%
Urinary tract/bladder/ kidney cancer	2%	2%	19%	19%	9%	9%	0%	0%
Gastric/small intestine, biliary tract, pancreas	17%	17%	6%	6%	2%	2%	0%	0%
* Warning: the results for PMS2 may be unreliable due to the limited sample size								

Although mutations in some Lynch syndrome genes carry a high risk of developing certain cancers where others have little or no elevated risk²⁴, many risk-management decisions, relating to life-changing prophylactic surgery or screening approach and frequency for example, are typically made without either the patient or the practitioner knowing the exact mutation or its associated risk profile. For example, current guidelines recommend hysterectomy is considered at age 40 or after childbearing is complete irrespective of gene fault.

The consumers' experience

Our survey asked:

What is your exact mutation?

Would you consider or have you considered having preventative surgery (such as total hysterectomy, bilateral oophorectomy or colectomy?)

Our survey revealed that many patients are missing vital information about which gene is faulty.

- Even when diagnosed, almost 30% of respondents weren't sure of their exact mutation (compared to only 10% of overseas respondents). This knowledge has direct implications for life-changing decisions.
- 49% of Australian women who had undergone preventive surgery to remove their ovaries and fallopian tubes carried a mutation that recent research suggests posed no elevated risk of ovarian cancer (i.e. either MSH6 or PMS2 mutations). Note: the survey did not ask when the women had the surgery.

Respondents commented

"The advice I have found to be most lacking is information on each specific gene mutation"

Recommendations

5: Genetic counsellors must inform their patients and family about their specific Lynch syndrome gene mutation and understand the relative risks associated with it.

5.1: Individuals and health professionals must have access to the latest research on the risks associated with each of the Lynch syndrome genes.

5.2: GPs must include a diagnosis of Lynch syndrome (including stating the specific gene involved) in all referrals to all other health professionals.



Misunderstood

The most important goal for people with Lynch syndrome is to manage their cancer risk so as to prevent cancer developing, or to diagnose cancer at the earliest possible stage so treatment is less invasive and its success is maximised. To do this, people need to know how best to manage their cancer risks.

Currently eviQ²⁵, an online service of the Cancer Institute NSW, provides treatment protocols for health professionals at the point of care. The goals of the Cancer Institute NSW are to reduce cancer incidence, increase survival rates, improve quality of life for cancer patients and those who care for them; and be an expert information source on cancer control for all.

Cancer prevention is a priority for those at high risk. Currently, the eviQ protocols for health practitioners in Australia recommend:

- Registration on a Familial Cancer Registry
- Risk reducing behaviour such as ensuring a healthy lifestyle
- A mixture of surveillance, risk-reducing surgery and medication to manage the risks of cancer for those with Lynch syndrome.

2.1 Registration on a hereditary cancer registry

According to the eviQ *Risk Management for Lynch Syndrome* protocols, all patients should be registered on a hereditary cancer registry to receive regular surveillance reminders, although notably the reminder systems for surveillance and follow up differ from state to state. It is important to note that State hereditary cancer registries are different to the database of individuals diagnosed with Lynch Syndrome held by individual Family Cancer Clinics.

The current situation

After our survey, LSA asked State hereditary cancer registries and Family Cancer Clinics for data about the number of people with a confirmed diagnosis of Lynch syndrome on the cancer registries, and how many people had been offered and had received genetic counselling and testing.

Each Australian state operates a different database to manage information about people with Lynch syndrome, although even when the same systems are used, the values and the data captured differ from state to state.

In New South Wales, those diagnosed with Lynch syndrome by a (public) Family Cancer Clinic are sent an invitation by post to join the NSW Hereditary Cancer Registry. If they accept this invitation, their details are registered on the NSW Hereditary Cancer Registry, a database managed by the Cancer Institute of NSW. Services provided by the NSW Hereditary Cancer Registry include receiving screening reminders, having access to information, assistance in contacting family members and access to relevant support groups. Similar databases exist in Queensland and South Australia but they are not formal hereditary cancer databases. In Western Australia, all positive results for Lynch syndrome are recorded by the Western Australia Familial Cancer Registry.

As at January 2017, the NSW Hereditary Cancer Registry includes only 1,180 people with Lynch syndrome. 75% of these have been genetically diagnosed. In January 2017, there were 124 people registered with Lynch syndrome in Western Australia. Since the conservative estimate of people with Lynch syndrome in Australia is around 85,000 and only 5% of these are understood to be diagnosed, it follows that there should be approximately 4,250 diagnosed with Lynch syndrome (assuming Australian adult population of 18.5 million).

In total, the NSW and WA (no data available from Queensland) registrants amount to just 1,304 individuals; meaning just over 1.5% of all Australians with Lynch syndrome are receiving screening reminders, update information and assistance from an appropriately resourced hereditary cancer registry.

Recommendation

6: Establish a National Register for Lynch syndrome that

- 6.1:** Contact all existing patients with Lynch syndrome and encourage them to join the register and automatically add all newly diagnosed patients to the registry (able to opt out, rather than opt in).
- 6.2:** Has a strong governance framework to ensure effective control of the registry data; ensuring strict privacy of participants, confidentiality of the information provided by individuals participating in the register and strict accountability of those managing the data for any breaches of these requirements.
- 6.3:** Provides timely screening reminders and follow up to participants
- 6.4:** Allows researchers to apply for access to de-identified data from the registry, subject to Human Ethics Approval
- 6.5:** Includes representatives from LSA in the Governance structure of the registry.

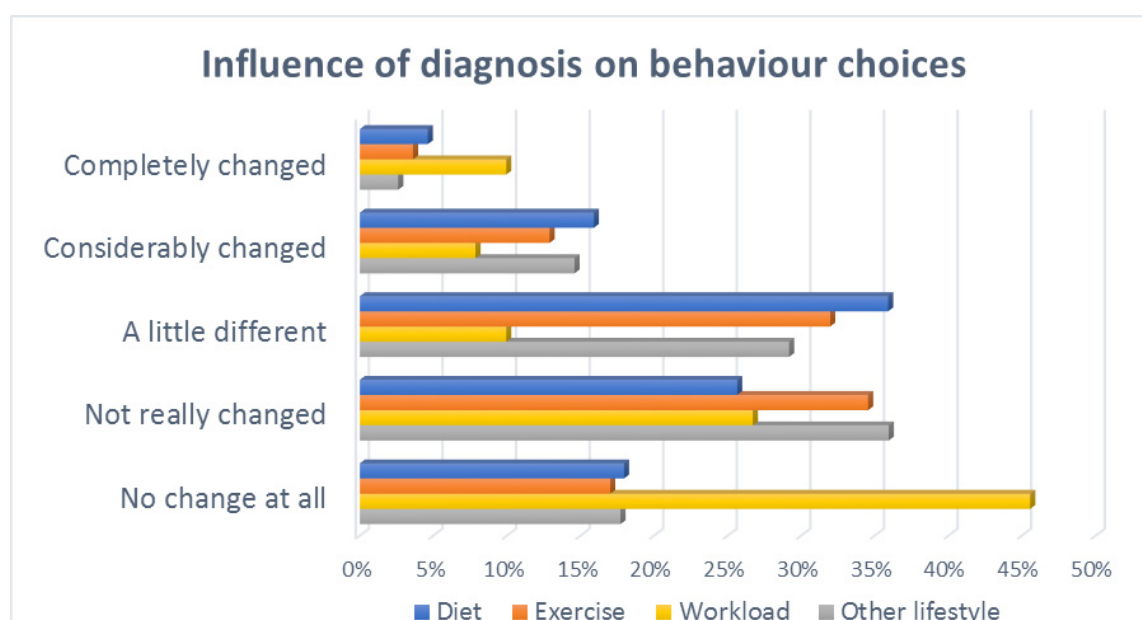


2.2 Risk-reducing lifestyle behaviours

The eviQ protocols recommend that health practitioners discuss with Lynch syndrome-affected individuals how lifestyle choices can affect their cancer risks. Recommendations include regular exercise, maintaining a healthy weight, eating a healthy diet, limiting alcohol, not to smoke and for women to breast-feed where possible.

This reinforces the long-standing recommendations of Cancer Australia for all Australians on how to reduce their risks of cancer. These recommendations are reinforced locally through eviQ protocols and internationally for people with Lynch syndrome by the International Society for Gastrointestinal Hereditary Tumours (InSIGHT).

Table 2: Influence of diagnosis on behavioural choices (Living with Lynch syndrome survey)



The consumers' experience

Our survey sought to identify whether a Lynch syndrome diagnosis motivated people to modify their behaviour by adopting healthier lifestyles, asking,

Since knowing about Lynch syndrome in your family, have you changed your diet; exercise regime; occupation/workload; other lifestyle factors?

Survey data revealed that many people made only modest changes to their diet, exercise and lifestyle following diagnosis.

- A mere 21% of respondents had substantially or completely changed their **diet**, while 44% had made virtually no change.
- Fewer than 16% had changed their **exercise activity** substantially or completely while little more than half (52%) had made no change.
- Curiously, the risk factor most likely to change completely was also the factor most likely not to change at all; work life balance. While 10% of respondents reported having completely changed their workload/occupation, compared to 5% who had completely changed diet, 4% exercise and 3% other lifestyle factors, 46% had made no change to their **workload/occupation** yet less than 20% had made no change to diet, exercise and other lifestyle factors.

Why a Lynch syndrome diagnosis has had such modest influence on lifestyle factors is likely to be an issue worthy of future research. However, these findings underscore the need to better communicate the role of lifestyle factors in reducing cancer risk and why it is particularly important for people with Lynch syndrome.

Recommendation

- 7: Design and distribute information, help and support appropriate to individuals with Lynch syndrome to explain the benefits of specific lifestyle and behaviour choices and promote risk reducing behaviours as part of an overall cancer management plan.

2.3 Surveillance and prophylactic surgery

The International Society for Gastrointestinal Hereditary Tumours (InSiGHT) has now identified 12 tumour types related to Lynch syndrome. While there appears to be broad consensus, globally, about diagnosis, cancer treatment and care of those with Lynch syndrome, current guidelines for managing the cancer risk differ markedly between countries^{26,27,28}. Further, guidance provided by Australian Family Cancer Clinics has evolved over time.

In Australia, current cancer risk management protocols are established by eviQ; a 'point of care clinical information resource that provides health professionals with current evidence-based, peer-reviewed, best practice cancer treatment protocols and information'. The development of content for eviQ follows a strict data governance process with protocols based upon the best and most comprehensive scientific evidence available at the time and follows the NHMRC hierarchy of levels of evidence.

eviQ protocols for the management of Lynch syndrome and its cancer risks are general guidelines developed for individuals who have NOT (yet) been diagnosed with a relevant cancer/tumour. They also recommend that care and monitoring activity should be individualised based on a patient's clinical situation and family history. The recommendations are summarised in Table 3 (following page).

Meanwhile, overseas experts like the Mallorca Group use a broader level of evidence in developing the Revised guidelines for the clinical management of Lynch syndrome (HNPCC).

These guidelines were developed at a workshop by a group of 21 experts from nine European countries and includes additional tailored surveillance recommendations. It is important to note that the group stated that there are still many aspects of Lynch syndrome about which new knowledge needs to be gained through further research.

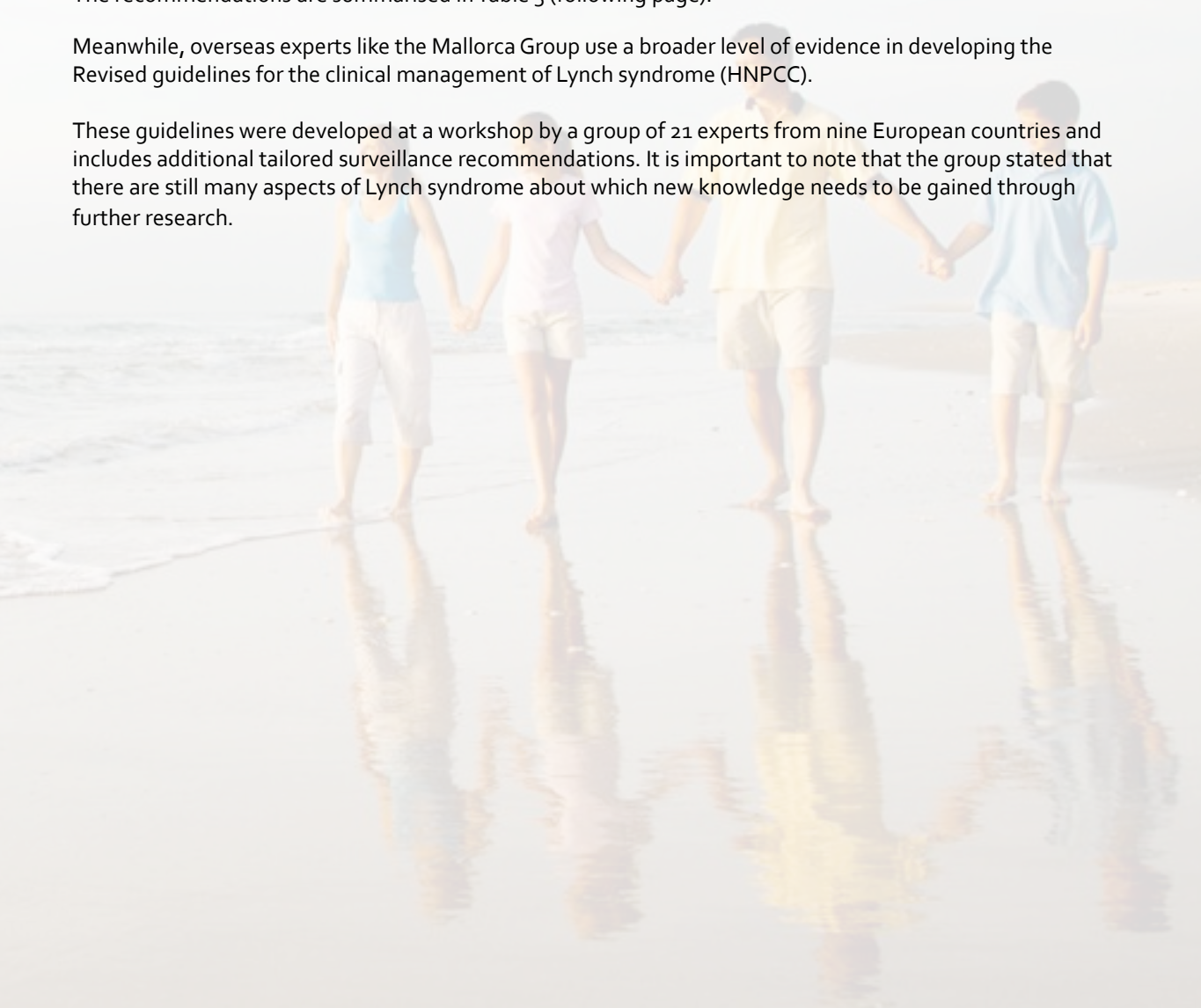


Table 3: eviQ protocols for the care of individuals with Lynch syndrome

Cancer Type	Australian recommendations for the care of individuals with Lynch syndrome	
Colorectal	Surgical	Consider subtotal colectomy in selected individuals
	Surveillance MSH6/PMS2	Annual colonoscopy from age 30 or 5 years younger than youngest affected if under 35 Review frequency at age 60
	Surveillance MLH1/MSH2	Annual colonoscopy from age 25 or 5 years younger than youngest affected if under 35 Review frequency at age 60
	Risk-reducing medication	There may be a reduction of risk taking aspirin however appropriate dose is not yet defined
Endometrial	Surgical	Recommended hysterectomy after childbearing complete or from age 40 or 5 years younger than the youngest affected, whichever comes first
	Surveillance	There is no evidence for a transvaginal ultrasound and/or aspiration biopsy
Ovarian	Surgical	Recommended risk-reducing salpingo-oophorectomy (RRSO) at time of hysterectomy Recommend HRT at time of RRSO and continue until usual time of menopause
	Surveillance	Do not offer serum CA125 and/or transvaginal ultrasound (TVU)
Gastric	Surveillance	Consider second-yearly gastroscopy from age 30 in families with gastric cancer or those at high ethnic risk
Urothelial	Surveillance	No evidence of benefit but patients encouraged to report symptoms

The consumers' experience: surveillance

Our survey asked participants about three aspects of surveillance: what surveillance they engaged in, who manages their surveillance and what out of pocket costs did they incur (if any)?

A surveillance regime is the testing you may receive regularly to look for signs of early cancer...Please tick one box per row to indicate the frequency of your surveillance regime and also tick the relevant family history box where an activity is prompted by past cancer experience in your own family.

Who manages your surveillance?

Do you pay out of pocket costs for your surveillance?

If yes, please estimate your annual out of pocket expenses?

What were your out-of-pocket expenses for?

'My genetic counsellor just wanted to speak about national statistical averages and completely ignored my family history'.

The most common cancer risks in Lynch syndrome, regardless of the gene involved are colorectal cancer for both men and women and endometrial cancer for women. LSA's survey, however showed that endometrial cancer was the most commonly occurring first cancer for women with Lynch syndrome.

In 2015, several survey respondents reported families who had experienced eight different Lynch syndrome related cancers. The current Australian guidelines (above) only recommend regular surveillance for one cancer risk for all adults with Lynch syndrome – colorectal cancer. No surveillance is recommended for the high risk of any other cancer type (with the exception of gastric cancer but only where there is a strong family cancer history).

Yet survey findings suggest risk management practices differ from one healthcare practitioner to the next. This may reflect their awareness of current research and risk management recommendations.

Our survey showed:

- Significant differences in the extent and frequency of surveillance that people received. Survey respondents reported various barriers which may have impacted on undertaking regular surveillance (see page 30).
- People with Lynch syndrome are receiving a much wider range of surveillance than that recommended to health practitioners by eviQ. Table 4 (following page) compares the experience of surveillance against the recommended protocols.

Table 4: Compares the experience of surveillance against the recommended protocols

	Current guidelines/protocols	Considerations	Experience of survey respondents
General	Guidelines assume that health practitioners, not patients, are managing their surveillance.		Survey participants were generally well-informed and motivated people managing their own surveillance and usually the first to be diagnosed in their family. 43% managed their own surveillance. 36% were the first to be diagnosed in their family.
Colorectal	Annual colonoscopy over age 25 or 30.	The average time for a colorectal cancer to develop is 10 years. The average time for a Lynch syndrome colorectal cancer is 30 months.	100% of respondents 25 years and under received annual or two yearly colonoscopy surveillance. One respondent had been diagnosed with bowel cancer and pancreatic cancer under 25 years of age. 6% of those who had reported cancer, had their first cancer diagnosis under 25 years of age. 20% of the 231 question respondents over age 25 years were <i>not</i> receiving annual colonoscopy surveillance.
Endometrial and ovarian	eviQ protocols recommend considering preventive surgery hysterectomy and RRSO for women and subtotal colectomy for selected individuals after age 40 years.	Research studies have shown that risk reducing surgery, also known as prophylactic or preventive surgery, has a significant positive effect on survival rates of people with Lynch syndrome. However, quality of life is also reported to be significantly affected by partial or subtotal colectomy (removal of the colon/rectum) and by hysterectomy and removal of ovaries and fallopian tubes.	86% of all respondents had either had or considered having preventive surgery. 62% of the female respondents had undergone risk-reducing surgery, mainly the removal of uterus, cervix, ovaries and fallopian tubes.
General risk	eviQ guidelines and evidence make no reference to this.	A carcinoembryonic antigen test (CEA) measures the amount of protein that may appear in the blood of some people who have certain kinds of cancers, especially colon and rectal cancer and also may be present in people with cancer of the pancreas, breast, ovary or lung.	52% of respondents to this question (175 people) received regular CEA blood testing.

	Current guidelines	Considerations	Experience of survey respondents
Breast	There are no protocols for the management of breast cancer within Lynch syndrome at this time.	<p>After recent research, leading international experts have recently added breast cancer to the list of tumour types considered related to Lynch syndrome. Further studies are underway.</p> <p>Screening for breast cancer is highly effective and breast cancer survival rates have risen from 70% in 1987 to 90% in 2010. Of the 179 women who responded to the survey, 109 reported a history of breast cancer as 6% of 1st and 2nd cancers and 20% of 3rd cancers.</p>	174 out of 214 Australian women responded to the question about surveillance and 59% receive regular breast screening, 38% annually, although some may be receiving population screening due to their age.
Urothelial	eviQ states that there is 'no evidence for the role of urine cytology in the surveillance of urothelial carcinoma in MMR gene carriers and consequently eviQ has no management recommendations'.		74% of those who answered this question underwent periodic urine cytology.
Prostate	No protocols are provided.	Prostate cancer is considered by InSiGHT as a Lynch syndrome-related cancer although the value of the PSA test is much debated.	Over 50% of male respondents received PSA testing every year, although some may be receiving population screening due to their age.
Skin	eviQ makes no mention of skin cancer either in its 'Cancer risk management and evidence' or in its 'Information for people with Lynch syndrome'.	Skin cancer is widely recognised as a Lynch syndrome-related cancer in those families with Muir Torre.	72% of 179 question respondents received skin checks 53% every 3 years or more frequently. 20 cases of skin cancer or pre-cancer were reported by 140 respondents who reported cancer/s.
Ovarian	eviQ guidelines do not distinguish between mutations when advising preventive surgery eviQ expressly advises against offering transvaginal ultrasound.	Ovarian cancer risks differ markedly depending on the exact gene mutation.	Of 170 respondents to this question, 26% receive pelvic ultrasound screening, which includes transvaginal ultrasound.

Importantly, the survey explored the role of GPs, genetics services, specialists and cancer organisation in providing information to support diagnosis and treatment. Survey participants reported that health professionals had little or no knowledge of Lynch syndrome and provided them with little or minimal information on the syndrome or on ways to reduce their cancer risk.

- 41% of respondents reported that talking to their GP was not useful or only somewhat useful.
- However, 83% of respondents reported that information on Lynch syndrome from the Lynch Syndrome Australia website was somewhat to very useful.

Our survey discovered multiple potential barriers to surveillance

The ability to pay for surveillance

- 73% had paid out of pocket expenses for their surveillance, mostly for hospital/doctor's fees
- 52% paid between \$500 - \$5000
- 20% paid between \$2000 - \$5000

Who managed their surveillance

- Almost half had to manage their own surveillance (understanding their risks, requesting appointments, making sure they were being screened regularly etc.)
- Only 14% had the help of an oncologist
- Only 1% had the support of a specialist nurse

Recommendations

8: Develop an improved, agreed National Clinical Care Standard for the management of Lynch syndrome.

8.1: The Lynch syndrome Clinical Care Standard must be: person centred; informed by evidence and current best practice; widely accepted and understood by health practitioners and individuals with Lynch syndrome. This means it must articulate the need to explicitly consider the specific gene mutated and the personal and family cancer history.

9: Ensure everyone with Lynch syndrome has timely and affordable access to the surveillance regimes in the Clinical Care Standard, regardless of where they live (including rural and remote consumers).

10: Adherence to surveillance regimes in the Clinical Care Standard is monitored by the national database and the care coordinator.

The consumers' experience: prophylactic surgery

Risk reducing surgery recommended by the eviQ protocols are as follows:

- A hysterectomy after childbearing complete or from age 40 or 5 years younger than the youngest affected family member, whichever comes first.
- Consider subtotal colectomy in selected individuals

Both of these major surgeries have life changing ramifications. Informed decisions should be made together with relevant health professionals and taking into consideration the risks associated with the specific gene mutation the person has, the risks of surgery and the impact of surgery on the person's quality of life.

Our survey asked:

Would you consider, or have you considered, having preventative surgery?

The survey findings revealed:

- 51% of individuals have considered or are considering having preventative surgery
- 63% of individuals have already had preventative surgery
- 42% of women have undergone a hysterectomy, 39% of those as a preventative measure
- 53% of hysterectomies included removal of ovaries.

Recommendation

- 11: Develop a Risk Reducing Surgery decision aid taking into consideration the different risks associated with each of the four mismatch repair genes for use by health professionals and consumers.



3

Missing Out

Broader issues for families with Lynch syndrome

The Australian Charter of Healthcare Rights states that patients have a right to access services to address their healthcare needs and the right to receive safe and high quality health services with professional care, skill and competency²⁹.

People with a lifelong, continuous and high risk of disease, like those with Lynch syndrome, need timely and affordable access to expert advice, treatment and care.

3.1 Affordable health care

Australia has one of the best health systems in the world. Eligible Australians have access to a universal health system through Medicare. Medicare provides free care as a public patient in a public hospital. Medicare also provides patients with a rebate for visits to doctors outside the hospital setting such as general practitioners and specialists. The Pharmaceutical Benefits System (PBS) provides subsidised medications to eligible Australians.

Many Australians also contribute to private health insurance which partially covers the cost of care in a private hospital. Just under half of Australians now have private health insurance for hospital treatment and just over half have 'extras' cover³⁰.

The need for regular, lifelong surveillance and potentially numerous cancer treatments, for not one but multiple family members, underscores the financial pressures that Lynch syndrome can impose on households. Access to affordable health care that is timely and of high quality is therefore particularly crucial for the survival of families with Lynch syndrome.

The consumers' experience

Our survey asked participants about their access to affordable health care and, where relevant, the financial impact of Lynch syndrome and the extent to which cost presents a barrier to routine surveillance or treatment. Questions included:

Do you pay out of pocket expenses for your routine surveillance?

Did you pay out of pocket expenses for your cancer treatment?

If yes, are you able to provide an estimate of costs?

The findings revealed that people with Lynch syndrome are paying significant gap fees for routine surveillance and cancer treatment; much of this resulting from concern regarding the expertise of local clinicians and efforts to secure access to specialists with known expertise in treating Lynch syndrome cancers. This often involves travelling considerable distances from home. In summary:

Of the Australians surveyed,

- 31% had treatment in a public hospital, 45% in a private hospital and 23% drew on a mix of public and private hospitals for their cancer treatment
- 77% had health insurance that included hospital cover at the time of their cancer treatment
- 81% had paid out of pocket expenses relating to their cancer treatment
- Costs were split fairly evenly between hospital, doctors', pathology fees and medications, with doctors' fees the most prevalent, closely followed by hospital fees (excess)
- Only 12% of respondents had received cancer treatment solely as a public patient in the public health system. Yet almost half (45%) still reported out of pocket costs of up to \$500 for pathology services, doctors' fees and medication
- Those treated as private patients in the public health system reported higher average costs of \$1,000-5,000, although two people paid over \$10,000 in out-of-pocket expenses. Gap payments for private patients in private hospitals also averaged \$1,000-5,000 although a greater proportion (29%) paid costs in excess of \$10,000 in cancer treatment gap fees

Importantly, qualitative responses reflected the significant impact that travel and gap fees had on many families. For example,

"I had my surgery in Melbourne but live in Northern NSW. I chose {travelling to} Melbourne [over Sydney] as we had family support there to help look after my children and we also had medical contacts for surgeons etc. Costs of airfares was high for 4 people back and forwards to Melbourne, particularly over the Dec/Jan holiday period for my pre-op tests and operations."

"I live in Darwin and to obtain the best treatment I travelled to Sydney to see specialists and have a PET scan (not available in Darwin at that time). I then had surgery in Sydney. My husband came to Sydney with me and we paid all our own travel and accommodation costs... In addition to all the above usual expenses I have a 6 year- old and 2 year - old that I had to pay for full-time care for while we were away."

"I had my surgery in Melbourne but live in Northern NT...we had medical contacts for surgeons"

"There was also travel costs from Whyalla to Adelaide to test a different chemotherapy as the side effects of the oral medication were debilitating"

"Queuing in a Medicare office about 10 days out of hospital, when I could barely stand, so I could get some money back into a bank account...I had emptied"

"I travelled from rural NSW to Sydney for cancer treatment. I had to sell my car to pay the hospital in-patient account"

"I won't get tested due to concern a positive result will impact future medical insurance or care"

Recommendation

- 12:** Development of Lynch Syndrome Centres of Excellence with access for all individuals with Lynch syndrome. Availability of flexible funding to assist individuals to access the right health professional at the right time in the right place.

3.2 Coordinated care

Managing the complex, lifelong risks associated with Lynch syndrome, making well-informed, timely and appropriate lifestyle choices and dealing with the impact on the whole family is not something that anyone should be expected to navigate on their own.

After diagnosis, patients must regularly meet with and rely upon their GP for help, support and advice about managing their cancer risk, cancer treatment and for timely and appropriate access to specialist care and lifestyle and wellbeing advice. Those with Lynch syndrome therefore need access to a knowledgeable and accountable primary care-practitioner.

A GP “plays a central role in the delivery of healthcare to the Australian community, is the most likely first point of contact and coordinates the care of patients. A GP also refer patients to other specialists and care for the whole person in the context of their work, family and community and provides advice and education on healthcare” (Royal Australian College of General Practitioners, 2017).

The RACGP’s current awareness campaign positions general practitioners as “your specialist in life”, emphasising their expertise and specialist skills. GPs are tasked with investigating a patient’s health history, helping to manage chronic conditions and support patient education and efforts to prevent disease, and therefore should be well placed to coordinate care for people with Lynch syndrome.

Furthermore, health care should be person-centred. The Australian Commission on Safety and Quality in Health Care define Person Centred Care as healthcare that is respectful and responsive to, the preferences, needs and values of patients and consumers. The widely-accepted dimensions of person-centred care are respect, emotional support, physical comfort, information and communication, continuity and transition, care coordination, involvement of family and carers and access to care³¹.



The consumers' experience

Our survey sought to explore the extent and quality of advice and support provided to people with Lynch syndrome by GPs and other health care providers. Questions included:

*Do you have a dedicated individual managing your surveillance? If so, who is this?
Have you ever delayed your surveillance or treatment?
What are your reason for delaying your surveillance or treatment?
What support or advice have you found to be most lacking?
Have you encountered any barriers to treatment?
How might your experience of diagnosis, surveillance and treatment be improved?*

The findings revealed a significant level of frustration with the level of support available from clinicians; GPs in particular. Typical of many responses were free text comments such as,

"I have yet to meet a GP that is aware of Lynch syndrome"

"I always have to explain what it is"

"I have found that all the GPs I have seen have no idea what Lynch syndrome is"

"When GP knowledge is less than patient knowledge this is a serious concern"

"GP didn't have a clue and sent me in circles, I ended up having to tell him how to do his job. . . and pay him for the effort!"

Unsurprisingly therefore, less than a third of respondents relied on their GP to coordinate their care.

- Only 29% of respondents' GPs managed or co-ordinated their surveillance/treatment
- 41% found talking to their GP about Lynch syndrome 'not useful'

When asked how their experience of diagnosis and treatment could be improved (more than one answer was possible)

- Over half said with a better-informed GP
- 43% more specific information from cancer organisations such as Cancer Australia and Cancer Councils
- 21% said a better-informed specialist
- 18% faster access to genetics services
- Information on recent advances e.g., Aspirin and lifestyle changes

Instead, an overwhelming 44% revealed they had to manage their own surveillance/treatment. In contrast, 29% were managed by GPs, 15% by oncologists and the remainder by various different specialists or family members. The choice of care coordinator appeared to have important implications for health care. For example,

- One third of respondents who managed their own care, had delayed surveillance or treatment.
- This was similar to the rate of delays for those coordinated by GPs but contrasted sharply against the low rate of delayed surveillance or cancer treatment for those whose care was managed by their oncologist or oncology nurse.

Access to an appropriate clinician is also important for facilitating seamless access to a range of important information and health services. Yet when asked about issues in accessing services, many of our survey respondents reported having faced challenges.

- 1 in 10 respondents had encountered barriers to treatment
- 25% of respondents had experienced problems accessing services
- 15% had problems accessing genetic testing
- 12% had problems getting to a diagnosis
- 17% had problems accessing treatment

Understanding survivorship

Further underscoring the challenges for lifelong care and support is the concept of survivorship. Cancer Australia says that 'an individual is considered a cancer survivor from the time of diagnosis, through the balance of his or her life. Family members, friends, and caregivers are also impacted by the survivorship experience³²'. However, survivorship means something very different for someone with Lynch syndrome than it does for someone facing a single cancer diagnosis. Individuals with Lynch syndrome face a continuous risk of multiple different primary cancers. Lifelong annual surveillance undermines the ability to say 'you are well, now get on with your life and forget about it'. Little is known about the short, medium and long-term psycho-social effects of living with Lynch syndrome on both an individual and their affected and non-affected family members.

In 2016, the Clinical Oncology Society of Australia (COSA) drew up a new model of survivorship care for all cancer patients, stating that, 'current models of care for cancer survivors are not survivor-centred, coordinated, or accessible. Nor do they effectively manage and minimise the burden of disease and treatment related side effects in the post-treatment phase'³³. Current models of survivorship appear to be failing traditional cancer patients and are far from helpful in supporting families with Lynch syndrome.

To that end, our survey also uncovered a number of challenges for people with Lynch syndrome. Concerns were most evident around:

- Problems obtaining life insurance and mortgage protection insurance
- Discrimination in gaining employment
- Problems accessing legitimate sick leave entitlements

These concerns were impacting some respondents' care choices. For example, refusal to be tested for Lynch syndrome, or delaying surveillance and treatment due to work commitments.

Recommendation

13: Individuals with Lynch syndrome have access to care coordinators help people manage their own care throughout their lives, assist with monitoring clinical management regimes and act as liaisons between patients and the health care system.

13.1: Health professionals involved in the care of individuals with Lynch syndrome should have a sound understanding of the diagnosis and lifelong management of Lynch syndrome.

3.3 Health literacy

Someone with Lynch syndrome faces a lifetime of managing their cancer risk of up to twelve tumour types and needs to understand their own particular health and specific needs whilst navigating a confusing system of health care and providers.

The Australian Commission on Quality and Safety in Healthcare defines health literacy as “the knowledge, motivation and competencies of a consumer to access, understand, appraise and apply health information to make effective decisions and take appropriate action for their health and health care.”³⁴

The health literacy environment is the world of infrastructure, policies, processes, materials and relationships that exist within the health system that make it easier or more difficult for consumers to navigate, understand and use health information and services to make effective decisions and take appropriate action about health and health care. Without these skills, it can be difficult for people to make the right choices when they are faced with important health care decisions³⁵.

Adequate health literacy levels are vitally important to support a patient’s involvement in the formal health care system, and the health decisions they make in the home, workplace and community. According to the Australian Bureau of Statistics, 60% of Australians do not have adequate health literacy skills.



The consumers' experience

We asked survey respondents to think about how they obtained information about Lynch syndrome and how useful they found each of the following: books; talking to my GP; talking to my genetic counsellor; talking to my oncologist; eviQ; Pubmed; web searches and forums; LSA website; Cancer Council websites; Cancer genetics websites; other.

The number of respondents indicating which websites were useful for sourcing information on Lynch syndrome suggests:

- 66% of respondents reported the LSA website was useful or very useful
- 38% found Cancer Council website(s) useful or very useful
- 28% reported eviQ online resources useful or very useful

Useful face-to-face sources of information on Lynch syndrome were:

- 74% talking to their genetic counsellor
 - 38% talking to their oncologist
 - 26% talking to their GP
- The survey did not include specific reference to their gastroenterologists

After our survey, we investigated the health literature on Lynch syndrome currently available in Australia and observed,

- Little consistency of terminology from cancer control organisations, including the continued use of outdated labels such as HNPCC.
- No consistency in defining or communicating the risks associated with Lynch syndrome.
- No literature to help someone with Lynch syndrome explain their unique diagnosis and prognosis to members of the family.

Recommendation

14: Health professionals consider the health literacy needs of individuals with Lynch syndrome.

- 14.1:** Individuals with Lynch syndrome should have access to comprehensive evidenced based information which can assist them during each phase of their journey from diagnosis to supporting self-management, surveillance and treatment as well as sharing this information with family members.
- 14.2:** Health professionals should encourage shared decision making with individuals with Lynch syndrome.
- 14.3:** Individuals with Lynch syndrome should have access to information targeted at all levels of health literacy.

4

Conclusion

These recommendations are just the start. Concerted commitment and collective action is needed from those organisations whose job it is to manage healthcare and long-term wellbeing for Australians. The recommendations will form the basis of Lynch Syndrome Australia's advocacy strategy for 2017/2018, starting with a series of meetings with those best placed to help change things for the better.

Naturally, our study had its limitations, including uncontrolled respondent recruitment. Self selection of course, could lead to ascertainment bias and we note, in particular, that the sample was skewed somewhat toward older female voices. Our conclusions should be read with this in mind. Nevertheless, a broad survey is considered appropriate methodology to test the environment and the collective experiences of families with Lynch syndrome and the relatively large number of participants lends strong support to the general conclusions that have been drawn. Such a survey, with its clinically informed design would generally be considered appropriate for a consumer organisation.

These findings should form the basis for greater and more targeted research, both qualitative and quantitative in nature, by well-funded stakeholders and cancer control organisations so that they might improve the experience of those living with Lynch syndrome and ensure no more Australians need be misdiagnosed, misunderstood and missing out.



5

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There are changes we all need to see; changes to help us meet these challenges, together.



Mailing Address

PO Box 292
The Summit, QLD, 4377

info@lynchsyndrome.org.au
Tel 0416 070 036



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Syndrome**
Australia

Many families. Many cancers.

One common cause.