



# Lynch syndrome -What is it?

Lynch Syndrome Australia



# Just bad luck?



# What is Lynch Syndrome?



Lynch syndrome (previously known as HNPCC) is an inherited genetic mutation which gives people an increased chance of developing certain cancers across their lifetime, often at a younger age than the general population (i.e. before 50 years of age).



# The numbers



4

Lynch syndrome is caused by a mutation in one of the body's mismatch repair (MMR) genes. These genes are:

MLH1, MSH2, MSH6, and PMS2.





- Colon\* and rectal cancer
- Endometrial cancer\*
- Small intestine cancer (MSH2 & MLH1)
- Hepato-biliary and pancreatic cancer (MSH2 & MLH1)
- Gastric cancer (MSH2 & MLH1)
- Ovarian non-serous cancer (MSH2 & MLH1)

*\*Most common cancers associated with Lynch syndrome.*

- Renal pelvis and ureter cancer (MSH2 & MSH6)
- Bladder cancer (MSH2 & MSH6)
- Sebaceous gland cancer (and adenoma – Muir-Torre syndrome)
- Prostate cancer (MSH2)
- Breast cancer (MLH1)
- Central nervous system cancer

# The numbers



1:280



# The numbers



# When would you suspect Lynch syndrome?





- Comprehensive Family History
- Tumour testing
- **Genetic Counselling**
- Mutation search
- Predictive testing



# Risk



## Lifetime risk of cancer

Cancer	MLH1 to age 70 yrs <sup>1, 2, 3</sup>	MSH2 to age 70 yrs <sup>1, 2, 3</sup>	MSH6 to age 70 yrs <sup>2, 4</sup>	PMS2 to age 70 yrs <sup>5</sup>	Lynch syndrome to age 70 yrs*	General population to age 70 yrs
Colorectal (male)	34%	47%	22%	20%	38%	3.1%**
Colorectal (female)	36%	37%	10%	15%	31%	2.2%**
Endometrial	18%	30%	26%	15%	33%	1.3%**
Gastric	6%	0.2%	Insufficient data	-	6%	0.38%**
Ovarian	11%	15%	Low	-	9%	0.57%**
Urothelial	0.2%	2.2%	0.7%	-	<3%	0.33%**
Small bowel	0.4%	1.1%	Insufficient data	-	<3%	0.12%#



# Risk



**PLSD**  
Prospective Lynch Syndrome Database

Prospective Lynch Syndrome Database (PLSD) - cumulative risk for cancer by age, genetic variant, and gender in carriers subject to colonoscopy

Any cancer

Carrier without previous cancer

Carrier with previous cancer

About

<http://lscarisk.org/>



# Management



Current management/surveillance of Lynch syndrome in Australia is based on the EviQ Guidelines



# EviQ guidelines




## Cancer risk management guidelines

Cancer type	Recommendations***	
Colorectal	<b>Surgical</b>	<ul style="list-style-type: none"><li>Consider subtotal colectomy in selected individuals.</li></ul>
	<b>Surveillance</b> MSH6	<ul style="list-style-type: none"><li>Colonoscopy every 1 to 2 years from age 25–30 years.<sup>6</sup> Annual surveillance is preferred in known mutation carriers.</li><li>Review frequency of colonoscopy at age 60 years with a view to reducing frequency.</li></ul>
	<b>Surveillance</b> PMS2	<ul style="list-style-type: none"><li>Colonoscopy every 1 to 2 years from age 35 years.</li><li>Review frequency of colonoscopy at age 60 years with a view to reducing frequency.</li></ul>
	<b>Surveillance</b> MLH1/MSH2	<ul style="list-style-type: none"><li>Colonoscopy every 1 to 2 years from age 25 years. Annual surveillance is preferred in known mutation carriers.</li><li>Review frequency of colonoscopy at age 60 years with a view to 2nd yearly frequency.</li></ul>
	<b>Risk-reducing medication</b>	<ul style="list-style-type: none"><li>Unless contraindicated, aspirin should be actively considered to reduce the risk of colorectal cancer. A low dose (100–300 mg per day) is recommended from the commencement of colonoscopy screening.</li></ul>



# EviQ guidelines



<b>Endometrial</b>	<b>Surgical</b> MLH1/MSH2/MSH6	<ul style="list-style-type: none"> <li>• Recommend hysterectomy after childbearing complete or from age 40 years.</li> </ul>
	<b>Surgical</b> PMS2	<ul style="list-style-type: none"> <li>• Recommend hysterectomy after childbearing complete or from age 50 years.</li> </ul>
	<b>Surveillance</b>	<ul style="list-style-type: none"> <li>• There is no evidence for transvaginal ultrasound (TVU) and/or aspiration biopsy.</li> </ul>
<b>Ovarian</b>	<b>Surgical</b>	<ul style="list-style-type: none"> <li>• Consider risk reducing salpingo-oophorectomy (RRSO) at time of hysterectomy in selected individuals.</li> <li>• Recommend HRT at the time of RRSO and continue until the usual time of menopause.</li> </ul>
	<b>Surveillance</b>	<ul style="list-style-type: none"> <li>• Do not offer serum CA125 and/or transvaginal ultrasound (TVU). See <a href="#">Cancer Australia for further information</a> </li> </ul>
<b>Gastric</b>	<b>Surveillance</b>	<ul style="list-style-type: none"> <li>• Consider 2nd yearly gastroscopy from age 30 years in families with gastric cancer or those at high ethnic risk - e.g. Chinese, Korean, Chilean and Japanese.</li> </ul>
<b>Urothelial</b>	<b>Surveillance</b>	<ul style="list-style-type: none"> <li>• No evidence of benefit but patients encouraged to report symptoms e.g. haematuria.</li> </ul>

\*\*\*The impact of lifestyle on cancer risk should be discussed e.g. exercise regularly, maintain healthy weight, have a healthy diet, limit alcohol intake, do not smoke and avoid excessive sun exposure.

<https://www.eviq.org.au/cancer-genetics/risk-management/1410-risk-management-for-lynch-syndrome>



# The Lynch syndrome patient with cancer



- Cancers occur younger than the population
- More than one cancer
- Treatment options



# The role of the Practice Nurse



- Take a comprehensive family history
- Regularly review the family history
- Coordinating surveillance
- Understanding where to find more information
- Emotional support



# Raising awareness



# Resources



- [Understanding genetic tests for Lynch syndrome-  
Information and decision aid](#)
- [Lynch Syndrome Australia](#)
- [NSW Cancer Institute](#)
- [Cancer Council NSW](#)
- [Cancer Australia](#)



# Lynch Syndrome Australia



- Living with Lynch syndrome workshops
- Peer to peer support group
- Comprehensive website



## Living with Lynch Conferences

Our Living with Lynch conferences are designed to provide current and relevant information and updates to individuals and families affected by Lynch syndrome, health professionals and related organisations.

In the past, these conferences have focused on bowel cancer surveillance, managing the additional risks in women, well-being, communicating with families and providing insights into international research efforts to provide greater hope for the future.

Details of past conferences can be found [found here](#).



# Lynch Syndrome Australia Report



Many families. Many cancers.  
One common cause.



Misdiagnosed, misunderstood  
and missing out:

Lynch Syndrome  
Australia's untold health story







**Many families. Many cancers. One common cause.**

[lynchsyndrome.org.au](http://lynchsyndrome.org.au)