

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

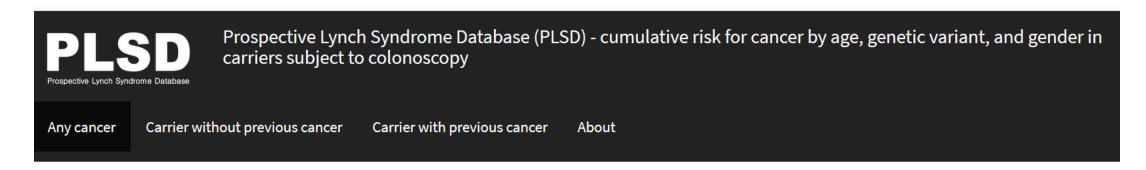


Lifetime risk of cancer

Cancer	MLH1 to age 70 yrs ^{1, 2, 3}	MSH2 to age 70 yrs ^{1, 2, 3}	MSH6 to age 70 yrs ^{2, 4}	PMS2 to age 70 yrs ⁵	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





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	Surgical	Consider subtotal colectomy in selected individuals.		
	Surveillance MSH6	 Colonoscopy every 1 to 2 years from age 25–30 years.⁶ Annual surveillance is preferred in known mutation carriers. Review frequency of colonoscopy at age 60 years with a view to reducing frequency. 		
	Surveillance PMS2	 Colonoscopy every 1 to 2 years from age 35 years. Review frequency of colonoscopy at age 60 years with a view to reducing frequency. 		
	Surveillance MLH1/MSH2	 Colonoscopy every 1 to 2 years from age 25 years. Annual surveillance is preferred in known mutation carriers. Review frequency of colonoscopy at age 60 years with a view to 2nd yearly frequency. 		
	Risk-reducing medication	 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. 		

EviQ guidelines



Endometrial	Surgical MLH1/MSH2/MSH6	Recommend hysterectomy after childbearing complete or from age 40 years.		
	Surgical PMS2	Recommend hysterectomy after childbearing complete or from age 50 years.		
	Surveillance	There is no evidence for transvaginal ultrasound (TVU) and/or aspiration biopsy.		
Ovarian	Surgical	 Consider risk reducing salpingo-oophorectomy (RRSO) at time of hysterectomy in selected individuals. Recommend HRT at the time of RRSO and continue until the usual time of menopause. 		
	Surveillance	• Do not offer serum CA125 and/or transvaginal ultrasound (TVU). See Cancer Australia for further information		
Gastric	Surveillance	 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. 		
Urothelial	Surveillance	No evidence of benefit but patients encouraged to report symptoms e.g. haematuria.		

^{***}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-tor-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







Misdiagnosed, misunderstood and missing out:

Lynch Syndrome Australia's untold health story







Many families. Many cancers. One common cause.

lynchsyndrome.org.au