Resources

Lynch Syndrome Australia has a comprehensive range of useful resources for both patients and health professionals on the website at **http://www.lynchsyndrome.org.au**

For convenience, the following additional resources maybe useful:

For patients:

- Understanding genetic tests for Lynch syndrome - Information and decision aid (produced by Centre for Genetics Education, NSW Health)
  http://www.genetics.edu.au/Publications-and-Resources/PublicationsBrochuresandPamphlets/Understanding%20Genetic%20Tests%20for%20Lynch%20Syndrome
- Lynch syndrome - A guide for people with Lynch syndrome and their family and friends (Cancer Institute NSW)
  https://www.cancerinstitute.org.au/understanding-cancer/cancer-in-nsw/hereditary-cancers/lynch-syndrome

For health professionals:

- A tool to plan your patient's surveillance regime based on the specific mutation and associated risks
  http://lsearisk.org/
- International Society for Gastrointestinal Hereditary Tumours (InSiGHT)
  https://www.insight-group.org/syndromes/lynch-syndrome/

About Lynch Syndrome Australia

Lynch Syndrome Australia (LSA) is an all-volunteer, survivor-led organisation committed to improving awareness, education, research, advocacy and support for everyone affected by Lynch syndrome.

LSA relies solely on volunteers and donations.

What we do:

- Raise awareness of Lynch syndrome amongst high risk groups, the medical profession and the general public
- Support research into Lynch syndrome to improve detection and surveillance
- Hold Living with Lynch syndrome events
- Provide presentations on Lynch syndrome
- Encourage and empower people to take charge of their own surveillance and general health
- Advocate for individuals with Lynch syndrome

Visit the LSA Website http://www.lynchsyndrome.org.au for:

- Blogs from health professionals experienced in managing individuals with LS
- Patient stories
- Information flyers for patients
- Information flyers for health professionals
- Posters for the waiting room
- Family history templates
- The latest research on Lynch syndrome
- Essential links

www.lynchsyndrome.org.au

info@lynchsyndrome.org.au

Lynch Syndrome Australia

Many families. Many cancers. One common cause.

Lynch Syndrome Information for health professionals

What is Lynch syndrome

Lynch syndrome is an inherited cancer predisposition condition caused by a DNA mutation in one of the four mismatch repair (MMR) genes MLH1, MSH2, MSH6, and PMS2. Carrying a mutation results in the person having a significantly increased risk of developing a range of cancers.

Lynch syndrome was previously known as Hereditary Non-Polyposis Colon Cancer (HNPCC).

People with Lynch syndrome have a higher risk of colon and rectal cancer, endometrial cancer, small intestine cancer, hepato-biliary and pancreatic cancer, gastric cancer, ovarian non-serous cancer, renal pelvis and ureter cancer, bladder cancer, sebaceous gland cancer (and adenoma – Muir-Torre syndrome), prostate cancer, breast cancer and central nervous system cancer.

Personal risks differ depending on which gene is affected, family cancer history and lifestyle factors and patients should be made aware of this difference.

Lynch syndrome is extremely under-diagnosed. Although Lynch syndrome is identified in approximately 3-5% of all colorectal cancers (CRCs), it is estimated that 95% of Lynch syndrome carriers are unaware of their Lynch syndrome status. These carriers are therefore unaware of their increased cancer risk and surveillance needs.

Diagnosing Lynch syndrome

Cancer history

A personal or family cancer history is an important diagnostic tool. Lynch Syndrome Australia provides a simple template for patients to use to assist in identifying a relevant family history. To access the template visit the **Health Professionals section** of the Lynch Syndrome Australia website.
If your patient has had a cancer from the list of Lynch syndrome-associated cancers before age 50 then you should refer her/him to a genetics service.

Lynch syndrome may also be indicated where the family history of cancer meets the **3,2,1 test:**

3 or more family members (including the patient) have been diagnosed with a Lynch syndrome associated cancer

2 consecutive generations or more are affected

1 affected family member is diagnosed before 50 years of age

Family history alone might fail to identify some families with Lynch syndrome; particularly where family size is small or where knowledge of family cancer history is incomplete (e.g. where family members are unknown, or family members are reluctant to discuss their details). It is important to consider these limitations and take a broad approach to assessing a family history.

The Cancer Council ***Clinical practice guidelines for the prevention, early detection and management of colorectal cancer*** recommend that all bowel cancer tumours should be tested for mismatch repair deficiency as a means to subsequently identify Lynch syndrome. This test looks for loss of proteins that indicate that a certain gene is not working in the tumour. Further testing will determine which mutations in the germline and which are only in the tumour. These patients should be referred to a family cancer clinic for genetic counselling and testing. It is also best practice in Australia to test all endometrial cancers under age 60 for mismatch repair deficiency.

Previous diagnostic criteria include the Amsterdam I and Amsterdam II criteria and the (Updated) Bethesda criteria.

A diagnosis of Lynch syndrome is a life-changing experience for a patient and their relatives and they could well take time to adjust to the diagnosis. Consider counselling or referral for ongoing support.

Genetic testing

If a patient is suspected of having Lynch syndrome, you should refer them to a Family Cancer Clinic for genetic counselling and, if recommended, genetic testing. A list of the Family Cancer Clinics and Genetic Services is available in the Health Professionals section of the Lynch Syndrome Australia website at **http://www.lynchsyndrome.org.au/**

Germ line mutations occur in one of the four-mismatch repair (MMR) genes associate with Lynch syndrome, i.e. MLH1, MSH2, MSH6, and PMS2. All patients whose tumours show this loss of staining should be referred to a family cancer clinic.

The Health Professionals section of the Lynch Syndrome Australia website provides further information on what should be included in a referral to a family cancer clinic. See: **http://www.lynchsyndrome.org.au/**. If your patient is diagnosed with Lynch syndrome then you should work with them to develop and maintain an appropriate cancer risk management plan. You should also ensure that your patient understands the need to communicate their diagnosis to at-risk blood relatives.

Risk management following diagnosis

Lynch syndrome cancers can be aggressive and often occur at an earlier age than in the general population. It is therefore essential that health professionals work with people with Lynch syndrome to properly assess their risk and take steps to manage and minimise their risk.

Understanding a patient's risk

Cancer risk for Lynch syndrome carriers is influenced by cancer history, gene type, familial and environmental factors (e.g., some families have extraordinarily high rates of particular Lynch syndrome cancers). Important information on cumulative, population cancer risk for people with Lynch syndrome by age, gene variant and gender is available on **www.lscarisk.org.** In addition, family cancer clinics provide evidence-based and more personalised risk estimates and screening advice.

Taking steps to manage risk

Health professionals can play a key role in identifying and assisting patients with Lynch syndrome including co-ordinate their risk reduction by considering chemo-prevention (with aspirin), preventive surgery and instigating an appropriate surveillance regime. In Australia, the NSW Cancer Institute produce evidence-based cancer treatment protocols for Lynch syndrome. These can be found at **http://www.eviq.org.au/**

Current recommendations are:

- Men and women: Annual colonoscopy is recommended from age 25 (MLH1 or MSH2 gene mutation) or age 30 (MSH6 or PMS2 gene mutation), or starting 5 years younger than the patient's youngest CRC affected relative which ever comes first;
- Women: Prophylactic total abdominal hysterectomy with bilateral salpingo-oophorectomy (TAH-BSO) is to be considered after childbearing is complete or by 40 years of age.

- The use of Aspirin as chemo-prevention is currently in trials in Australia.

Indeed, surveillance recommendations have changed extensively since the first Lynch syndrome patients were identified. You must consider a patient's risk management strategy and surveillance regime, together with a patient's own and family history of cancer, lifestyle/environmental factors and continually evolving Australian and global best practice. International recommendations for best practice may be considered where the patient's family history or other factors are relevant. The International Society for Gastrointestinal Hereditary Tumours (InSiGHT) is an international multidisciplinary, scientific organisation which includes useful information on Lynch syndrome at **https://www.insight-group.org/syndromes/lynch-syndrome/**

Enrolment of a patient to the state Hereditary Cancer Registry (if available) is also strongly advised, as they provide a screening reminder service as well as other information and services.

A diagnosis of Lynch syndrome may affect childbearing decisions. You should also consider a referral to a fertility specialist for discussions on preimplantation genetic testing and assisted conception.