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MOUNT SINAI HOSPITAL
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Understanding Lynch Syndrome

Information for people who may have Lynch syndrome and their family members

MOUNT SINAI HOSPITAL
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RESEARCH TREATMENT EDUCATION SUPPORT TO IMPROVE TREATMENT

Introduction

Welcome to our Familial Gastrointestinal Cancer Registry.

We are a group of genetic counsellors, researchers, surgeons, pathologists, gastroenterologists and molecular geneticists who work together to try to understand some types of **hereditary** cancer. Hereditary means passed down from parent to child through genes.

- Our role is to identify families that have a possible hereditary cancer syndrome.
- By doing so, we can find those who have a higher risk to develop certain types of cancers and recommend appropriate ways to reduce their risks

What support is available?

The goal of the Familial Gastrointestinal Cancer Registry is to offer **education and support** to families with LS and other types of hereditary cancer.

In addition to genetics, we focus on prevention and early detection of colorectal cancer and also help organize screening for families with and without LS.

Our Registry collaborates on research with other international groups that also specialize in hereditary cancer. Through research we aim to learn more about possible causes, treatments, effective screening and the genetics of colorectal and other cancers associated with LS.

This booklet is meant to provide an overview of Lynch syndrome and not to replace a thorough genetics assessment.

If you have any questions about the information provided, please contact us or another genetics clinic or speak to your physician.

Other LS-related cancer screening

At this time, routine screening for other LS-related cancers has not been shown to be effective.

Based on family history of other types of cancer, other screening procedures may be recommended on an individual basis.

Some families with LS may develop specific skin findings (lesions) called keratoacanthomas or sebaceous adenomas/carcinomas). **Any unusual skin findings should be reported to a doctor**.

Individuals with LS are encouraged to discuss all changes in general health with their doctor.

Please note: Screening recommendations must be given by a genetics specialist and/or doctor after a complete assessment of a family.

Please talk to your doctor if you have questions or concerns about screening recommendations for you.

Is cancer hereditary?

Everyone has a chance of developing cancer in their lifetime.

As we get older, the cells that line our organs are damaged by environmental factors, such as diet, lifestyle and as a normal part of the aging process.

Over time, this damage adds up and can lead to cancer.

►This is thought to explain the majority of cases of cancer, which we call **sporadic** (happens by chance).

A smaller number of cancers are explained by a hereditary factor (something running in a family) that increases one's risk to develop cancer.

In this brochure we will be reviewing a hereditary cancer condition called Lynch syndrome **(LS)** or Hereditary Non-Polyposis Colorectal Cancer **(HNPCC).**

We will review:

- Cancers that are associated with **LS**
- Background genetics
- The testing process
- Screening information

What is Lynch Syndrome or HNPCC?

LS is a hereditary syndrome (can be passed down from parent to child through genes) that causes higher risks for cancer.

The most common cancer associated with LS is **colorectal cancer** (CRC), which refers to cancer that starts in either the colon or the rectum.

The chance of developing colorectal cancer in the average person is about 1 in 16, or 6%, whereas in people with LS this risk is much higher.

LS accounts for about 2% of all colorectal cancers.

Cancer of the **endometrium** (lining of the uterus) in women is the second most common cancer associated with LS.

What are the benefits and limitations of genetic testing?

Colorectal Cancer Screening*

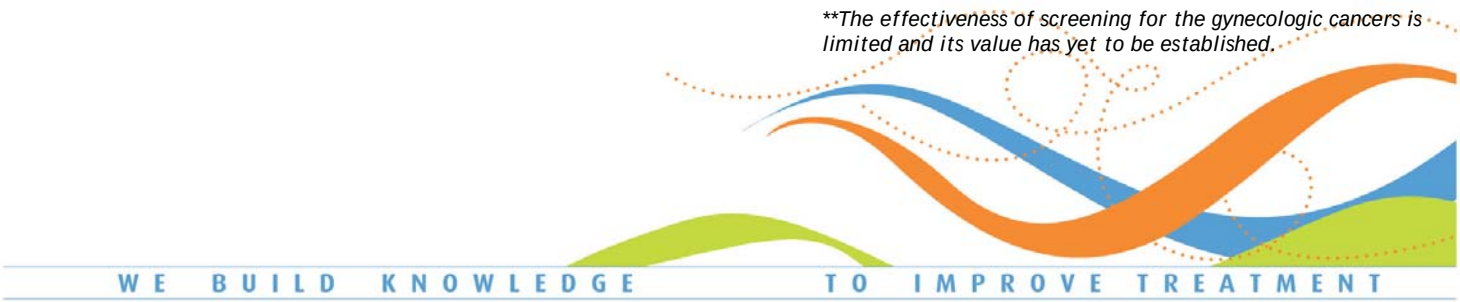
- Colonoscopy (a procedure used to see inside the colon and rectum) **every 1-2 years starting at age 20-25**
- Some families may be advised to start screening at an earlier age

Gynecological Cancer Screening**

- Trans-vaginal and pelvic ultrasound **every year**
- CA125 blood test **every year**
- Discuss oral contraceptive use with physician
- Pay attention to signs and symptoms of endometrial cancer such as irregular or heavy vaginal bleeding and go see your doctor immediately if either of these symptoms occurs
- Prophylactic (preventative) removal of uterus, fallopian tubes and ovaries may be considered when child-bearing is completed.

**Evidence shows that regular colonoscopy reduces the risk of developing colorectal cancer in LS patients.*

***The effectiveness of screening for the gynecologic cancers is limited and its value has yet to be established.*



What are the benefits and limitations of genetic testing?

The decision to have genetic testing involves **carefully considering** what the results could mean.

BENEFITS:

- Genetic testing can be helpful because it gives information about a person's risk to develop certain types of cancer.
- Having this information might encourage someone to start having the right cancer screening done and to better manage their health. For example, genetic test results could influence the type of surgery a patient decides to have if diagnosed with cancer.

LIMITATIONS:

- Genetic testing may be hard for some people who feel that knowing their risk would cause them anxiety (worry or stress).

HELP IS AVAILABLE:

- Before having genetic testing, a person must talk about the benefits, risks and limitations of genetic testing with a genetic counsellor.
- Afterward, some individuals may decide that testing is right for them while others may choose not to be tested.

What is Lynch Syndrome or HNPCC?

These cancers are also more common in LS than in the average person:

- small bowel
- ureter and kidney (specifically transitional cell carcinomas)
- Ovary
- Stomach
- Hepatobiliary tract (e.g. pancreas and bile duct)
- Brain
- Skin (specifically sebaceous adenomas/ carcinomas and keratoacanthomas)

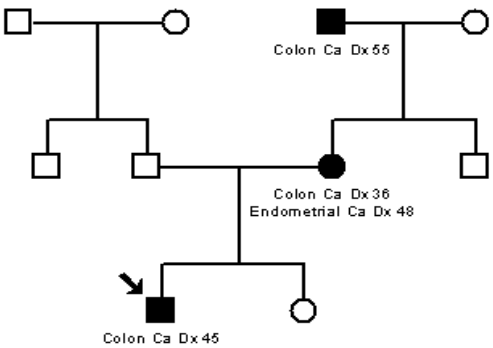
Although the risks for the cancers listed above are a bit higher for someone with LS, **the actual risks are still quite low.**

Who is at risk for LS?

We see differences between families with sporadic cancer and families with LS .

LS families often have:

- More than one person with CRC and other LS-related cancers (i.e. endometrial cancer),
- More than one generation with these cancers
- More than one cancer in a person
- Cancers diagnosed at younger ages (usually under age 50).



A genetic counsellor can tell if your family is at risk for LS.

Commonly asked questions about genetic testing for LS

What if I have an LS gene mutation?

If a mutation is identified in a LS gene, it confirms the diagnosis of LS. Your genetic counsellor will discuss screening recommendations for you and your family as well as the availability of genetic testing for your at-risk family members.

Can my children be tested for the mutation?

Genetic testing is not usually offered to children under the age of 18 because the cancer risk associated with LS usually occurs after that age. Exceptions can be made in certain situations, e.g. if a very young cancer has been seen in the family. Please speak to your genetic counsellor with any concerns.

What if my relative (or I) did not inherit the LS gene mutation known to be running in my family?

This would mean they do not have LS. Their risk to develop cancer would be the same as someone in the general population. Population screening recommendations would be given. It also means their children would not be at risk to inherit LS.

Commonly asked questions about genetic testing for LS

What if the tumour sample does not have characteristics of LS?

If the tumour does not have features of LS it means that LS was probably not the cause of that cancer.

BUT the tumour test is just a screening tool, so it cannot completely rule out LS.

It also does not rule out **all** hereditary causes of cancer. Since there may still be an inherited form of cancer in the family, your family history will be reviewed again and the right screening suggestions will be given to you and your family.

What if the tumour showed features of LS, but no genetic mutation was seen with my blood test?

Depending on the **family history** and the **tumour result**, LS may still be a possibility in the family.

Your genetic counsellor will look at your results together with your family history and discuss screening recommendations for you and your family.

What causes LS?

LS is hereditary, meaning that it can be passed down from parent to child through genes. Genes carry instructions to help our body work properly.

There are five genes currently known to be associated with LS, named *MLH1, MSH2, MSH6*, *PMS2* and *EPCAM (TACSTD1)* that each of us have and that help protect us from developing cancer.

Individuals with LS are born with a mutation (problem) in one of these genes.

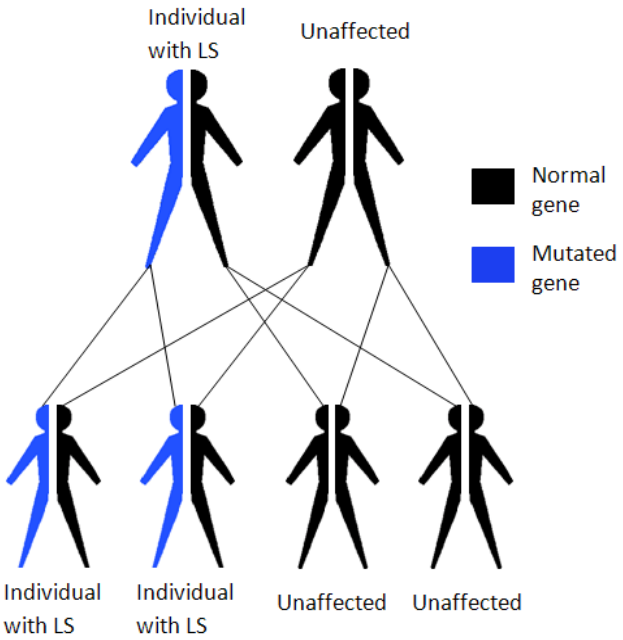
Everyone has two copies of every gene, one copy that was inherited from their father and one copy from their mother.

Children and siblings of someone with LS have a 50% (1 in 2) chance of inheriting that same LS gene mutation. This is called **autosomal dominant inheritance.**

It is **not** the cancer itself that is passed down from parent to child, it is a **higher chance** of developing cancer that is passed down.

The gene mutation makes a person's chance of developing cancer higher but it **does not mean that they will definitely develop cancer.**

Inheritance of LS



Lynch syndrome has an **autosomal dominant pattern** of inheritance

What does this mean?

- Someone with a gene mutation for LS has a 50% chance of passing that mutation on to each of their children

How does genetic testing for LS work?

A person referred to a genetics clinic will be asked to give **family history** information.

A **genetic counsellor** will decide if the family looks like they might have sporadic cancer or a possible hereditary cancer syndrome like LS.

If a family is suspected of having LS, genetic testing may be offered.

Steps in genetic testing for LS:

1. Look at tumor samples. Most tumors associated with LS show special characteristics. If these characteristics are seen then we move on to the second step: genetic testing using a blood sample.

2. Genetic testing looks for a mutation in one of the five genes that are known to be associated with LS. If a mutation is found in a family, relatives can be tested to see if they have that mutation.

Sometimes the testing process does not find a mutation in a family. In these cases we may not be able to confirm or rule out a hereditary cancer syndrome.