What is Li-Fraumeni Syndrome?

Li-Fraumeni Syndrome (LFS) is one of many different genetic conditions we know of today. A gene is a section of our DNA, or genetic code, that controls a particular function within the body, and when there is a mistake within this code it is known as a mutation. In this case the affected gene is called TP53, which makes a protein known as p53. This protein is very important in processes that stop our cells from becoming cancerous, and as such is often referred to as the 'guardian of the cell'. Having this mutation means that cells are not as effectively protected against becoming cancerous. The risk of developing cancer is therefore increased. It is an inheritable condition – a patient with LFS will have a 50/50 chance of passing it on to any children.

How do I get tested?

If you believe you may be at risk (see 'Am I eligible for genetic testing?' section), ask your oncologist or GP to refer you to a genetics unit, where you will meet with a genetics counsellor. They can answer any questions you may have about LFS, and the pros and cons of being tested.

The process involves a simple blood test and you will normally get your results in 4-8 weeks.

What does having LFS mean for my health?

Having LFS does not automatically cause illness. What it does mean is that there is a greatly increased *risk* of developing cancer. The most commonly seen cancers in LFS patients are breast, brain tumours, leukaemia, soft tissue sarcomas, bone sarcomas and adrenocortical carcinomas, although the risk of developing any type of cancer is greater than normal. Cancers often develop early and affect children and young adults at a far greater rate than the general population.

What treatment is available to me?

There is currently no cure for LFS. However, for some cancers there are measures that can be taken to reduce the risk. Avoiding smoking and too much sun exposure (particularly burning), will minimize your risk of smoking related cancers and some skin cancers. Women can opt for annual breast MRIs to detect any abnormalities, or alternatively undergo a risk-reducing mastectomy.

It is important to make your GP aware of your diagnosis, and to discuss any unusual or concerning symptoms with them. They can then be prepared to fast-track you for assessment and immediate investigation if necessary.

I have been diagnosed with LFS – should my family be tested too?

If you have been diagnosed with LFS, your immediate family should be offered testing too. Siblings, children and parents should all have the opportunity to have a genetic test.

If you have young children, deciding on their behalf whether to test them or not can be a complex and difficult question, and is something that can be discussed with your genetics counsellor. They can also offer advice on how to introduce the idea of genetic testing to older children, who may have enough understanding to be involved in the decision making process.

If you wish to avoid any future children inheriting LFS, there are options available to help you with this. Ask your doctor to refer you to an assisted conception unit to learn more about what is available to you.

Who in my child's life should I tell?

It is helpful to explain your child's diagnosis to their teacher, school nurse or anyone else working closely with them. They can offer emotional support and understanding through any hospital appointments or treatment that may be happening. For further advice and support for your child, ask your GP or oncologist about any extra counselling that is available to them.

Am I eligible for genetic testing?

Undergoing a genetic test is recommended by the Institute of Cancer Research for patients meeting any of the following criteria:

Any individual with:

- Adrenocortical cancer at any age
- Choroid plexus cancer at any age
- Rhabdomyosarcoma below age 5
- Breast cancer below age 30 (and BRCA1/2 negative)
- Two or more primary cancers, including one below age 46, of the following: sarcoma, breast, brain, adrenocortical or any childhood cancer.

***OR* two relatives:**

With sarcoma, breast, brain, adrenocortical or any childhood cancer

- one diagnosed below age 36
- another diagnosed below 46

***OR* three relatives:**

- One with sarcoma below age 45
- Another with any cancer below 45
- A third with any cancer below 45 or a sarcoma at any age

If you believe you are at risk and wish to be tested, talk to your oncologist or GP who will be able to refer you to a genetics counselor.

Help And Support

The George Pantziarka TP53 Trust has been set up to help patients and families diagnosed with Li-Fraumeni Syndrome. Please contact us through the website, www.tp53.co.uk if you wish to discuss anything or talk to other LFS patients.

The following websites may also be helpful to you:

- www.lfsassociation.org - An American organisation dedicated to LFS research and support
- livinglfs.blogspot.com – Another LFS patient group based in the USA
- www.clicsargent.org.uk - Caring for children and young people with cancer
- www.maggiescentres.org - Centers set up around the UK to provide support to patients with cancer and their families
- www.thehaven.org.uk - Centers providing support for patients with breast cancer
- www.geneticalliance.org.uk - A UK charity helping people with genetic conditions

George Pantziarka TP53 Trust

Li-Fraumeni Syndrome

Information for patients and families

www.tp53.co.uk