Understanding What Causes LFS

LFS is caused by a mutation in the TP53 tumor suppressor gene. When working properly, this gene controls cell growth, preventing cancers from forming. Individuals with LFS lack a properly functioning p53 gene.

Know the Facts on Diagnosis

Determining if a person or family has LFS begins with taking a family history. Individuals with an adrenocortical carcinoma or choroid plexus carcinoma, as well as those meeting the criteria listed within, should be evaluated for LFS.

A referral will often be made to a genetic counselor, a health care provider who specializes in diagnosing hereditary conditions, to provide families with an opportunity to discuss the implications of genetic testing. Genetic testing involves analyzing a sample of blood or saliva for alterations in the TP53 gene. Ideally, the process of genetic testing should begin with a family member who has had cancer. If an alteration causing LFS is identified, then other relatives should be tested to determine who else may be at risk.

Important Li

Awareness of LFS is Important

LFS is a rare genetic disorder that greatly increases the risk of developing many types of cancer. Cancers related to this condition may occur at any age, but a characteristic feature of LFS is a high risk for cancer occurring in childhood. Approximately 40% of individuals with LFS will develop cancer in childhood. LFS affects both men and women and this condition has been identified in families all over the world.

The Most Common Types of Cancer Associated with LFS Include:

- Breast cancer
- Adrenocortical cancer
- Leukemia
- Soft tissue sarcomas
- Osteosarcomas
- Brain tumors
- Colon cancer
- Stomach cancer
- Lung Bronchoalveolar cancer

If You Meet Any of the Following Criteria, Talk to Your Physician About LFS Screening:

- You have been diagnosed with a cancer within the LFS spectrum of cancers (soft tissue sarcoma, osteosarcoma, brain tumor, pre-menopausal breast cancer, adrenocortical cancer, leukemia or bronchoalveolar lung cancer) before age 46 AND have at least one first or second degree relative with an LFS related cancer before 56 years of age.
- You have been diagnosed with multiple cancers, 2 of which belong to the LFS spectrum of cancers, with your first cancer before age 46.
- You are a woman diagnosed with breast cancer before age 35 and have tested negative for a BRCA1 or BRCA2 mutation.
- You have a history of adrenocortical cancer or choroid plexus carcinoma.

Why Diagnosis Can Make a Difference

While the diagnosis of LFS can seem overwhelming, it is critical that patients be aware of the disorder so they may adequately manage their healthcare. Because **patients with LFS often develop cancers earlier**, regular check-ups and cancer screenings are extremely important – the sooner a cancer is diagnosed and treated, the better the chances of a successful treatment. Coordination with individual medical providers is also crucial to ensure that the LFS diagnosis is being considered in the treatment of conditions, particularly in regard to the use of radiation therapy in cancer treatment.

LI-FRAUMENI SYNDROME

LFS

ASSOCIATION

Find Support for the LFS Community at the LFSA

Li-Fraumeni Syndrome Association
P.O. Box 6458 Holliston, MA 01746
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