Li-Fraumeni syndrome

Aflac Cancer & Blood Disorders Center
CHILDREN'S HEALTHCARE OF ATLANTA

Li-Fraumeni syndrome (LFS) is a syndrome that predisposes individuals to develop several types of benign and malignant tumors. In childhood, this often includes:

- Choroid plexus carcinoma, a type of brain tumor
- Adrenocortical carcinomas, which is present in the adrenal glands located on top of the kidneys
- Osteosarcomas, or bone tumors; and rhabdomyosarcomas, or soft tissue tumors

Cause

LFS is caused by genetic changes that occur in the TP53 gene, which is often called "the guardian of the genome" and has several vital functions, including preventing tumor development and controlling the cell cycle. Some cases of LFS may occur on their own without being passed down from a parent. However, in most cases genetic alterations, or changes in the TP53 gene that cause LFS, are inherited from one parent.

Diagnosis

Diagnosis of LFS occurs through genetic testing, which is often facilitated by a genetic counselor and/or clinical oncology team.

Treatment and surveillance

Due to the increased risk of cancer, surveillance is recommended for both children and adults with LFS. This is best done at a center where physicians, genetic counselors and geneticists have expertise in LFS.

A diagnosis of LFS may change treatment plans for existing cancers, as well as the medical management and future surveillance plans, as individuals with LFS are extremely sensitive to radiation and are at an increased risk for secondary cancers.

Who is at risk?

Once an individual has been identified with LFS, all family members at risk may choose to pursue genetic counseling and testing. A genetic counselor can help identify family members who may be at risk for LFS.

References

ncbi.nlm.nih.gov/books/nbk1311/
Abeloff's Clinical Oncology 5th edition, pages 143-152