About TP53 Mutations and Li-Fraumeni Syndrome

About Genes

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work properly, they help stop cancer from developing.

Sometimes changes to genes can happen. These changes are called mutations. Mutations can make the genes stop working and raise the risk for certain types of cancer. Some mutations can be passed through families. When that happens, family members who have the mutation have a *hereditary cancer syndrome*.

The *TP53* gene plays a major role in controlling how cells grow and divide. When *TP53* is not working right, cells can grow out of control. This can lead to cancer. A person who inherits a mutated copy of *TP53* has a hereditary cancer syndrome called Li-Fraumeni syndrome.

Li Fraumeni Syndrome

Li-Fraumeni syndrome (LFS) raises the risk of getting many types of cancer. LFS-related cancers often happen in childhood or young adulthood. People with LFS often develop more than one type of cancer in their lifetime. LFS is very rare.

LFS and Cancer Risk

People with LFS have a 1 in 2 chance to get cancer by age 30. Almost all women with LFS (93 in 100) get cancer in their lifetime. The lifetime cancer risk is somewhat lower for men with LFS (68 in 100).

People with LFS often get one or more of these cancers:

- Adrenocortical carcinoma (cancer in the outer layer of the adrenal gland)
- Bone and soft tissue sarcomas
- Brain tumors
- Breast cancer

They may also get these cancers:

- Colorectal
- Leukemia
- Lung
- Pancreas
- Skin cancers

Recommendations

MEN AND WOMEN

Starting in infancy or at time of diagnosis: Yearly total body MRI and brain MRI; physical exam every 6 months

Starting at 18 years of age: Yearly dermatalogy exam

Starting at 25 years of age: Colonoscopy and upper GI endoscopy every 2-5 years

WOMEN

Starting at 20 years of age: Yearly breast MRI, clinical breast exam every 6 months

Starting at 30 years of age: Mammogram and breast MRI every year (scheduled 6 months apart)

Consider surgery to remove both breasts

CHILDREN

Starting at birth: Physical exam and ultrasound of abdomen and pelvis every 3–4 months

KIDS AND SIBLINGS

Siblings and children of people with LFS have a 1 in 2 chance of also having LFS. We recommend genetic testing and counseling for at-risk family members of any age.

Family members who do not have LFS probably do not have a higher risk of getting cancer. They should follow general cancer screening guidelines.

Knowing which side of the family carries LFS is important. That information can help us decide who is at risk for cancer. A genetic counselor can help you discuss LFS with your family members.

Do you have questions about your risk for cancer? Our doctors and genetic counselors can help find the cancer screening plan you need. Call Huntsman Cancer Institute's Family Cancer Assessment Clinic to learn more: 801-587-9555.

For more patient education information: Call 1-888-424-2100 toll free • Email cancerinfo@hci.utah.edu • Visit www.huntsmancancer.org
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