Mass General Brigham
Mass General Cancer Center

TP53 gene (Li-Fraumeni syndrome): what you need to know

What does it mean to have a *TP53* pathogenic variant?

Having a *TP53* pathogenic variant, also known as a mutation, means a person has a version of the *TP53* gene that does not work properly. This may cause a cancer predisposition condition called Li-Fraumeni syndrome (LFS) or LFS-like syndrome.

What is my risk for cancer if I have a *TP53* pathogenic variant?

If you have a *TP53* pathogenic variant causing LFS or LFS-like syndrome, you have an increased risk to develop one or more cancers which can occur anytime from early childhood through late adulthood.

Individuals with a *TP53* pathogenic variant with the classic form of LFS have the following risks:

- For people assigned female at birth, the lifetime risk of developing cancer is > 90%.
- For people assigned male at birth, the lifetime risk of developing cancer is > 70%.
- For all people, the lifetime risk of developing a 2nd cancer is 40-50%.

What type of cancer am I more likely to develop if I have a *TP53* pathogenic variant?

The *TP53* gene is important for preventing cancer in many different parts of the body. Therefore, individuals with a pathogenic variant in this gene causing LFS or LFS-like syndrome are at risk to develop several different types of cancer. The types of cancer most common in LFS or LFS-like syndrome are:

- Sarcoma
- Breast cancer (in people assigned female at birth)
- Brain tumors
- Acute leukemia
- Adrenal cortical cancer

Many other types of cancer have also occurred in families with LFS or LFS-like syndrome.

If I have a *TP53* pathogenic, what is the chance my family members will have it too?

There is a 50% chance that a person with a pathogenic variant will pass it on to each of their children. In most cases, siblings of a person with a pathogenic variant have a 50% chance to have the pathogenic variant. Additionally, other family members are at risk to have the pathogenic variant.