LI-FRAUMENI SYNDROME

WHAT IS LI-FRAUMENI SYNDROME?

Li-Fraumeni syndrome (LFS) is a rare genetic condition caused by inherited pathogenic variants (mutations) in a gene called *TP53*. Individuals with LFS have nearly 100% lifetime risk of cancer of various types that can develop anytime from early infancy to late adulthood. Individuals with LFS have a 50% chance of passing on the condition to their children. Siblings, parents, and other relatives may also have LFS. Individuals with LFS regularly undergo intensive cancer-related screenings and procedures (e.g., whole-body MRI, brain MRI, breast MRI, colonoscopy). There is no cure for LFS or effective ways to reliably detect and treat all possible cancers. Ideally, people with LFS receive care from an interdisciplinary team of health care providers, including a genetic counselor, to ensure comprehensive care. Below are examples of multiple challenges that individuals with LFS and their families may face.

PERSONAL CHALLENGES

Chronic grief
- Concurrent and multiple cancer diagnoses and deaths in the family and LFS community

Multidimensional loss & PTSD
- Trauma from cancers, deaths, and other losses related to uncertainty (e.g., ambiguous loss, unacknowledged loss)

Scanxiety
- Anxiety surrounding upcoming screenings and results

COMMUNITY CHALLENGES

Access to quality healthcare
- Invalidating health encounters and role reversal where the person with LFS becomes the educator for providers

`Family communication
- Difficulty talking about LFS, risk, genetic testing, reproductive beliefs, and health decisions

`Supportive networks
- Difficulty finding support for individuals with LFS and family

STRUCTURAL CHALLENGES

Health policy
- Lack of health policies for affordable screening and care for all individuals with LFS

Health insurance coverage
- Private and public insurance policies that do not cover all LFS screening or cancer care

Scarcity of health systems
- Difficulty finding local specialty care for LFS, resulting in burdensome travel costs

RESOURCES

LFSA
LFSASSOCIATION.ORG

NATIONAL CANCER INSTITUTE
LFS.CANCER.GOV

LIVINGLFS
LIVINGLFS.ORG

Authors: Catherine Wilsnack, Camella Rising, and Carly Grant