Introduction

Li-Fraumeni Syndrome (LFS) is a genetic cancer predisposition syndrome that is defined as any germline mutation in the TP53 tumor suppressor gene. It is passed down in an autosomal dominant pattern, with a 50% chance of passing the mutated gene to each child. In response to stress signals, the tumor suppressor gene controls a wide range of processes including apoptosis, DNA replication and repair, and regulatory control over the cell cycle. When this gene is mutated, cells lack control over these processes. There are two different types of criteria used to diagnose LFS, the classical and the Chompret criteria, which are defined by patients with these processes. There are two different types of criteria used to diagnose LFS, the classical and the Chompret criteria, which are defined by patients with any first degree relatives diagnosed with particular cancers at certain ages. There are certain cancers that are associated with LFS, and prevalence of these cancers varies based on age. In the pediatric/young adult population, risk is highest for sarcomas, brain tumors, adenocortical carcinoma, leukemia, and early onset breast cancer. Cumulative cancer risk associated with LFS has been estimated to be approximately 50% by age 40 years and up to 90% by age 60 years.

Patients are recommended to follow regular surveillance screening guidelines, which have been shown to improve overall survival. Multiple screening guideline protocols are utilized, including that of the MD Anderson Children’s Cancer Hospital pediatric LEAD clinic. LFS can have a significant physical and psychological impact on children and families that it affects.

Practice

The mission of the LEAD program at MDACC is to improve the patient experience, quality of life, and long-term survival for children diagnosed with LFS by providing age-appropriate screening and supportive care services from professionals with LFS expertise in a dedicated program.

Li-Fraumeni Syndrome Education and Early Detection Clinic

Making a difference in quality of life and long-term survival through screening and education

Heather Meador, APRN, CPNP, CPHON & Whitney Throckmorton, MPAS, PA-C

Outcomes

At this time, 224 pediatric patients have been tested for TP53 mutations at MDACC, and 34 have tested positive for LFS. There are currently ~20 patients receiving care in the MDACC pediatric LEAD clinic. Of those 18, five patients have received treatment for at least one malignancy. Once a patient turns 21, they transition to our adult LEAD clinic, as they focus on additional malignancies that affect the adult population.

References


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