TP53 (p53) testing for Li-Fraumeni Syndrome (LFS)

Record Status
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Authors' objectives
Li-Fraumeni syndrome (LFS) is an autosomal dominant cancer predisposition syndrome associated with a significantly increased risk of cancer, often occurring when the patient is at a young age. It has been estimated that individuals with LFS have a 60% chance of malignancy by age 45 and a 95% chance by age 70. The cancers that occur most often in LFS families include premenopausal breast cancer, soft tissue sarcomas, osteosarcomas, brain tumors, and adrenocortical carcinomas. In addition, colorectal cancer, leukemia, lung cancer, choroid plexus carcinoma, and malignant phyllodes tumors (sarcomas of the connective tissue in the breast) appear to be more common among LFS kindreds than among cancer patients in the general population. Although the above cancers are most frequent, LFS patients may develop a wide variety of malignancies. LFS patients are also at an increased risk for multiple primary tumors, which is uncommon among the general population. Two forms of LFS have been recognized: classic LFS and Li-Fraumeni-like syndrome (LFLS). Patients with classic LFS meet the following criteria: the diagnosis of a sarcoma before age 45; a first-degree relative diagnosed with any cancer at age < 45; and an additional first- or second-degree relative diagnosed with any cancer at age < 45 or a sarcoma at any age. Patients with LFLS meet some, but not all, of the above criteria. Two sets of less stringent criteria (the Birch criteria and the Eeles criteria) have been developed for the diagnosis of these patients, and a third set of criteria (the Chompret criteria) have been proposed for identifying patients appropriate for genetic testing. The majority of patients satisfying the criteria for classic LFS (and a smaller proportion of patients satisfying the Birch, Eeles, or Chompret criteria) are found to carry germline variants in the TP53 gene, which is located on chromosome 17 at band p13.1. TP53 (also known as p53), a tumor suppressor gene, encodes a transcription factor with critical functions in cell cycle control and apoptosis (programmed cell death). Somatic variants in TP53 are found in more than half of sporadic cancers, illustrating the importance of TP53 in tumor suppression. Given the wide spectrum of tumors that may occur in TP53 variant carriers, and the fact that many are difficult to detect early, effective screening is difficult. At this time, there are no published guidelines for the surveillance of TP53-positive individuals that have been shown to improve health outcomes. The treatment of LFS patients is tumor specific and may be limited in that some treatment modalities (particularly radiotherapy) are not recommended due to their DNA damage-inducing properties.

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