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LETTER TO THE EDITOR

Tricuspid mass-curious case of Li-Fraumeni syndrome: A letter to the editor

Mohammad S Al-Haggar, Zahraa A Abdelmoneim

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Abstract

We focus specifically on the rare occurrence of cardiac thrombi in Li-Fraumeni syndrome (LFS). LFS is a hereditary risk to a diverse range of specific, uncommon, malignancies. Children and young adults have a heightened susceptibility to many malignancies, particularly soft-tissue and bone tumors, breast malignancies, central nervous system malignancies, adrenocortical carcinoma, and blood cancers. Additionally, LFS patients may experience other cancer types such as gastrointestinal, lung, kidney, thyroid, and skin cancers, along with those affecting gonadal organs (ovaries, testicles, and prostate). An accurate diagnosis of LFS is crucial to enable affected families to access appropriate genetic counseling and undergo surveillance for early cancer detection.

Key Words: Li-Fraumeni syndrome; Cancers; Cardiac thrombus; Genetic counseling; Surveillance

Core Tip: Li-Fraumeni syndrome (LFS) is an inherited propensity to various distinct, frequently uncommon malignancies. Pediatric and adolescent age groups are more likely to acquire many malignancies, including soft-tissue and bone tumors, breast cancer, central nervous system cancers, adrenocortical carcinoma, and blood cancers. LFS individuals may also develop various types of cancer, including gastrointestinal, lung, kidney, thyroid, and skin cancers, as well as those affecting the gonadal organs. An accurate diagnosis of LFS is critical for afflicted families to receive appropriate genetic counseling and be monitored for early cancer detection.

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TO THE EDITOR

Li-Fraumeni syndrome (LFS) is linked to elevated susceptibility to a wide range of cancers occurring in all age groups[1]. Individuals with LFS face a lifelong cancer risk of at least 70% for men and over 90% for women. The most common malignancies in LFS include adrenocortical malignancies, breast malignancies, nervous system malignancies, bone malignancies, and soft-tissue malignancies. LFS is connected with an elevated occurrence of multiple cancer forms, including leukemia, lymphomas, gastrointestinal cancers, prenatal choriocarcinoma, and other reported malignancies[2,3].

LFS diagnosis is confirmed when a proband fulfills all three traditional LFS criteria and/or presents a pathogenic (or likely pathogenic) germline variant in *TP53* detected through molecular analysis. Typical LFS criteria (60%-80% show a germline *TP53* harmful mutation)[2]: (1) A patient has a sarcoma diagnosed before the age of 45; (2) A first-degree relative has malignancy before the age of 45; and (3) A first- or second-degree relative has malignancy before 45, or with sarcoma at any age.

The Chompret criteria for clinical diagnosis of LFS has new diagnostic tools to detect diseased individuals in addition to the conventional items described before. Anyone with a personal and family history that fits one of the following three criteria may be diagnosed with LFS and tested for the *TP53* gene mutation:

Criterion 1: A tumor from the LFS cancer spectrum diagnosed before the age of 46. This includes soft tissue sarcoma, bone tumor, pre-menopausal breast malignancy, central nervous system (CNS) cancer, adrenal cortical malignancy, blood cancer, and lung malignancy.

At least one first- or second-degree family member has an LFS-related cancer, with the exception of breast cancer if the individual is under the age of 56 or has several cancers.

Criterion 2: An individual with various cancers, excluding many breast malignancies, two of them are on the LFS cancer spectrum, and the first of them developed before the age of 46 (A case report).

Criterion 3: An individual has adrenocortical carcinoma or a malignancy in the choroid plexus, which is a membrane surrounding the brain, independent of family history.

Furthermore, individuals have anaplastic rhabdomyosarcoma, females with breast malignancy before the age of 31, and individuals have hypodiploid acute lymphoblastic leukemia and SHH medulloblastoma, regardless of family history, should be evaluated.

The documented case[1] involved a 30-year-old female patient with a background of multiple cancers meeting the criteria for LFS. She visited the cardio-oncology clinic after a heart tumor in the right ventricle was discovered with follow up echocardiogram, which was being performed to monitor two atrial septal defects. During her assessment, she reported no recent symptoms such as chest discomfort, palpitations, exertional dyspnea, orthopnea, or peripheral edema. Clinical examination revealed no signs of murmurs, arrhythmias, jugular venous distention, lung crepitation, or leg swelling. A transthoracic echocardiogram identified a 1 cm × 1 cm mass near the tricuspid valve. The patient subsequently had a cardiothoracic operation, during which a biopsy revealed a 2 cm mass on the posterior leaflet of the tricuspid valve with a slender stalk. The mass was excised from the stalk, and the specimen was found to be a large, structured thrombus without malignancy. Additionally, her atrial septal defects were repaired during the surgery. Her surgical repair was uncomplicated.

The case report[1] did not mention the exclusion or discussion of other potential causes of a cardiac thrombus, such as the type of chemotherapy that the patient received, history of central venous catheter insertion, or any additional symptoms or signs suggestive of infective endocarditis.

CLINICAL IMPLICATIONS

Diagnosing LFS necessitates regular oncological management and surveillance for malignancies to prevent their occurrence. Surveillance involves a thorough clinical assessment and imaging of the abdomen and pelvis every 3-4 months from birth until age 18. Additionally, a yearly CNS examination and whole-body magnetic resonance imaging (MRI), including a brain MRI, are required at onset of diagnosis.

For patients 18 years and older, a clinical examination should be conducted every six months, with an annual imaging of the abdomen and pelvis and a skin examination. Females should undergo a physical breast assessment every 6-12 months starting at age 20-25, an annual breast MRI starting at age 20-30, and a mammogram along with a breast MRI annually from age 30 to 75. Upper endoscopy and colonoscopy are needed every 2–5 years starting at age 25[4,5].

Genetic counseling is crucial, as LFS is inherited in an autosomal dominant pattern. Patients with LFS have inherited a *TP53* harmful mutation from one parent, while 7-20% have a *de novo TP53* pathogenic variant. Children of someone with an established LFS diagnosis (meeting typical LFS criteria or having a heterozygous germline *TP53* harmful mutation) have a 50% chance of having the variant and the associated cancer risks. Predictive testing for at-risk family members, as

well as prenatal and preimplantation molecular analysis, are available if a *TP53* germline harmful mutation is identified in the family[6].

LFS is an uncommon genetic condition that elevates the occurrence of various malignancies. Detecting this syndrome requires monitoring for additional malignancies and providing thorough genetic counseling to enable early diagnosis and treatment of cancers. The presence of intracardiac thrombus in such cases requires further confirmation by ruling out other causes of cardiac thrombi and through molecular testing.

FOOTNOTES

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