

Li-Fraumeni syndrome: update, new data and guidelines for clinical management

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Abstract

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Authors

Thierry Frebourg, Anne Abel, Catherine Bonaiti-Pellie, Laurence Brugières, Pascaline Berthet, Brigitte Bressac-de Pallerets, Annie Chevrier, Agnès Chompret, Odile Cohen-Haguenauer, Olivier Delattre, Josué Feingold, Jean Feunteun, Didier Frappaz, Jean-Paul Fricker, Paul Gesta, Philippe Jonveaux, Chantal Kalifa, Catherine Lasset, Bruno Leheup, Jean-Marc Limacher, Michel Longy, Catherine Nogues, Daniel Oppenheim, Danièle Sommelet, Florent Soubrier, Claude Stoll, Dominique Stoppa-Lyonnet, Henri Tristant

Service de génétique, CHU et Inserm EMI 9906, Faculté de médecine et de pharmacie, 76183 Rouen.

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The Li-Fraumeni syndrome (LFS) is an inherited form of cancers, affecting children and young adults, and characterized by a wide spectrum of tumors, including soft-tissue and bone sarcomas, brain tumours, adenocortical tumours and premenopausal breast cancers. In most of the families, LFS results from germline mutations of the tumor suppressor TP53 gene encoding a transcriptional factor able to regulate cell cycle and apoptosis when DNA damage occurs. Recently, germline mutations of hCHK2, encoding a kinase, regulating cell cycle via Cdc25C and TP53, were identified in affected families. The LFS working group recommendations are the following: (i) positive testing (screening for a germline TP53 mutation in a patient with a tumor) can be offered both to children and adults in the context of genetic counseling associated to psychological support, to confirm the diagnosis of LFS on a molecular basis. This will allow to offer to the patient a regular clinical review in order to avoid a delay to the diagnosis of another tumor; (ii) the 3 indications for positive testing are : a proband with a tumor belonging to the narrow LFS spectrum and developed before age 36 and, at least, a first- or second-degree relative with a LFS spectrum tumor, before age 46, or a patient with multiple primary tumors, 2 of which belonging to the narrow LFS spectrum, the first being developed before 36 or a child with an adenocortical tumour; (iii) presymptomatic testing must be restricted to adults; (iv) the young age of onset of the LFS tumors, the prognosis of some tumors, the impossibility to ensure an efficient early detection, and the risk for mutation carriers to develop multiple primary tumors justify that prenatal diagnosis might be considered in affected families.

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li-fraumeni syndrome Inheritance: Autosomal dominant; Prevalence: 1-9/100000 (United Kingdom), 1-9/100000 (United States); Age of onset: All ages; Age of death: any age Other clinical definitions for LFS have been proposed and called Li-Fraumeni like syndrome (LFL). In these families affected relatives develop a diverse set of malignancies at unusually early ages. Four types of cancers account for 80% of tumors occurring in TP53 germline mutation carriers: breast cancers, soft tissue and bone sarcomas, brain tumors (astrocytomas) and adrenocortical carcinomas. Clinical and Molecular Studies of Li-Fraumeni Syndrome and TP53-associated Disorders. Recruiting. NCT04367246. 10. Clinical, Epidemiologic, and Genetic Studies of Li-Fraumeni Syndrome. Recruiting. Li-Fraumeni syndrome (LFS) is a rare familial cancer-predisposing syndrome, which is inherited in an autosomal dominant pattern. LFS patients often present with multiple primary tumors and an early age of onset. The lifetime risk of cancer is estimated to be 73 % for males and nearly 100 % for females. Germline mutations in the tumor suppressor gene, TP53, are associated with LFS and can be detected in more than 60 % of classic LFS families. Clinical genetic testing for TP53 is available for individuals with a suspected or known clinical diagnosis of LFS or a molecular diagnosis based on a his The National Comprehensive Cancer Network has created expert guidelines for management of cancer risk in men and women with Li-Fraumeni Syndrome. People with a TP53 mutation may also be eligible for pancreatic cancer screening clinical trials. Visit our research study page for links to clinical trials for early detection of pancreatic cancer. Reproductive options. For patients of reproductive age, advise about options for prenatal diagnosis and assisted reproduction including pre-implantation genetic diagnosis. Risk to relatives. Advise about possible inherited cancer risk to relatives, options for risk assessment, and management. Recommend genetic counseling and consideration of genetic testing for at-risk relatives. Updated 02/03/2020