

Li-Fraumeni syndrome

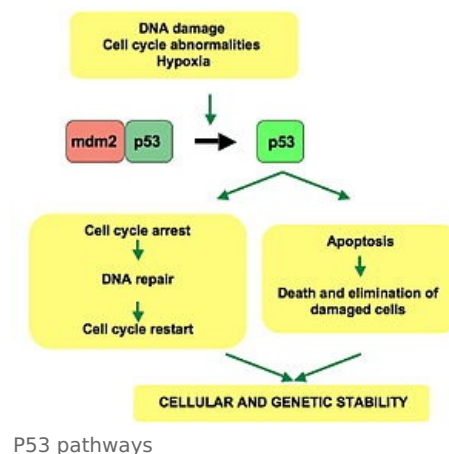
Li-Fraumeni Syndrome (OMIM: 151623) is a hereditary tumor syndrome associated with a generally increased predisposition to cancer (without clear organ specificity).

Etiology

- The cause a germline mutation tumor supressor gene *TP53* (17p13.1);
- A similar clinical picture (so-called Li-Fraumeni syndrome 2 OMIM: 609265) is caused by a mutation in the *CHEK2 gene* (22q12.1; OMIM: 604373), otherwise also associated with hereditary breast cancer.

Clinical image

- The syndrome is characterized by a highly variable phenotype:
 - osteosarcomas and soft tissue sarcomas
 - breast cancer
 - adrenal medullary adenocarcinoma
 - CNS tumors
 - leukemia
- It is usually a very early onset of cancer



Occurrence

- It is characterized by a very frequent occurrence of tumors in the family ("tumor families"). Incidence in the world is 1-9/100,000.

History

The syndrome was described by Frederick Pei Li and Joseph F. Fraumeni jr. in 1969. ^[1]

Links

Related articles

- Hereditary cancer
 - Neurofibromatosis
 - Wilms tumor

Source

- ŠÍPEK, Antonín. *Genetically determined neoplastic diseases* [online]. The last revision 8. 6. 2007, [cit. 17. 4. 2010]. <<http://www.genetika-biologie.cz/hereditarni-nadorove-syndromy>>.

1. LI, F P – FRAUMENI, J F. Soft-tissue sarcomas, breast cancer, and other neoplasms. A familial syndrome?. *Ann Intern Med* [online]. 1969, vol. 71, p. 747-52, Available from <<https://www.ncbi.nlm.nih.gov/pubmed/5360287>>. ISSN 0003-4819.

Bibliography

- KLEIBL, Zdeněk – NOVOTNÝ, Jan. *Hereditary cancer syndroms*. 1. edition. Prague : Triton, 2003. 31 pp. ISBN 80-7254-357-1.