Talk:Syndrom Li-Fraumeni

Li-Fraumeni syndrome 151623 (https://omim.org/entry/151623)) is a hereditary cancer syndrome associated with a generally increased predisposition to the development of cancer (without clear organ specificity).

Etiology

- The cause is a germline mutation tumor-supresorové gene P53|TP53 ***(17p13.1);***
- A similar clinical picture (so-called Li-Fraumeni syndrome 2, is caused by a mutation of the CHEK2 gene, otherwise also associated with hereditary breast cancer.

Clinical picture

- The syndrome is characterized by a highly variable phenotypeem:
  - osteosarkomand soft tissue sarcomasy měkkých tkání;
  - **breast cancer**;
  - adenocarcinoma of the adrenal medulla;
  - CNS tumor;
  - Leukemia.
- It is mostly a very **early onset** of cancer.

Occurence

- The very frequent occurrence of tumors in the family (" tumor families ") is characteristic..

History

The syndrome was described by Frederick Pei Li and Joseph F. Fraumeni Jr. in 1969.

[1].

Odkazy

Související články

- Hereditární nádorové syndromy
  - Neurofibromatóza
  - Wilmsův tumor

Zdroj

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Reference

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Použitá literatura

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