

Oncogenetic testing and follow-up for Li-Fraumeni syndrome

1. Diagnostic testing criteria

A person should be only offered counselling and genetic testing if he or she fulfils either the criteria for Classic Li-Fraumeni Syndrome, Li-Fraumeni Like Syndrome or the revised Chompret criteria, or for early onset breast cancer.

Classic Li-Fraumeni Syndrome (LFS)

- A proband with a sarcoma diagnosed before age 45 years,
AND
- A first-degree relative with any cancer before age 45 years,
AND
- A first- or second-degree relative with any cancer before age 45 years or a sarcoma at any age.

Li-Fraumeni Like Syndrome

Birch definition:

- A proband with any childhood cancer OR with sarcoma, brain tumour, or adrenocortical carcinoma diagnosed before age 45 years,
AND
- A first- or second-degree relative with a typical LFS cancer (sarcoma, breast cancer, brain tumour, adrenocortical carcinoma, or leukaemia) at any age,
AND
- A first- or second-degree relative with any cancer before age 60 years.

Eeles definition:

- Two first- or second-degree relatives with LFS-related malignancies at any age.

Chompret criteria

- A proband with a tumour belonging to the LFS tumour spectrum (soft tissue sarcoma, osteosarcoma, brain tumour, pre-menopausal breast cancer, adrenocortical carcinoma, leukaemia, or bronchoalveolar lung cancer) before age 46 years, AND at least one first- or second-degree relative with an LFS tumour (except breast cancer if the proband has breast cancer) before age 56 years or with multiple tumours,
OR
- A proband with multiple tumours (except multiple breast tumours), two of which belong to the LFS tumour spectrum and the first of which occurred before age 46,
OR
- A proband who is diagnosed with adrenocortical carcinoma or choroid plexus tumour, irrespective of family history.



Early onset breast cancer

- For individual with breast cancer ≤30 years with a negative BRCA1/BRCA2 test, offer a TP53 test

2. Additional recommendations

- Individual risk assessment should be done by professionals with sufficient skills and experience, and should include extensive counselling and sufficient attention to patient preferences and support.
- Discuss with the patient the possibility to perform prophylactic bilateral mastectomy. However, the patient should be informed that there is no proof that preventive measures have a benefit overall.

3. Follow-up of women at high risk

- For women with a proven TP53 mutation who opt for screening rather than for prophylactic bilateral mastectomy, yearly MRI is recommended from the age of 25 years onwards.
- Yearly mammography is not recommended because of the higher susceptibility to radiation.
- Ultrasound is useful to reduce the number of false positives when MRI is difficult to interpret.

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