Federaal Kenniscentrum voor de Gezondheidszorg
Centre Fédéral d'Expertise des Soins de Santé
Belgian Health Care Knowledge Centre

Clinical recommendations

- Family history should be evaluated using a validated prediction model (e.g. PREMM1,2,6) or the revised Bethesda criteria. Individuals considered at risk should be referred for genetic counselling. A first step may be the retrieval and immunohistochemical analysis of stored samples of family members after appropriate consent. This is possibly followed by germline mutation analysis of the referred individual.

- Investigation of all colorectal cancers by immunohistochemistry (IHC) of the four mismatch repair (MMR) proteins or by microsatellite instability (MSI) testing is recommended. In case of a positive family history (e.g. based on PREMM1,2,6) or other risk factors, both IHC and MSI should be performed if either MSI of IHC performed alone remains inconclusive.

- Immunohistochemistry and MSI tests should only be performed in laboratories that are ISO accredited for these tests.

- If the only reason for germline mutation analysis is a positive IHC for MLH1, germline mutation analysis should be accompanied by MLH1 promotor methylation or BRAF mutation analysis.

- Patients with a positive IHC or MSI result should be offered referral for genetic counselling, which may result in germline mutation analysis.

- In families with a known causal mutation, predictive testing should be offered to all relatives from the age of 18 onwards and after genetic counselling.

- In confirmed Lynch syndrome patients, yearly surveillance (including colonoscopy) is recommended. To maximally prevent the associated risk of endometrial and ovarian cancer, hysterectomy and bilateral oophorectomy is an option to be discussed with mutation carriers who have completed their families, especially after the age of 40 years. The option of surveillance for endometrial cancer should also be discussed with the patient; it should be mentioned that currently the benefit is unproven.

- In families without identified causal mutation, the decision for surveillance should be based on the family or the personal history.

- Participation of patients in the FAPA registry[a] is recommended and should be offered to patients concerned.

[a] Familial Adenomatous Polyposis Association

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