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

Oncogenetic testing for persons with Multiple Endocrine Neoplasia type 1 (MEN1)

Multiple Endocrine Neoplasia type 1 (MEN1) is a polyglandular genetic syndrome characterized by tumours of the parathyroid glands, pancreatic islet cells and/or anterior pituitary gland. Parathyroid tumours with primary hyperparathyroidism is the most common presentation (95% of all MEN1 cases). In addition to these three 'major' locations, tumours can also occur in 'minor' locations, such as the adrenal cortex. MEN1 is usually inherited (as an autosomal dominant disorder), but *de novo* mutations of the menin gene associated with MEN1 are found in about 10% of patients.
Epidemiological data are not available for Belgium, but the incidence has been estimated to be 0.25% from post mortem studies.

Criteria for clinical suspicion of MEN1

- One of the three major MEN1-associated tumours in combination with one minor MEN1-associated tumour (adrenocortical tumours or neuroendocrine tumours of the stomach, lungs or thymus).
- Multiple MEN1-associated tumours in one organ.
- One MEN1-associated tumour at an age < 35 years and a family member with a different MEN1-associated tumour.

Criteria for clinical diagnosis of MEN1

- At least two of the three major MEN1-associated tumours (parathyroid tumours, neuroendocrine tumours of pancreas/duodenum, anterior pituitary tumours).
- One of the three major MEN1-associated tumours in a first-degree relative of a case with a clinical diagnosis of MEN1.

Clinical Recommendations

- Pre- and post-test genetic counselling should be offered to all patients with a clinical diagnosis or suspicion of MEN1 (strong recommendation)
- All patients with a clinical diagnosis of MEN1 should be offered MEN1 genetic testing (strong recommendation)
- In patients with a clinical suspicion of MEN1, MEN1 genetic testing may be considered (weak recommendation)
- MEN1 mutation analysis should be offered to all first-degree relatives of MEN1 mutation carriers* (strong recommendation)

** Or first-degree relatives of patients with clinical MEN1 who died before genetic testing was carried out.*

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