Genetic testing in primary hyperparathyroidism

Peter Vestergaard
Bente Lomholt Langdahl
Leif Mosekilde

Genetic testing should be performed if:

1) In the individual patients more than one endocrine neoplasia is present (e.g. primary hyperparathyroidism plus medullary thyroid carcinoma or phaeochromocytoma or Marfanoid phenotype in multiple endocrine neoplasia type 2, primary hyperparathyroidism plus pituitary tumors or neuroendocrine tumors in multiple endocrine neoplasia type 1) [1-3]
2) If more than one patient in a family has primary hyperparathyroidism (RET proto-oncogene, MENIN gene, HRPT2 gene) [2-4]
3) In very young patients, especially in the presence of hyperplasia of the parathyroid glands, although RET proto-oncogene mutations may be associated with single adenomas [5].

If a patient has undergone parathyroid surgery and calcium continues to be increased despite seemingly succesful therapy or if urine calcium/creatinine clearance is very low (<0.02), then CaSR mutation analysis should be performed [6].

Bibliography

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