

Case Report

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Case Presentation MEN2A

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SUMMARY

Multiple endocrine neoplasia type 2 (MEN2) is an autosomal dominant disorder that can be distinguished as three different syndromes: multiple endocrine neoplasia type 2A (MEN2A), MEN2B and familial medullary thyroid carcinoma (FMTC). This disorder is usually caused by the mutations of the rearranged during transfection protooncogene gene (RET) or the neurotrophic tyrosine kinase receptor type 1 gene (NTRK1).

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Introduction

Mutations in the E7 proto-oncogene on chromosome 10 have been identified in MEN 2A, MEN 2B and familial medullary thyroid carcinoma. The RET protein is a receptor tyrosine kinase. MEN 2A and familial medullary thyroid carcinoma mutations result in activation of certain intracellular pathways.

	Risk of Medullary thyroid carcinoma	Pheochromocytoma	Primary hyperparathyroidism	Other extra endocrine signs
MEN2A	80-100%	0-50% (risk depends on genotype)	0-20% (risk depends on genotype)	10% (proliferating disease, Cutaneous (hirsutism))
MEN2B (MUT, RET, R463G mutation)	100%	100% (risk depends on genotype)	0%	100% (facial freckles, skeletal manifestations, Marfan syndrome)

Background

MEN2A is subdivided into four phenotypes:

1. Classical MEN2AA family (or individual) with at least two MEN2A clinical features: MTC, PHEO, and/or HPT (includes: hyperparathyroidism, parathyroid adenoma, or parathyroid hyperplasia).
2. MEN2A with CLAClassical MEN2A family (or individual) that also developed cutaneous lichen amyloidosis (CLA).
3. MEN2A with HSCRCClassical MEN2A family (or individual) that also developed Hirschsprung's disease (HSCR).
4. FMTCFMTC (familial MTC) is used when four or more family members had MTC only and no clinical evidence of PHEO, HPT, or any MEN28-specific non-endocrine clinical features in affected or at-risk family members. Note, FMTC may be a disease variant of classical MEN2A with decreased penetrance of PHEO and HPT.

Case report

34years old female patient referred from surgery clinic after thyroidectomy due to thyroid nodules found to have medullary thyroid carcinoma, for follow up, during investigations found to have high calcitonin, pan CT for staging done. Showed bilateral adrenal adenomas. While bp is normal, and no palpitations. 24 hour urine for catecholamines done, found to have 3 times upper normal. Physical examination was normal- no nevi or other features. After discussion with patient and her family decision made to do bilateral adrenalectomy. Patient started preparation with phenoxybenzamine, then after 2 weeks of medicine, she underwent surgery smoothly without complications. Histopathology came with diagnosis bilateral pheochromocytoma, Hormonal replacement with thyroxine and steroids, fludrocortisone started.

RET protooncogene requested for her came positive, Then requested for her kids found 3 out of 5 kids positive. The decision to do total thyroidectomy for those who had positive RET mutation [1-4].

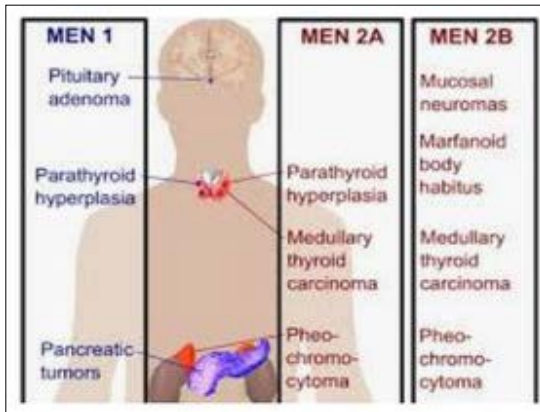
Images



Recommendations

- Identify the germline RET mutations in the etiology of multiple endocrine neoplasia type 2
- Explain the epidemiology of multiple endocrine neoplasia type 2.
- Review the use of total prophylactic thyroidectomy and lymph node dissection in the management of multiple endocrine neoplasia type 2.

- Summarize the importance of collaboration and communication among the interprofessional team to provide counseling and genetic testing to first degree relatives of those affected with multiple endocrine neoplasia type.



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