Multiple Endocrine Neoplasia, Type I

Alternative Names
MEN1
MEN I
Endocrine Adenomatosis, Multiple
MEA I
Wermer Syndrome

Record Category
Disease phenotype

WHO-ICD
Neoplasms > Neoplasms of uncertain or unknown behaviour

Incidence per 100,000 Live Births
2-5

OMIM Number
131100

Mode of Inheritance
Autosomal dominant

Gene Map Locus
11q13.1

Description
Multiple endocrine neoplasia Type 1 (MEN1) is a rare autosomal dominantly inherited form of cancer. It is characterized by the development of neuroendocrine tumors of the parathyroid, pancreas, and anterior pituitary gland. However, it is less commonly associated with cancer of the adrenal cortical gland. Some patients with MEN1 may have tumors not related to the adrenal glands. Affected individuals present with primary hyperparathyroidism (90%) followed by pancreatic neuroendocrine tumour (35-75%). Other signs include hypercalcemia and hypophosphatemia. The disease affects both sexes equally, and the penetrance is 94% by the age of 50.

The diagnosis is based on the presence of two endocrine tumors in addition to biochemical and genetic testing. Annual screening is recommended for the patients and at-risk family members. Treatment approach is oriented to a specific tumor that present in affected individual.

Molecular Genetics
MEN1 is caused by inactivating mutations in the MEN1 gene which is located on the long arm of chromosome 11. MEN1 gene encodes the menin protein, which is functioning as a tumor suppressor. This protein is widely expressed in endocrine and nonendocrine tissues. Loss of function of menin leads to abnormal cell division and cancer development in sites where this protein is abundant. Several mutations have been reported in the MEN1 gene.

Epidemiology in the Arab World

Saudi Arabia
Raef et al. (2011) described a large Saudi family with MEN1. Seven out of ten siblings presented with hyperparathyroidism, pituitary adenomas, adrenal adenomas and pancreatic endocrine tumours. Parents were non consanguineous. The father was affected with adrenal tumours while the mother and three siblings of the affected patients were normal. Raef et al. (2011) identified a novel monoallelic deletion of 5 kb genomic DNA involving the promoter and exons 1 and 2 of MEN1 gene. A long-term periodic assessment was recommended for the positive cases.

References
Raef H, Zou M, Baitei EY, Al-Rijjal RA, Kaya N, Al-Hamed M, Monies D, Abu-Dheim NN, Al-Hindi H, Al-Ghamdi MH, Meyer BF, Shi Y. A novel deletion of the MEN1 gene in a large family of multiple endocrine neoplasia type 1 (MEN1) with aggressive phenotype. Clin Endocrinol (Oxf). 2011; 75(6):791-800. PMID:21627674

Related CTGA Records
MEN1 Gene

External Links
http://emedicine.medscape.com/article/126438-overview
http://www.cancer.net/cancer-types/multiple-endocrine-neoplasia-type-1

https://www.niddk.nih.gov/health-information/health-topics/endocrine/multiple-endocrine-neoplasia-type-1/Pages/fact-sheet.aspx
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=652

Contributors

Ameera Balobaid: 10.06.2016