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Retinal atrophy and MEN1 syndrome: case report

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Abstract:

In this study, we present the case of a patient with multiple endocrine neoplasia type 1 (MEN1) syndrome. The patient presented with primary hyperparathyroidism (pHPT) which was associated with a microadenome in the right pole. This case presents additional complications related to an extensive medical history of eye diseases and cancers in the family. Family medical history includes multiple cases of hyperparathyroidism and degenerative eye disease, as well as a previous death in the family due to metastases from another abdominal cancer.

Keywords: Retinal atrophy, MEN1, HPT, Cancer.

INTRODUCTION

MEN1 syndrome is a rare genetic disorder characterized by the development of multiple endocrine tumors, leading to serious hormonal disturbances. MEN1 is characterized by hyperparathyroidism, pancreatic adenomas, and pituitary adenomas. Here we present a complex case of a patient presenting with various complications including a family history of cancer and visual disturbances.

View case

Medical history: We had a 43-year-old female patient with MEN1 syndrome. The patient suffers from primary hyperparathyroidism (pHPT) associated with the presence of a small adenoma in the right pole of the gland. The patient had undergone treatment for strabismus two years ago. But what is interesting about the case is the medical history of this family

Family medical history:

Two sisters suffered from hyperparathyroidism (HPT) in their youth. A sister died at the age of 35 from peritoneal carcinoma that was the result of another abdominal cancer. Two sisters and a brother suffer from retinal degeneration

Association of MEN1 and retinal dystrophy: MEN1 causes endocrine neoplastic developments, and there are no direct data linking it to retinal degeneration. The MEN1 gene is known to be a tumor suppressor gene, as the Menin protein regulates the cell cycle and suppresses tumors. Possible association between hereditary tumors and retinal disorders: Gene mutations affecting different proteins may play a role in both conditions, but there are no direct data suggesting a relationship between MEN1 and retinal dystrophy.

The role of Menin protein in the structure and function of the retina is as follows:

- Gene regulation and gene expression Menin acts as a gene regulator by interacting with chromatin and other proteins. This interaction can affect gene expression of important proteins in retinal cells. It helps regulate transcription factors such as the MLL (Mixed-Lineage Leukemia) complex, which can affect the differentiation and development of retinal cells (1).

- Cell cycle and differentiation Menin plays a role in regulating the cell cycle by inhibiting or activating certain genes, contributing to reticulocyte differentiation and avoiding abnormal growth or tumors. By affecting cell signaling pathways such as TGF-β pathway and Wnt pathway, it can affect cell survival and differentiation (2,3).

- Neurological function and nerve cell support Menin is important in maintaining the normal function of nerve cells, including those in the retina. Some suggest that it may have a role in supporting nerve health and maintaining retinal cell homeostasis, which can influence the retina's response to light signals.

- Protection from oxidative stress Menin contributes to the regulation of the response to oxidative stress, which can affect retinal cells that are regularly exposed to metabolic stress resulting from light signals. Impaired Menin function may increase the susceptibility of retinal cells to oxidative damage, which may contribute to the development of degenerative retinal diseases.

Possible mechanisms:

* Interaction with growth factors: Menin protein interacts with growth factors such as insulin-like growth factor (IGF-1) and other growth factors that play a role in retinal development.
* Interaction with chromatin: May affect gene regulation by interacting with chromatin proteins that modulate gene expression of essential proteins in the retina.

Conclusion: Menin protein plays an important role in maintaining retinal health and function by regulating gene expression, supporting neuronal differentiation, and protecting it from oxidative stress. A defect in this protein can lead to disorders in the growth and function of retinal cells, which may affect the structure of the retina and contribute to the development of degenerative diseases in it.

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