Neurofibromatosis Type 2

Neurofibromatosis type II is an autosomal-dominant disorder associated with various neoplasms of the central nervous system. It is caused by a mutation in the NF2 gene, located on chromosome 22q12. The most typical finding is bilateral vestibular schwannomas, which are benign tumors involving cranial nerve VIII. Other findings include multiple meningiomas and ependymomas, which can be intracranial or spinal in location. Patients also demonstrate café au lait spots and cataracts.

PLAY PICMONIC

Pathophysiology

Autosomal-Dominant

Domino

This disease is inherited in an autosomal dominant fashion.

Chromosome 22

(22) Chrome Chromosome

NF2 is caused by a mutation in the NF2 gene, located on chromosome 22q12.

Signs and Symptoms

Vestibular Schwannoma

Vest-bull-headphones on Swan-gnome

The most characteristic manifestation of the disease is the development of symmetric cranial nerve VIII tumors called vestibular schwannomas, also known as acoustic neuromas. These present with the progressive loss of hearing and balance.

Meningioma

Men-in-jeans

Meningiomas are brain tumors that arise from the meninges. They are typically benign but have a higher incidence of malignant transformation in NF2 patients. Their presentation depends on their location as they can occur in a wide variety of places within the brain and throughout the central nervous system.

Ependymoma

Panda-mummy

Ependymoma is a tumor that arises from the ependymal cells in the brain. It presents most often in the 4th ventricle in children and spinal canal in adults with symptoms related to hydrocephalus such as nausea and vomiting.

Cafe Au Lait Spots

Coffee-cow

Cafe au lait spots are hyperpigmented macules seen in NF2 and various other genetic syndromes including neurofibromatosis type 1. People with NF2 usually have fewer café au lait spots than people with NF1.

Cataracts

Cadillac-cataracts

Clouding of the lens of the eye that commonly occurs in neurofibromatosis type II.