Neurofibromatosis

Introduction

- Neurofibromatosis (NF) is a genetic disease that causes tumors to develop in the nervous system. There are three types of neurofibromatosis that are each associated with unique signs and symptoms
- Neurofibromatosis type 1 (NF1) causes skin changes (cafe-au-lait spots, freckling in armpit and groin area); bone abnormalities; optic gliomas; and tumors on the nerve tissue or under the skin. Signs and symptoms are usually present at birth.
- Neurofibromatosis type 2 (NF2) causes acoustic neuromas; hearing loss; ringing in the ears; poor balance; brain and/or spinal tumors; and cataracts at a young age. It often starts in the teen years.
- Schwannomatosis causes schwannomas, pain, numbness, and weakness. It is the rarest type.

Neurofibromatosis

Clinical manifestation /Symptoms

Neurofibromatosis I

•The classic symptom of NF I is light brown patches of pigment on the skin, called café-au-lait spots.

•Neurofibromas are often found growing on the nerves and in various organs of the person's body.

•Lisch nodules, which are small tumors on the iris (colored part of the eye), may appear around adolescence, but usually do not cause problems.

•Hearing loss, headaches, seizures, scoliosis, and facial pain or numbness may also be present.

•Mental retardation is present in 2 to 5 percent of individuals with neurofibromatosis I, while other persons affected may have learning problems and hyperactivity.

Neurofibromatosis

Clinical manifestation /Symptoms

Neurofibromatosis II

•Tumors on the eighth cranial nerve, which can lead to hearing loss, headaches, problems with facial movements, problems with balance, and difficulty walking.

•Hearing loss may be noted as early as the teenage years.

•Other clinical signs of NF II may include seizures, neurofibromas (skin nodules), and café-au-lait spots (although this is not as common as in NF I).

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Causes

❖Neurofibromatosis is caused by genetic defects (mutations) that either are passed on by a parent or occur spontaneously at conception. The specific genes involved depend on the type of neurofibromatosis

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Inheritance Pattern

❖All three types of NF are inherited in an autosomal dominant manner. Other cases may result from new (de novo) mutations in the gene. These cases occur in people with no history of the disorder in their family

Neurofibromatosis

Gene Panel

NF1, NF2, SPRED1

No of Genes : 3
Sample Type : EDTA-blood sample - 4 ml
TAT : 6 Weeks
Methodology : NGS

Diagnostic Tests

anderson
Clinical Genetics

Management

Neurofibromatosis

Reference

https://rarediseases.info.nih.gov/diseases/10420/neurofibromatosis

https://rarediseases.org/rare-diseases/neurofibromatosis-type-1-nf1/

https://stanfordhealthcare.org/medical-conditions/brain-and-nerves/neurofibromatosis/symptoms.html