Neurofibromatosis

Neurofibromatosis (NFT) is the most common of Neurocutaneous disorders.

Classification

There are as many as 6 distinct types, the two most common of which are:

Neurofibromatosis type 1

Neurofibromatosis type 2

Associated conditions

1. Schwann-cell tumors on any nerve (but bilateral VSs are virtually nonexistent)

2. spinal and/or peripheral nerve neurofibromas

3. multiple skin neurofibromas

4. aqueductal stenosis

5. macrocephaly: secondary to aqueductal stenosis and hydrocephalus, increased cerebral white matter 6. intracranial tumors: hemispheric astrocytomas are the most common, solitary or multicentric meningiomas (usually in adults). Gliomas associated with NF1 are usually pilocytic astrocytomas. Brainstem astrocytomas include both contrast-enhancing pilocytic lesions and those that are non-enhancing and radiologically diffuse

7. unilateral defect in superior orbit → pulsatile exophthalmos

8. neurologic or cognitive impairment: 30–60% have mild learning disabilities

9. see Spinal Deformity in Neurofibromatosis

10. visceral manifestations from the involvement of autonomic nerves organ glia within the organ.Up to 10% of patients have abnormal gastrointestinal motility/neuronal intestinal dysplasia related to neuronal hyperplasia within submucosal plexus

11. ≈20% develop plexiform neurofibromas: tumors from multiple nerve fascicles that grow along the length of the nerve. Almost pathognomonic for NF1

12. syringomyelia

13. malignant tumors that have increased frequency in NFT: neuroblastoma, ganglioglioma,sarcoma, leukemia, Wilm’s tumor, breast cancer

14. pheochromocytoma: is occasionally present