Neurofibromatosis Type 1

- Von Recklinghausen disease
- Peripheral neurofibromatosis
- ~ 50% have macrocephaly; in part 2° ↑ WM volume
- OPG can cause progressive vision loss
- Café au lait spots are earliest finding
- Most common neurocutaneous and inherited tumor syndrome

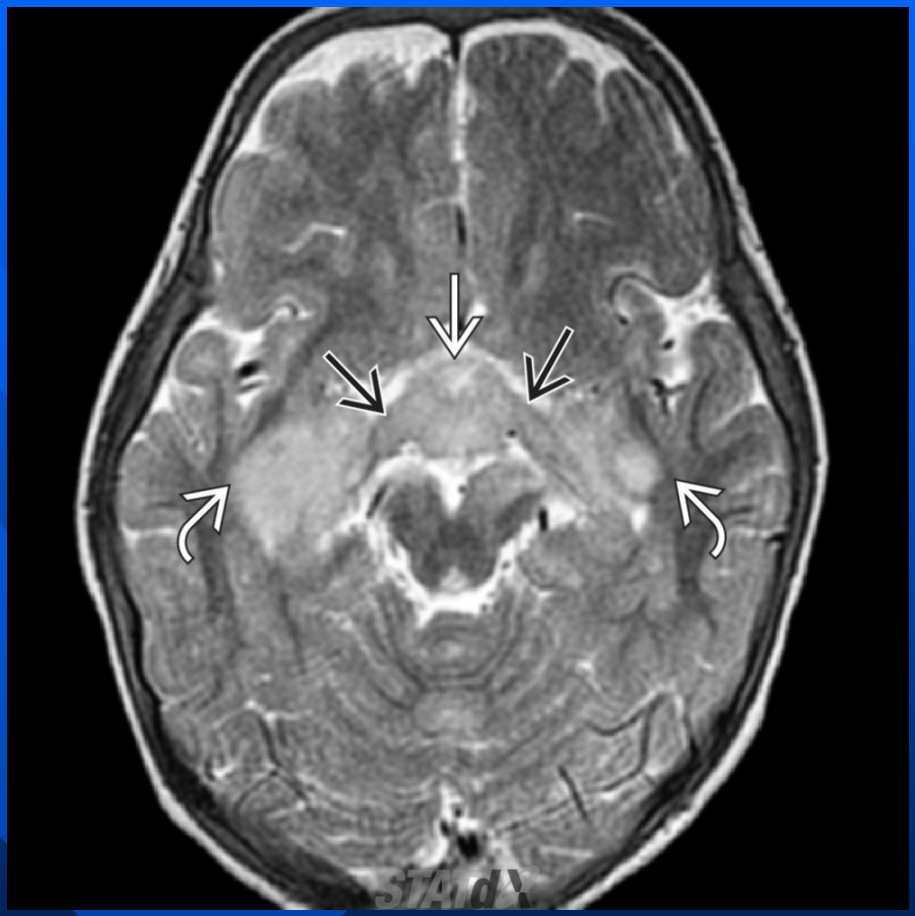
NF 1

- Autosomal dominant; gene locus is chromosome 17q12
- Best diagnostic clue
  - FASIs on T2WI in 70-90% of preteen children
  - Plexiform neurofibromas
  - Optic pathway gliomas

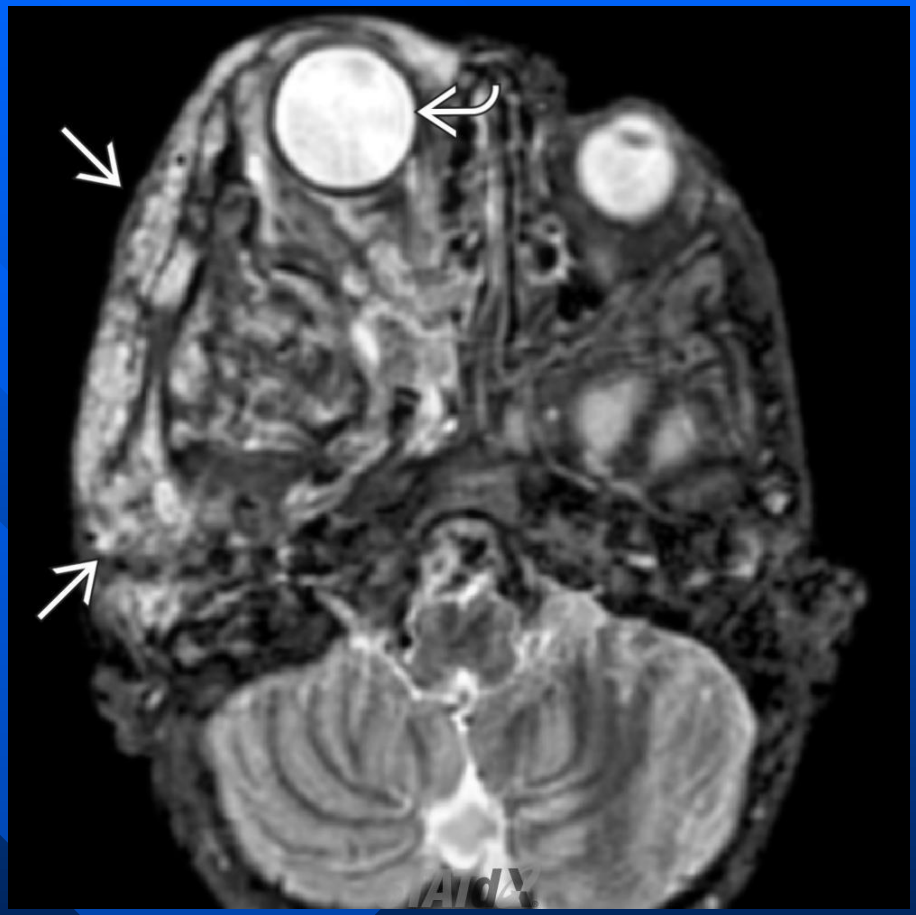
Imaging

- FASI (focal areas of signal intensity) on T2WI in 70-90% of preteen children. Dysplastic Myelin
- Plexiform neurofibromas
- Optic pathway gliomas
- Parenchymal gliomas
- Sphenoid wing and occipital bone dysplasia found in association with plexiform tumors
- WM lesions may also involve cerebellar white matter, globus pallidus, thalamus, brainstem
- WM lesions are hyperintense and typically poorly defined; no mass effect
- Vascular dysplasias → stenosis, moyamoya, aneurysm

Axial T2WI MR shows an optic pathway glioma infiltrating the chiasm (white solid arrow), the optic tracts (black solid arrow) within the suprasellar cistern, and the medial temporal lobes (white curved arrow). The tumor was identified at the time of a screening MR exam of the brain of this 21-month-old infant with newly diagnosed NF1.

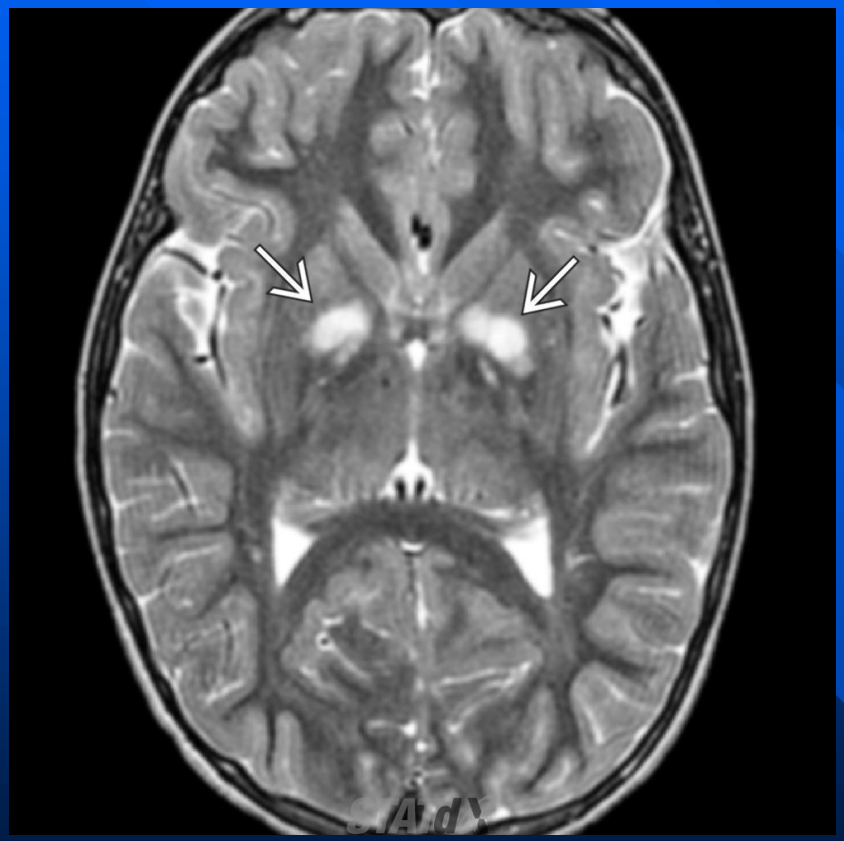


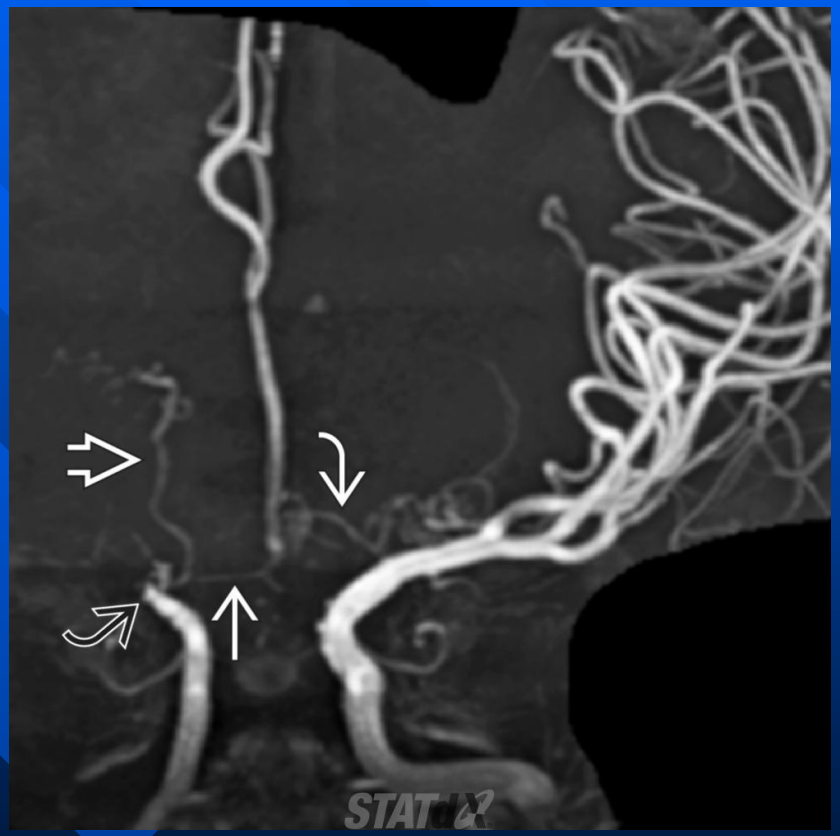
Axial T2WI FS MR shows an extensive plexiform neurofibroma (white solid arrow) of the right orbit and temporal region.
The right sphenoid wing is eroded, with resulting exophthalmoenlarged (bupthlalmos).
The right globe (white curved arrow)
The affected right globe is enlarged (bupthlalmos).



Axial T2WI FS MR shows an extensive plexiform neurofibroma (white solid arrow) of the right orbit and temporal region. The right sphenoid wing is eroded, with resulting exophthalmos of the right globe (white curved arrow). The affected right globe is enlarged (bupthlalmos).

NF 1





NF1 (myelin vacuolization)