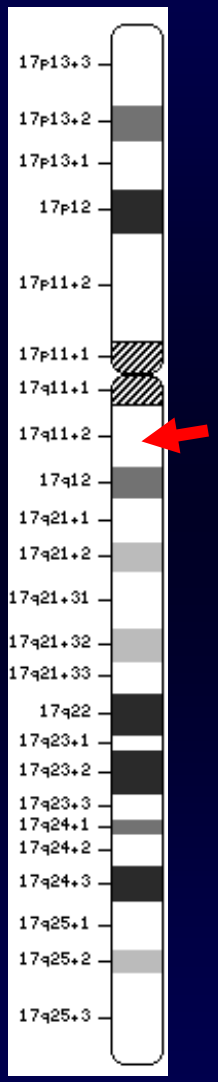
Neurofibromatosis (NF1)



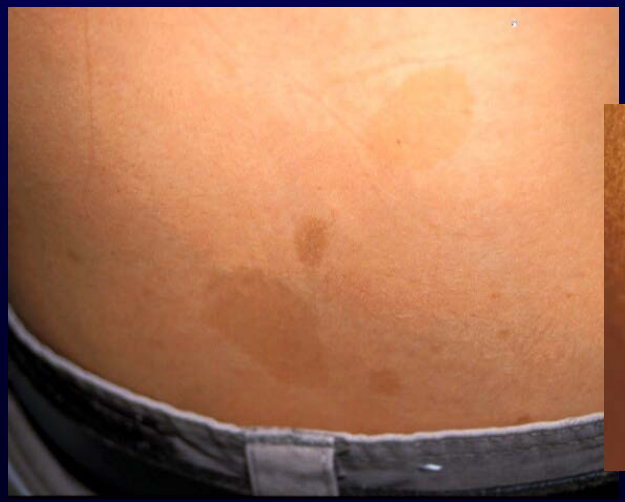
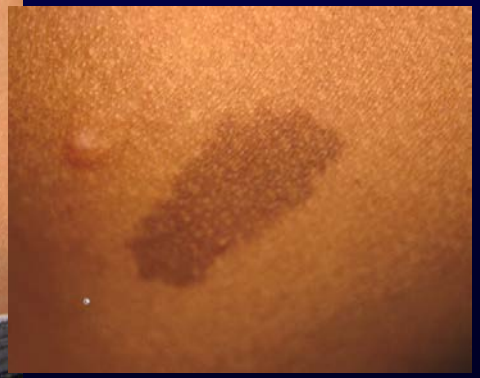
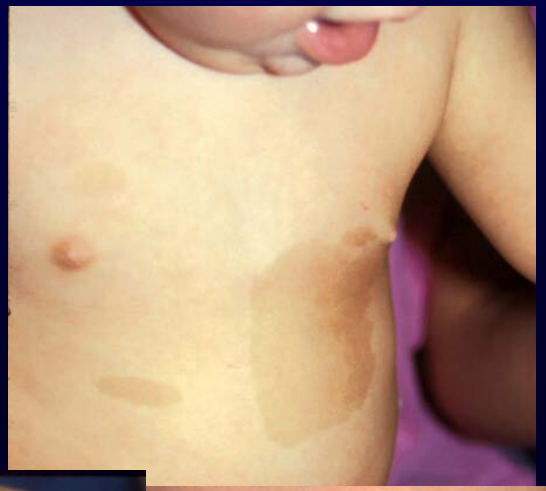
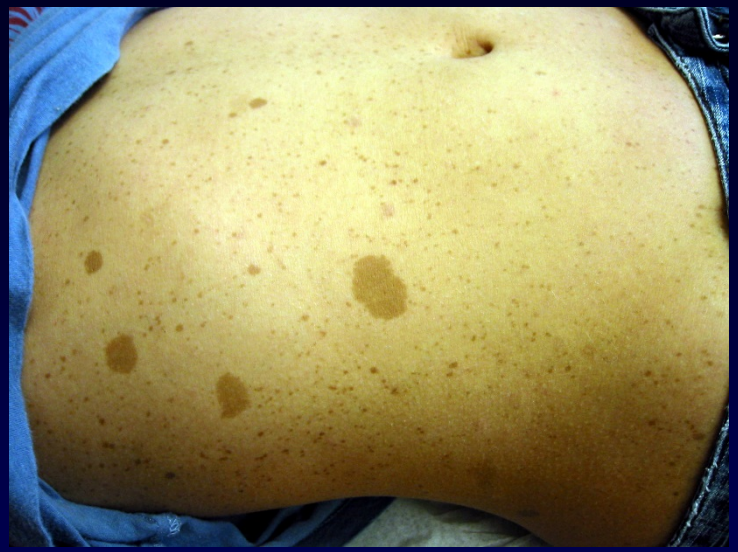
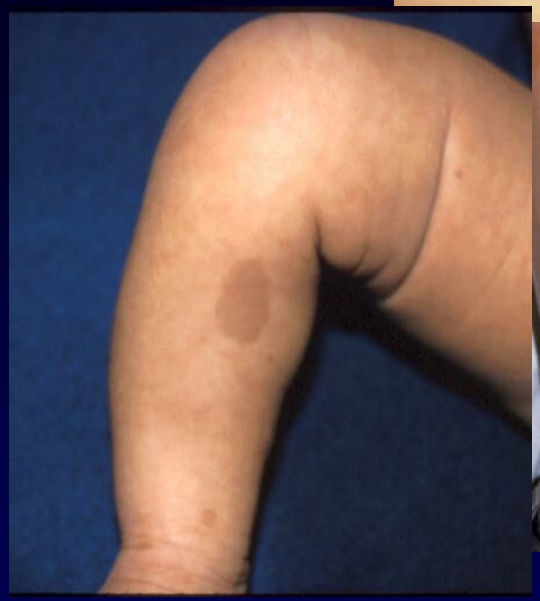
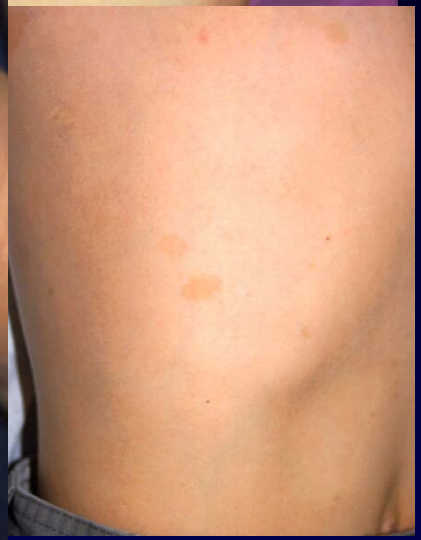
- 1/3000 incidence
- Autosomal dominant inheritance
- Gene on chromosome 17
- Mutations occur throughout the gene
- Protein (neurofibromin) has tumor suppressor effects

NF Diagnostic Criteria: 2 or more

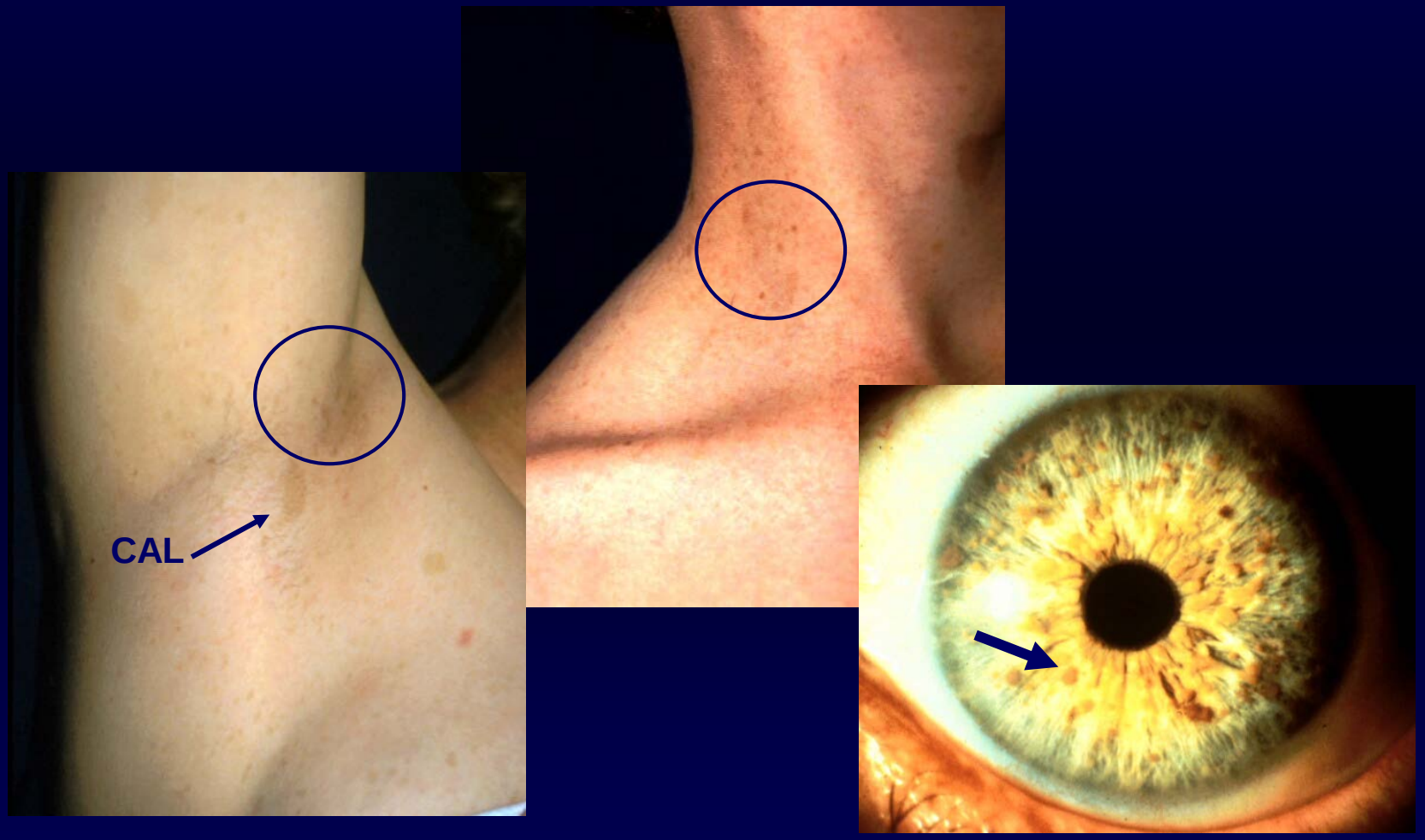
- ≥ 6 café-au-lait spots
  - 1.5 cm or larger, postpubertal
  - 0.5 cm or larger, prepubertal
- Freckling: axilla or groin
- ≥ 2 Lisch nodules
- ≥ 2 neurofibromas or ≥ 1 plexiform
- Optic glioma: nerve or pathway
- A distinctive bony lesion
  - sphenoid wing aplasia
  - tibial pseudoarthrosis
- 1^{o} relative with NF1

NFF, 1997

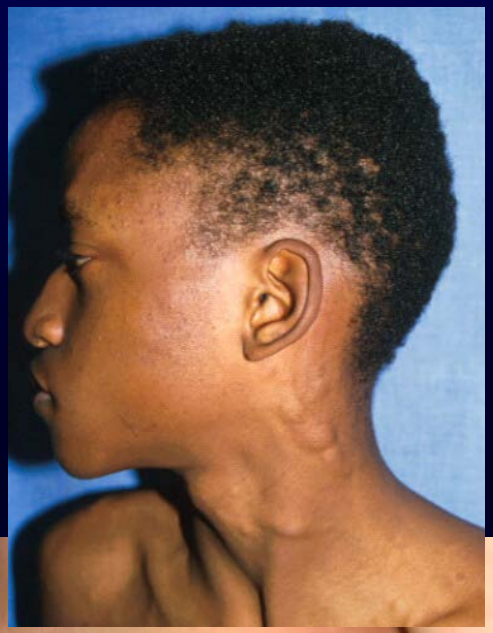
Café-au-lait spots in NF1

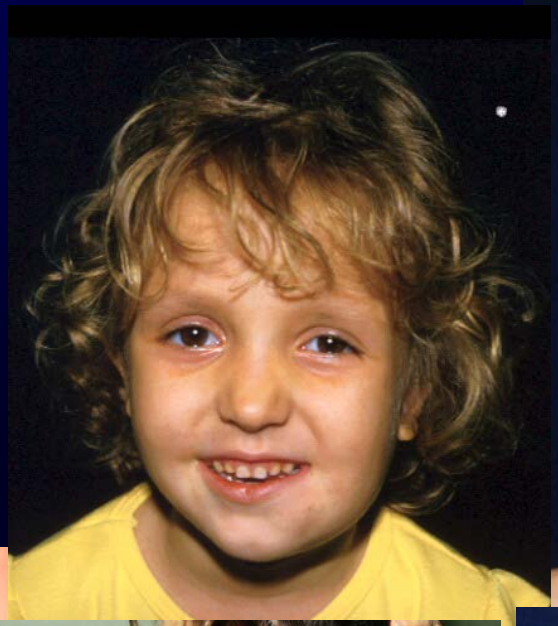







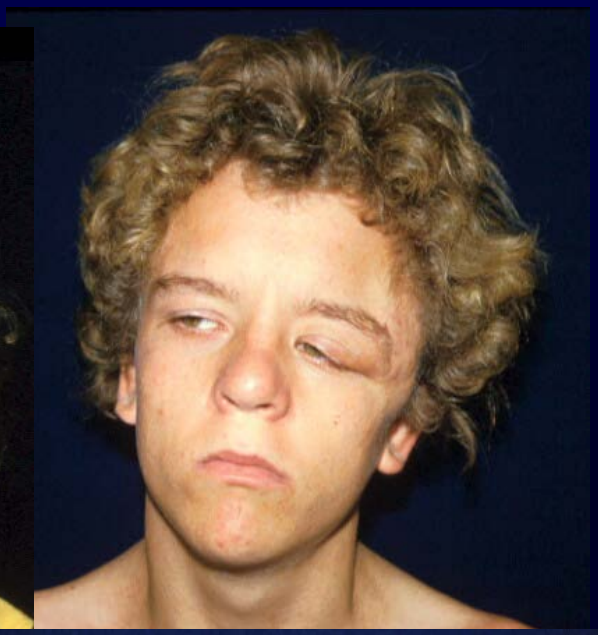
Skin freckles and Lisch Nodes: NF1

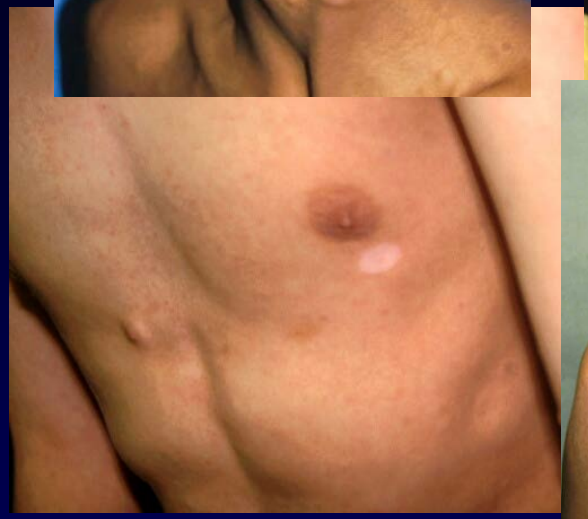


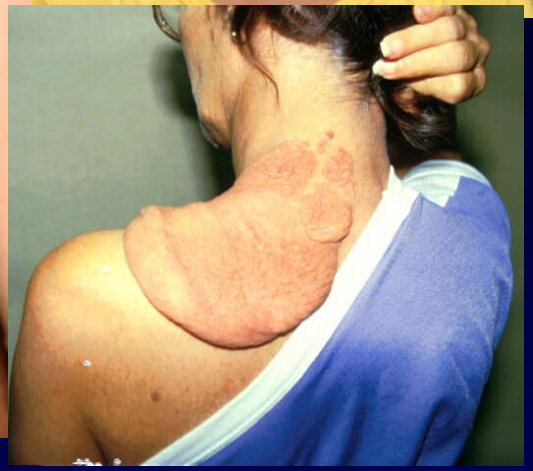
Neurofibromas in NF1

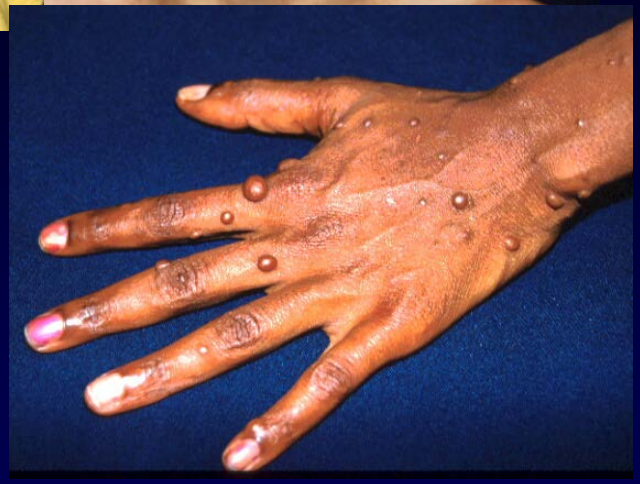








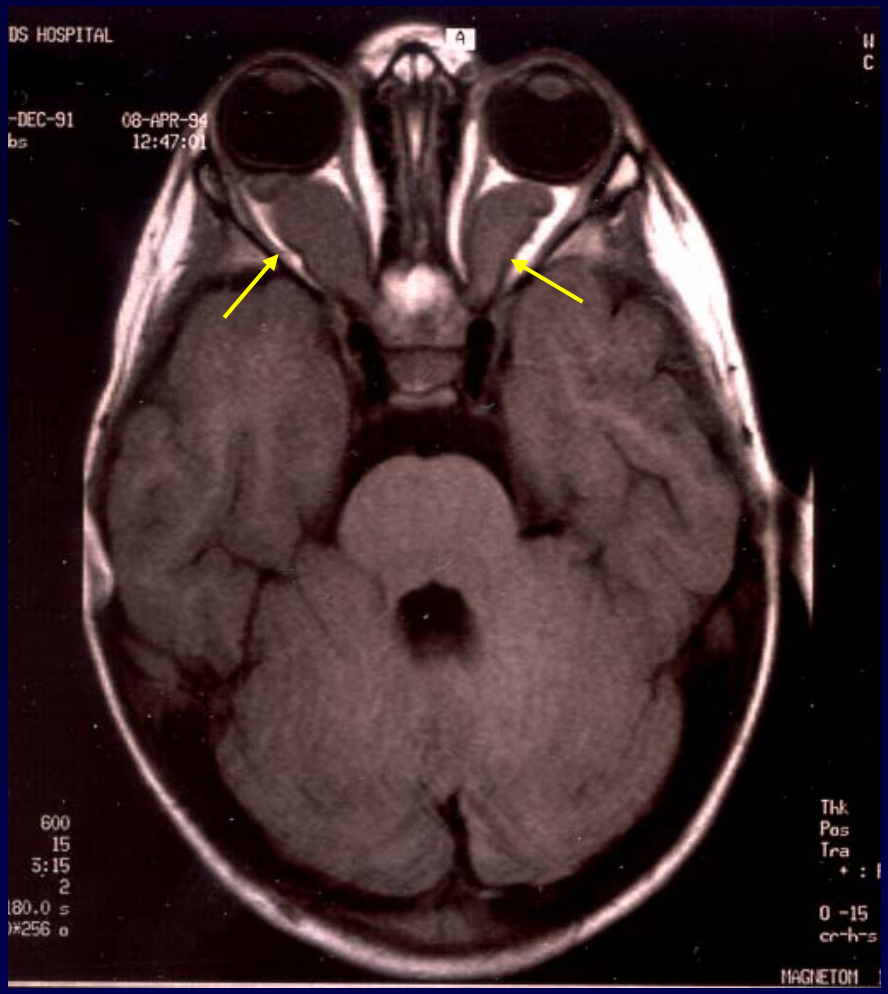


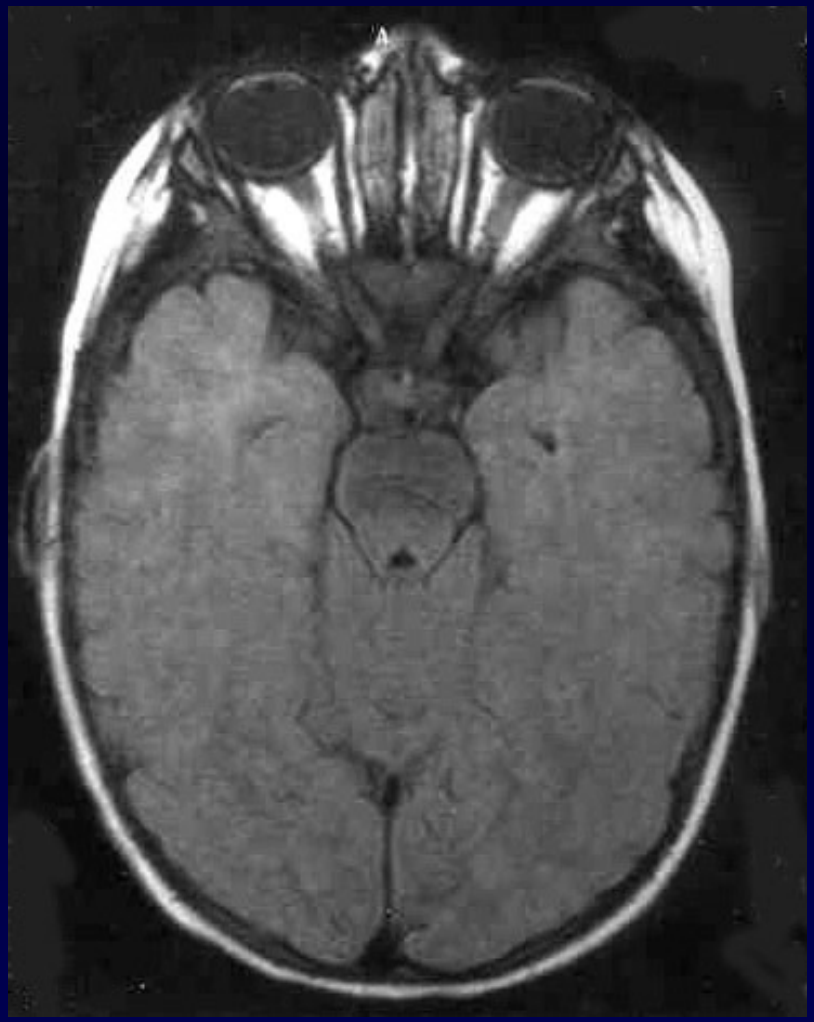


Brain Findings in NF1

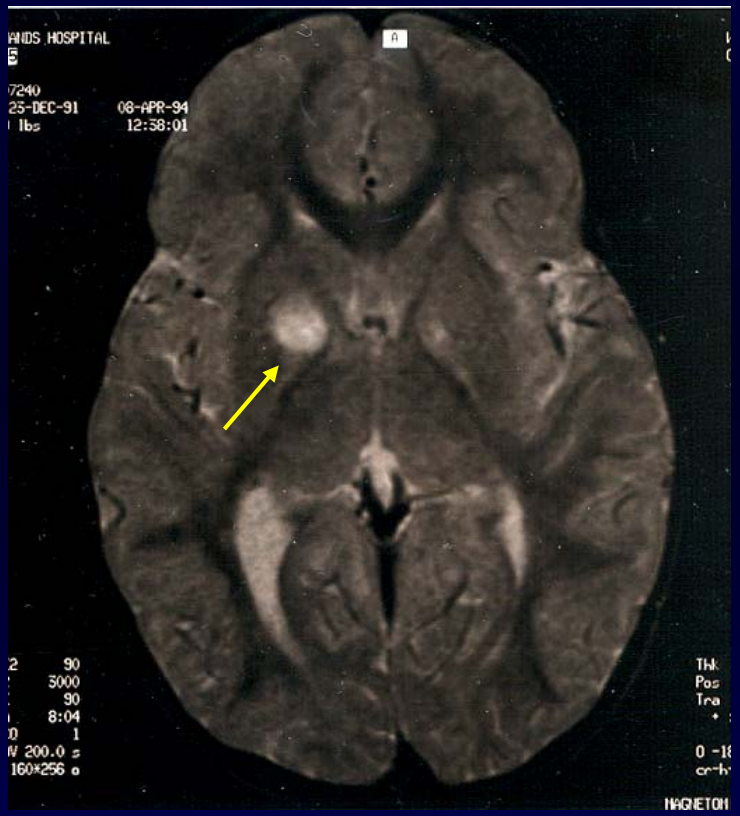
- Optic nerve glioma
- Generalized macrocephaly
- Bright spots in mid-brain, brainstem and cerebellum
- Learning disabilities
- Precocious puberty

NF1: optic glioma





T2 Signals on Brain MRI



- 43-79 % prevalence
- Histology:
  - Intramyelinic edema
  - glial cell dysplasia and hyperplasia
- Associations:
  - Not with macrocephaly
  - With cognitive/learning problems

North K, Am J Med Genet, 2000

NF1 Learning Disabilities

- 30-60% have problems
- IQ usually normal
- Cognitive deficits involve:
  - School performance
  - Visual-spatial ability
  - Executive function
  - Expressive and receptive language
  - Attention skills (e.g., ADHD)