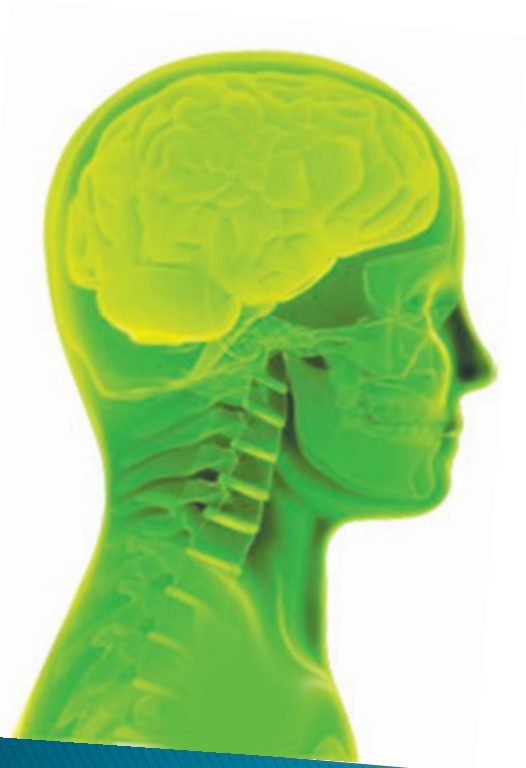
Weill Cornell Medical College

NewYork-Presbyterian
Phyllis and David Komansky
Center for Children's Health

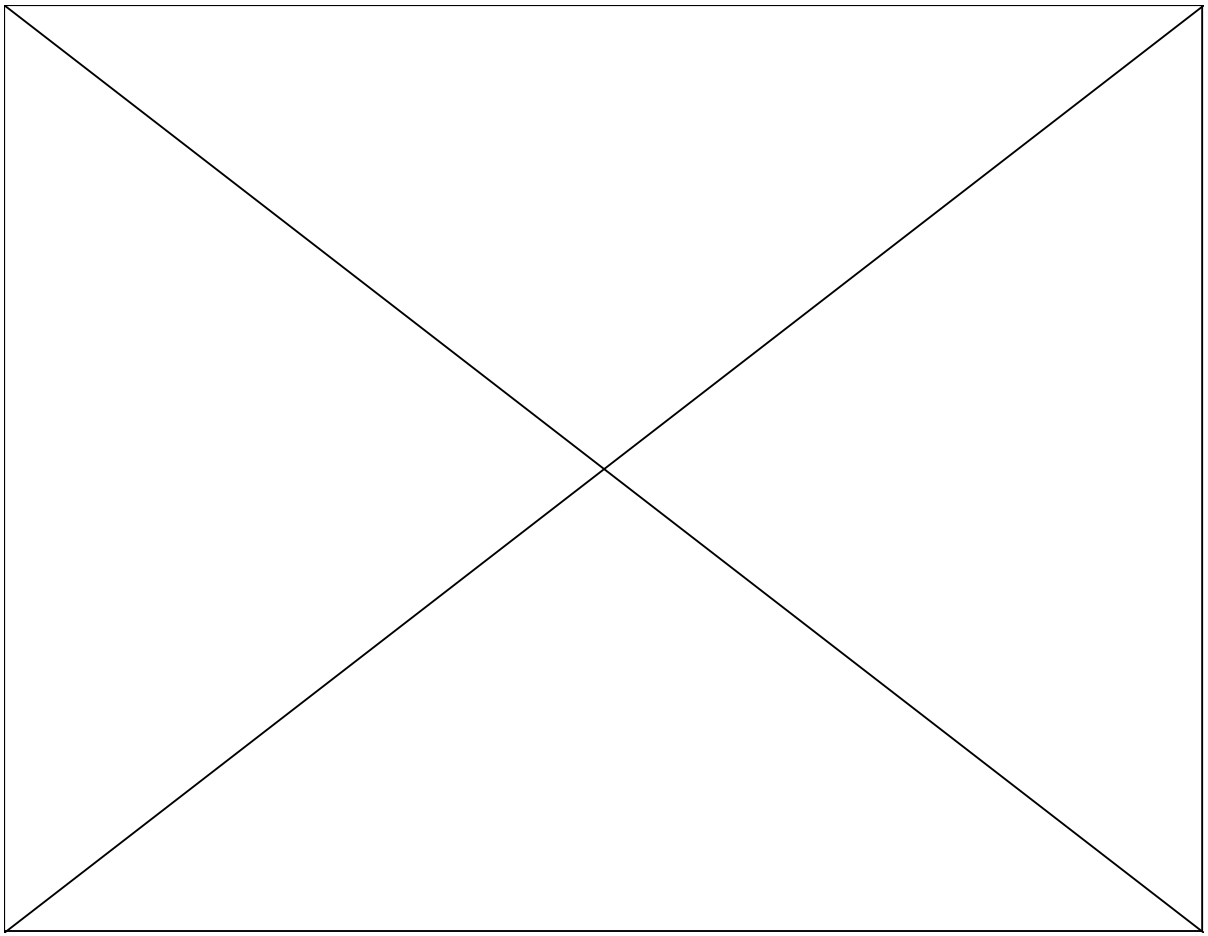
Pediatric
Brain and Spine Center

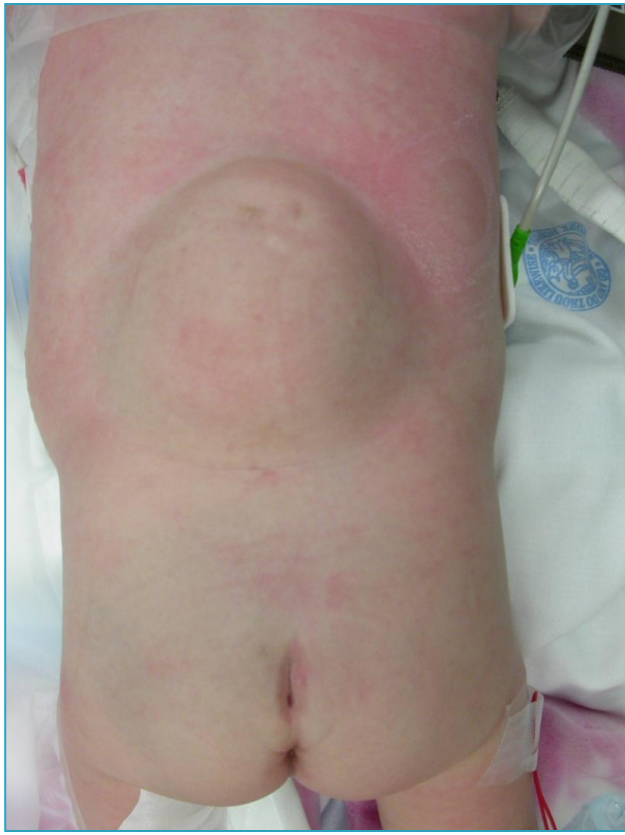


Cutaneous markings, detecting occult spinal dysraphism during the newborn exam and common neurocutaneous disorders in the office setting

Mark Souweidane, MD
Kaleb Yohay, MD

Question



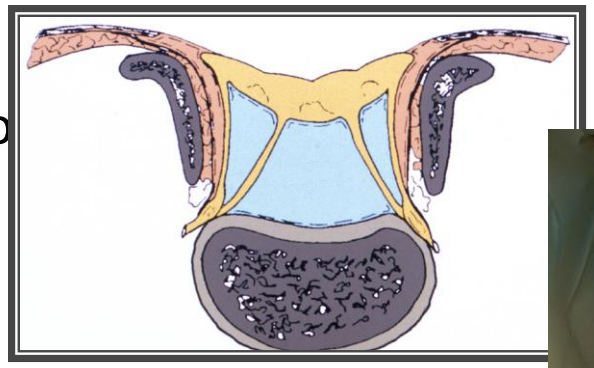


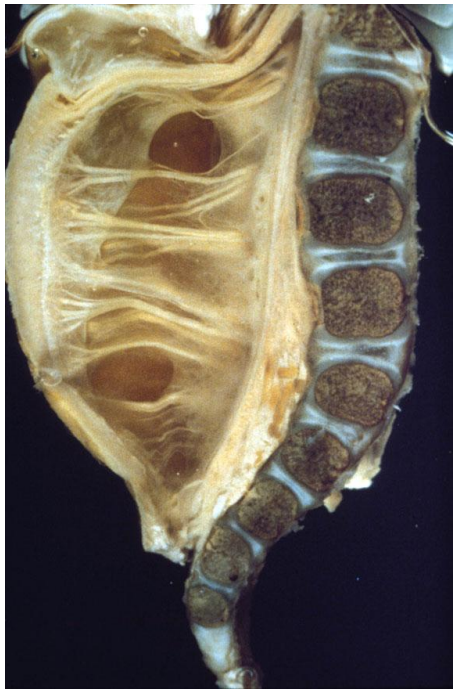
Spinal Dysraphism

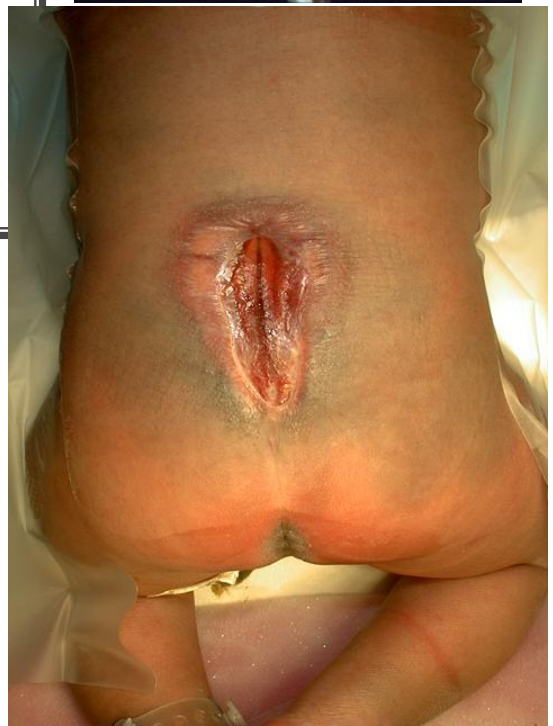
- Dysraphism (midline fusion defects)
- Neural Tube Defect (NTD)
- Occurs at variable rates
  - Overall incidence (0.4–0.6 per 1000)
  - Temporal decline (2.0–2.3 per 1000 from 1930–1940)
  - Geographical variation (0.1–0.4 Great Britain)
- Importance of early diagnosis lies in the propensity for late neurologic and infectious complications

Spinal Dysraphism

- Open NTD (Spinal Bifida Aperta)
  - Visible neural tissue/absent epithelialization
  - CSF leak/potential fo meningitis
  - Tethering at the junction of neuroectoderm and cutaneous ectoderm
  - Requires immediate (48 hours) repair
  - Associated with CNS malformations (hydrocephalus, chiari type II, etc.)

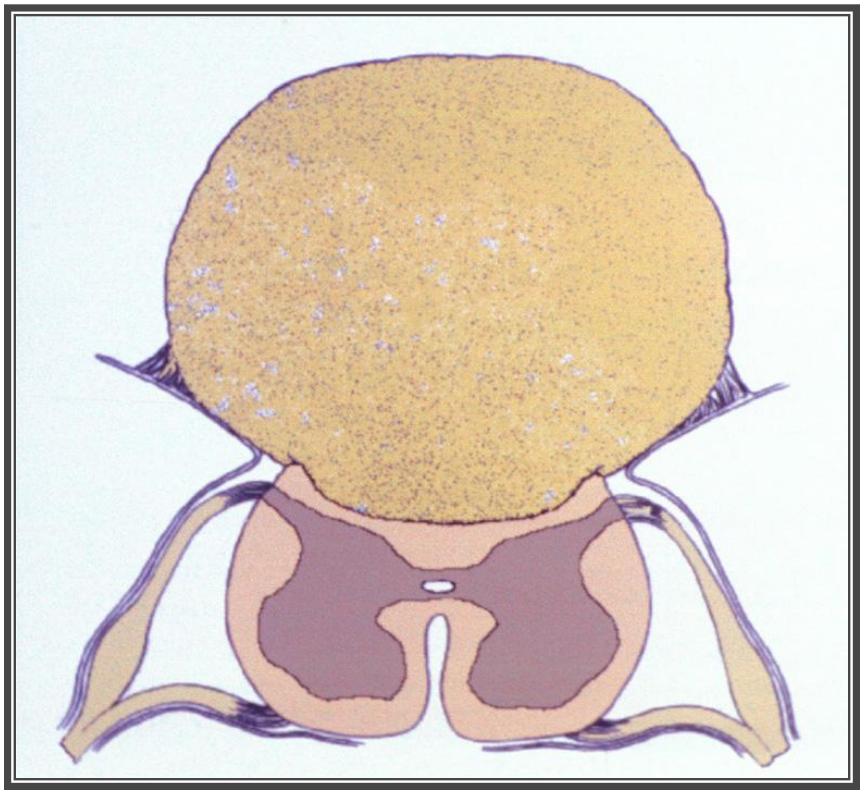






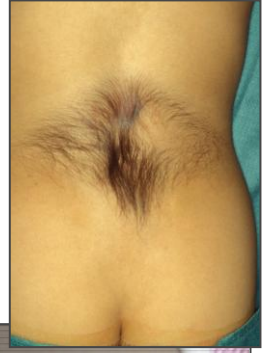
Spinal Dysraphism

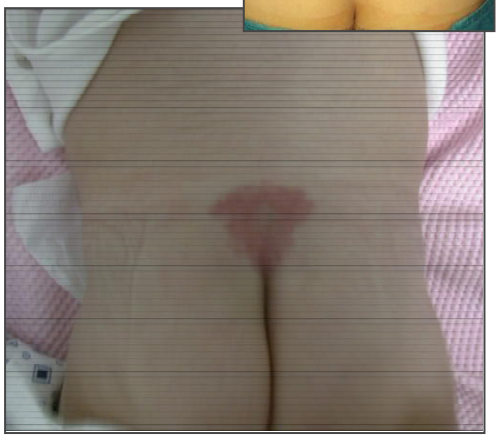
- Closed NTD (Spinal Bifida Oculta)
  - No visible neural tissue/epithelialization
  - Cutaneous stigmata are common
  - No CSF leak/no risk of meningitis
  - Tethering is variable based upon the type of NTD
  - Does not require immediate repair (elective at approximately 6 months)
  - Not associated with CNS malformations

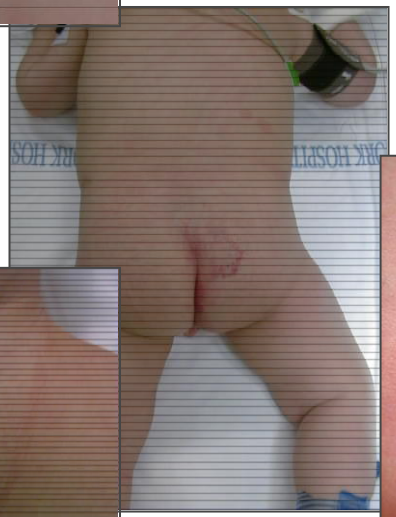


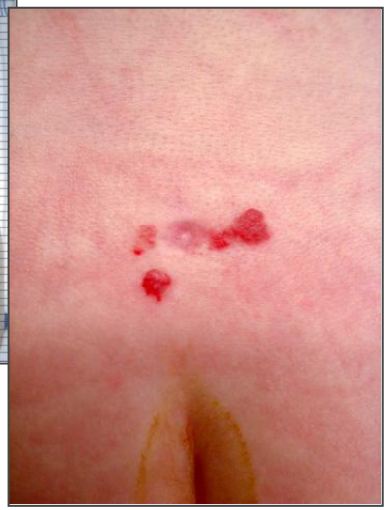
Closed Neural Tube Defect: (Spina Bifida Occulta)

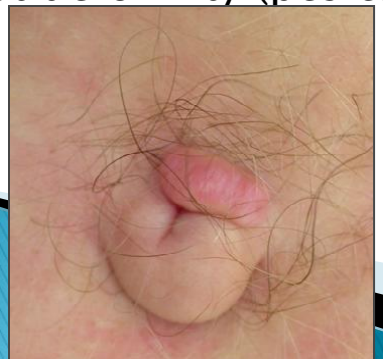
- Cutaneous findings
  - Lipomatous mass
  - Angiomata
  - Rudimentary tail
  - Atretic meningocele (manque)
  - hypertrichosis
- Anogenital malformations
  - Imperforate anus or anal atresia
  - hyposadius
- Urologic dysfunction
  - Frequent urinary tract infections
- Orthopedic
  - Limb asymmetery
  - Foot deformity (pes cavus)





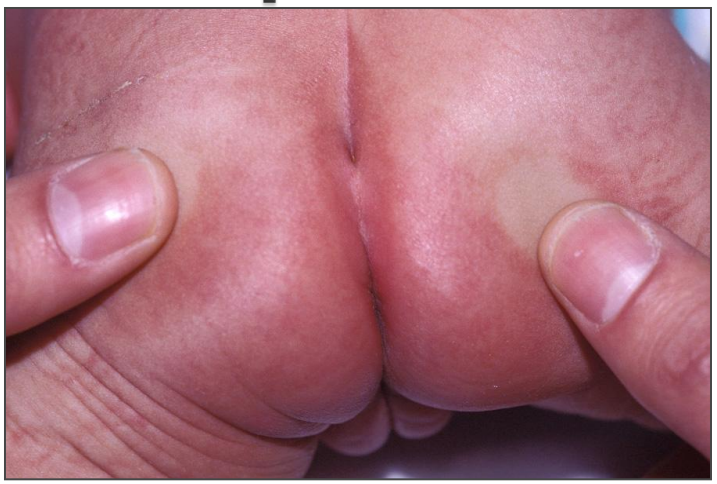


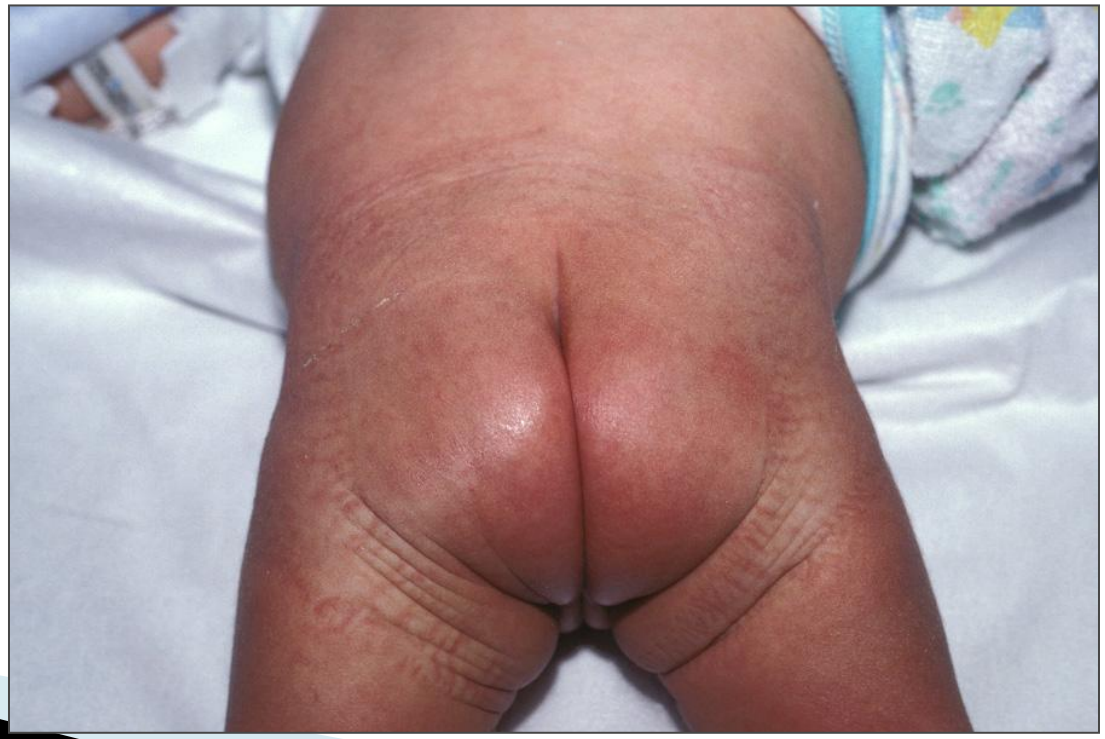




Sacral/Coccygeal Dimple

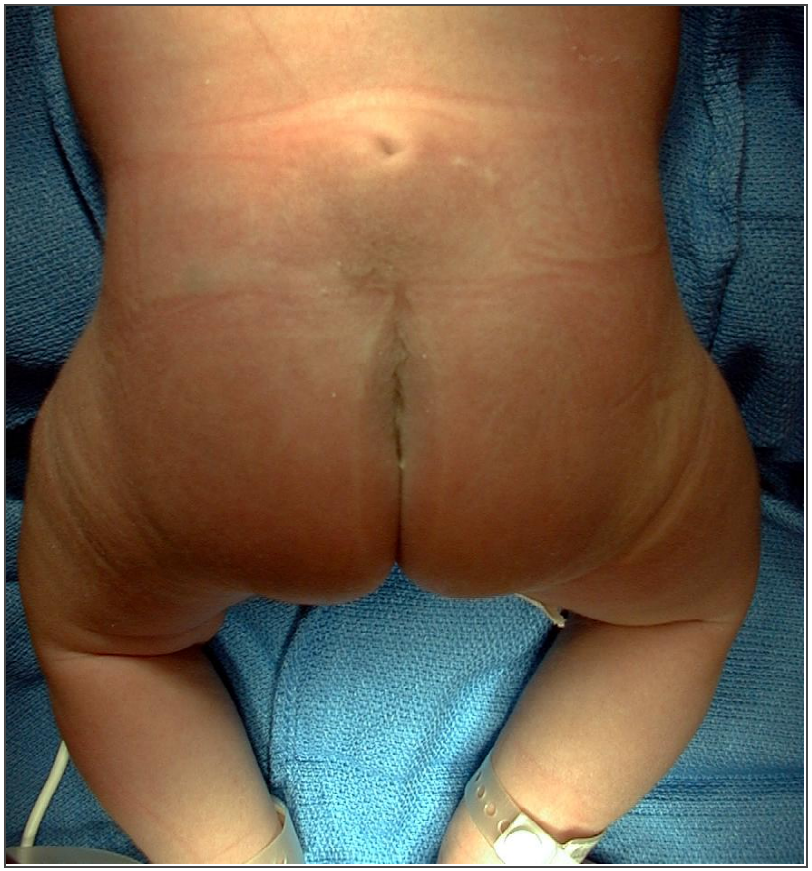
- Not a clinically relevant dysraphic state
- Located within the glueal fold
- Terminates at the tip of the coccyx





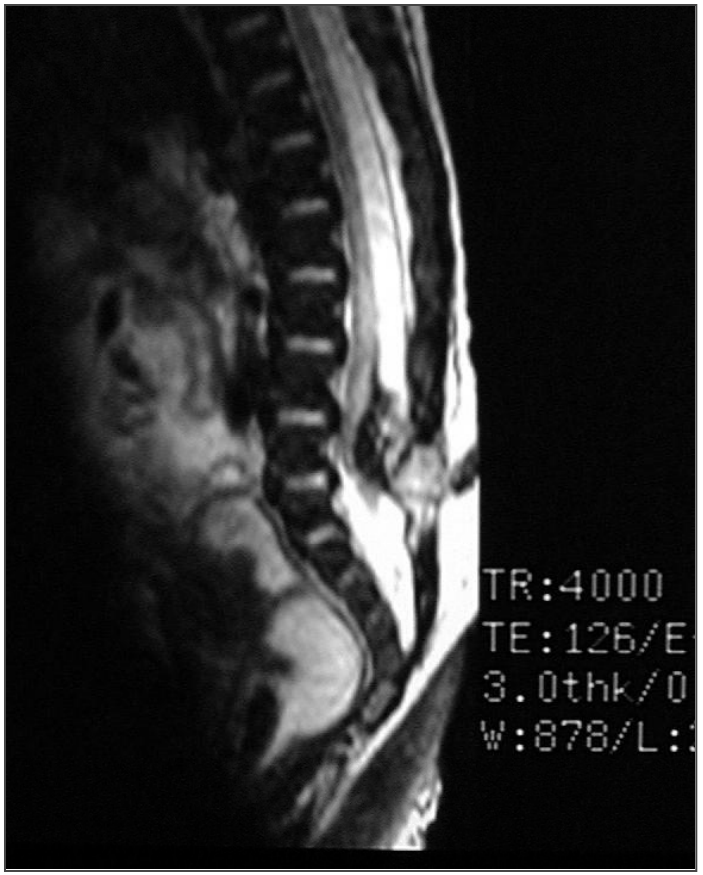
Dermal Sinus Tract

- History
  - Recurrent meningitis
- Clinical Manifestations
  - Dimple, pit, or punctum, above the gluteal crease
  - Associated hair
- Urgent referral



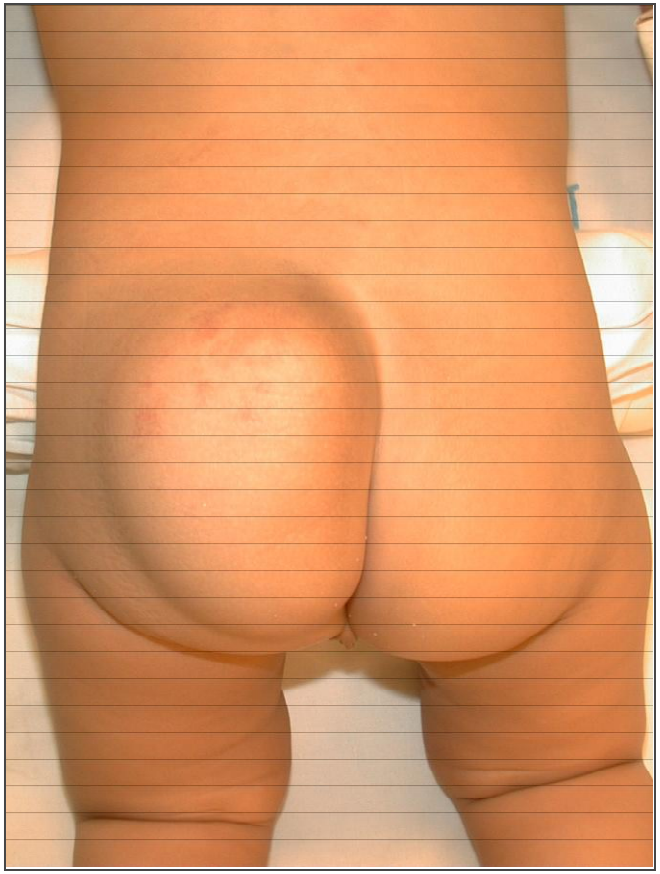
Dermal Sinus Tract

- Tethered spinal cord
- Invagination of skin and skin appendages
- Terminates intradurally
- Tethering at the terminal dermal sinus and fibrolipomatous filum terminale

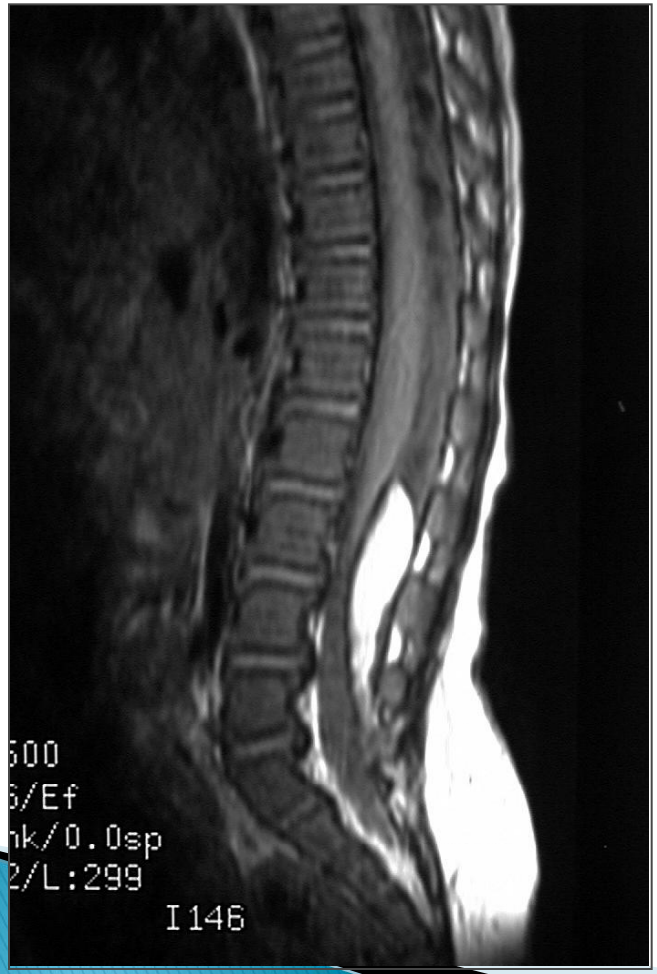


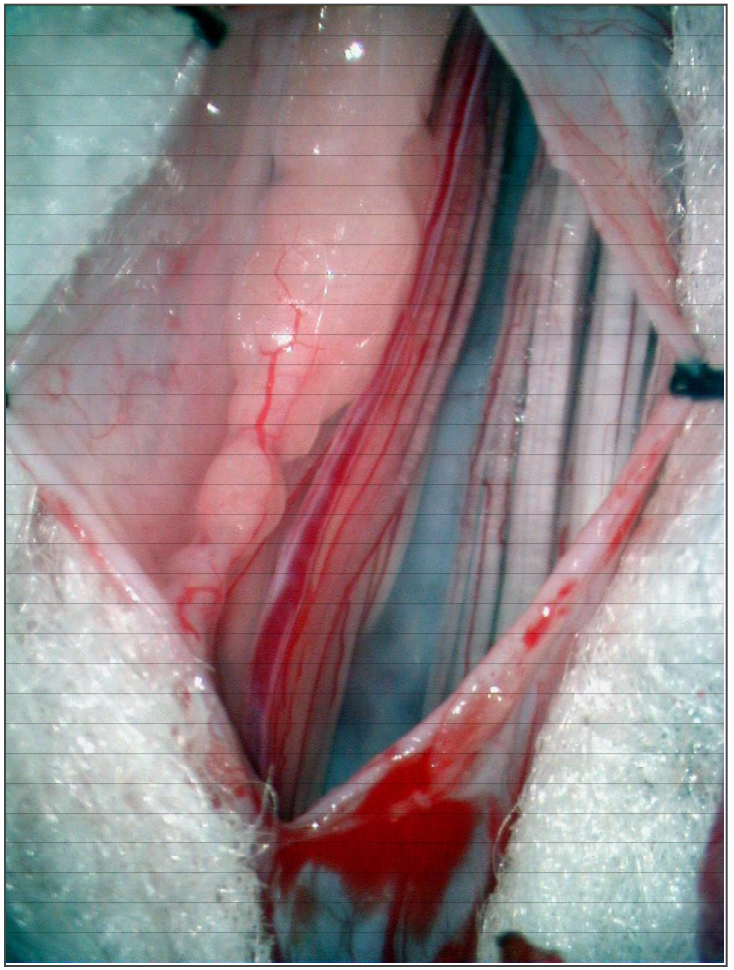
Spinal Lipomas

- Commonly asymptomatic at birth
- Cutaneous signature or subcutaneous mass (lipomyelomeningocele)
- Tethering occurs at the site of attachment between the lipoma and the spinal cord
- Lipoma is not neoplastic

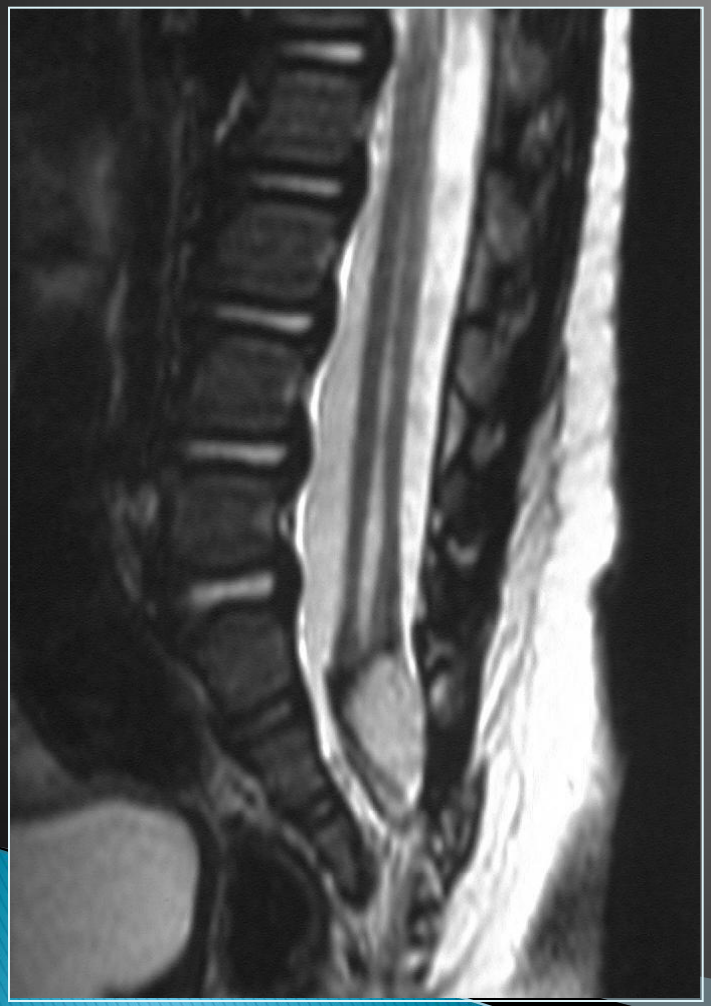


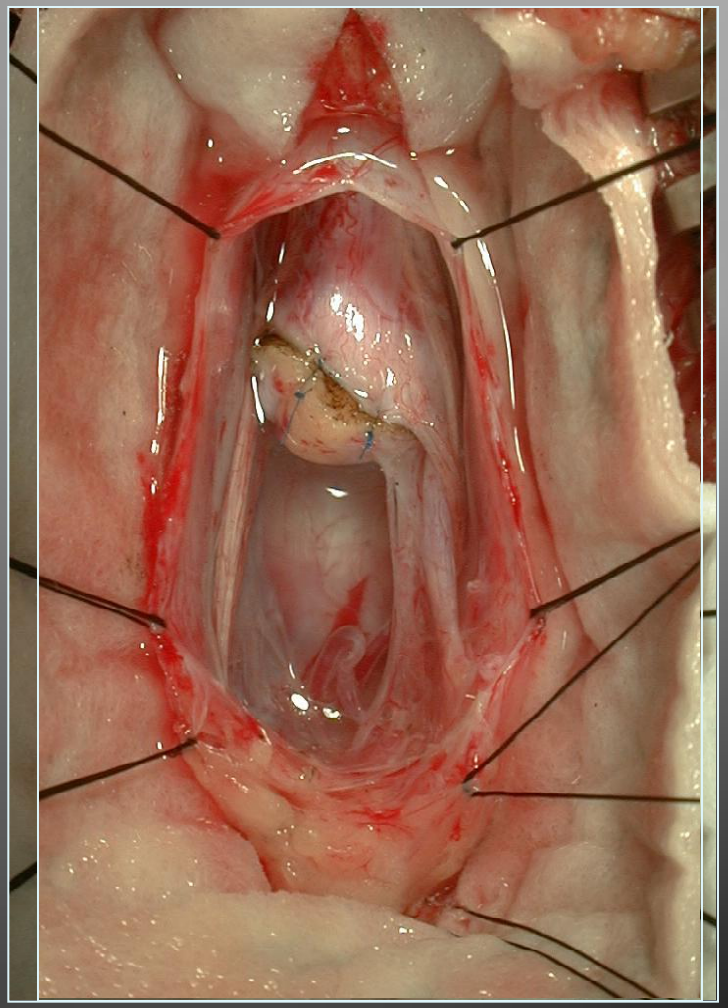
Spinal Lipomas





Spinal Lipomas



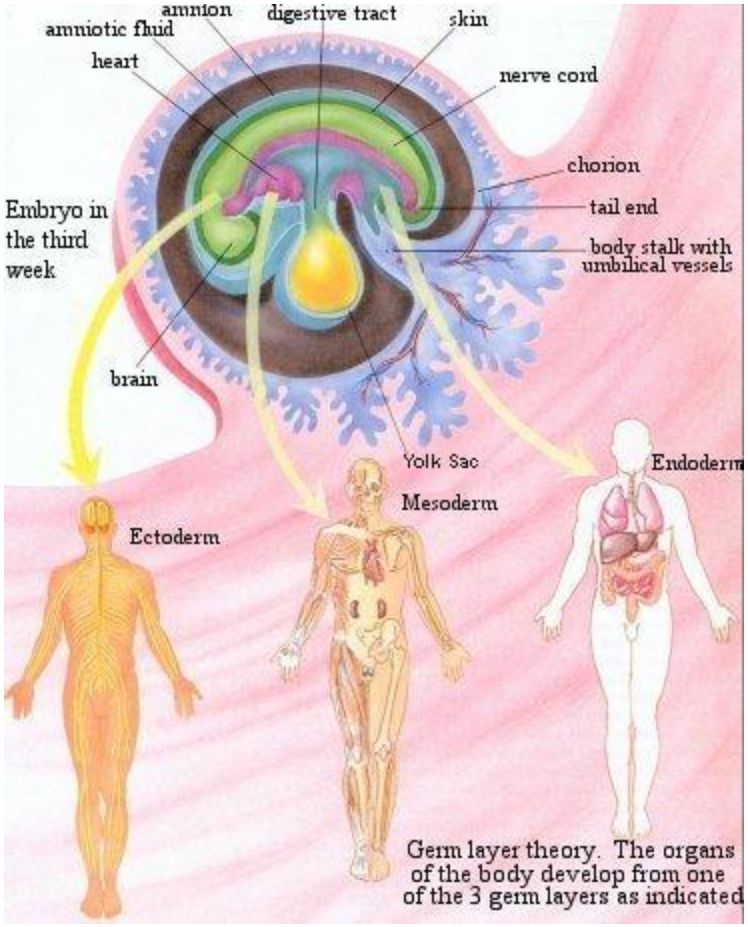


Cutaneous Stigmata: What to do?

- Image and refer to neurosurgeon
  - Dimple over lumbar spine (above gluteal crease)
  - Midline or paraspinal subcutaneous lipoma
  - Midline or paraspinal subcutaneous cyst
  - Caudal appendage (“tail”)
  - Haiy path (hypertrichosis)
  - Circumscribed atophic skin (“cigarette burn”)
- Reassure family
  - Dimple over the coccyx (within gluteal crease)
  - Isolated café-au-lait spot
  - Melanotic nevus

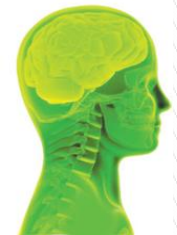
What are neurocutaneous disorders?

- A disorder that affects the skin and nervous system.
- Most (but not all) neurocutaneous disorders are genetic.

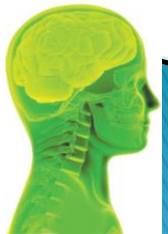


What are neurocutaneous disorders?

- Tuberous Sclerosis
- Neurofibromatosis Type 1
- Neurofibromatosis Type 2
- Sturge-Weber Syndrome
- Ataxia-Telangiectasia
- Von Hippel-Lindau disease
- Hypomelanosis of Ito
- Linear Sebaceous nevus syndrome
- Neurocutaneous melanosis
- Parry-Romberg syndrome
- Leopard syndrome
- Incontinentia pigmenti
- Gorlin-Goltz Syndrome
- Osler-Weber-Rendu disease

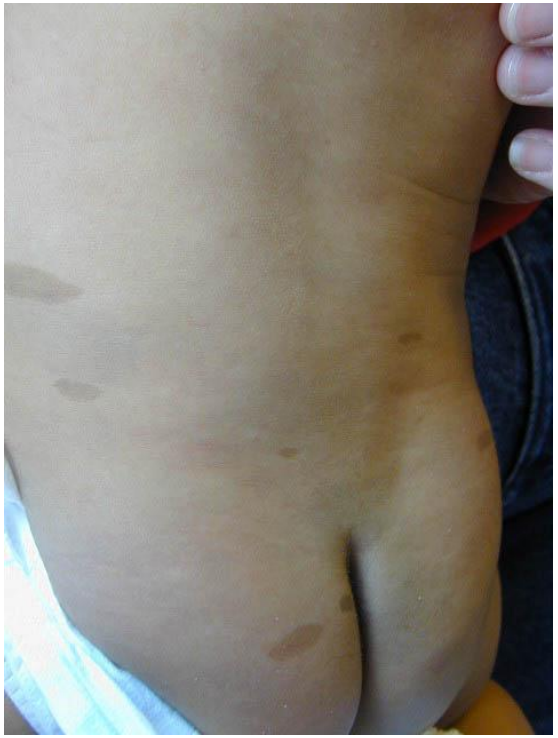


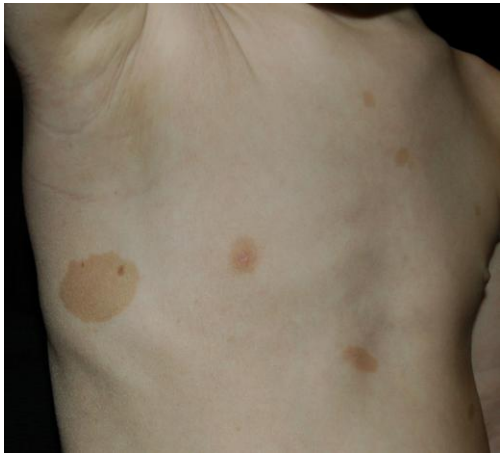
Syndrome	Incidence	Genetics
Neurofibromatosis Type 1	1 in 3000	AD
Tuberous Sclerosis	1 in 10,000	AD
Neurofibromatosis Type 2	1 in 40,000	AD
Sturge-Weber	1 in 50,000	Sporadic
Ataxia-Telangiectasia	1 in 50,000	AR
Neurocutaneous Melanosis	Rare	Sporadic

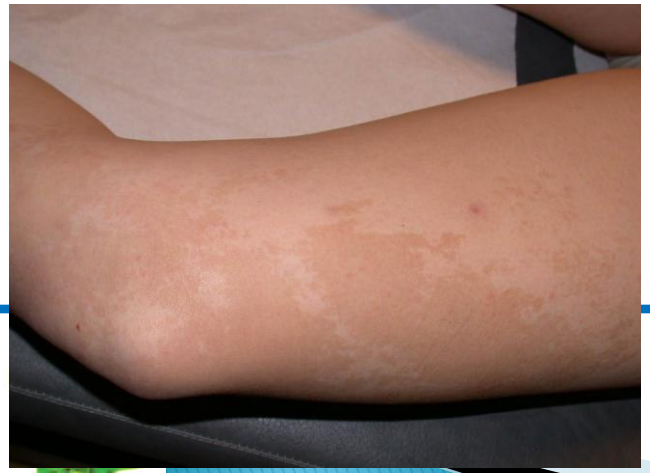


Café-au-lait spots

Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

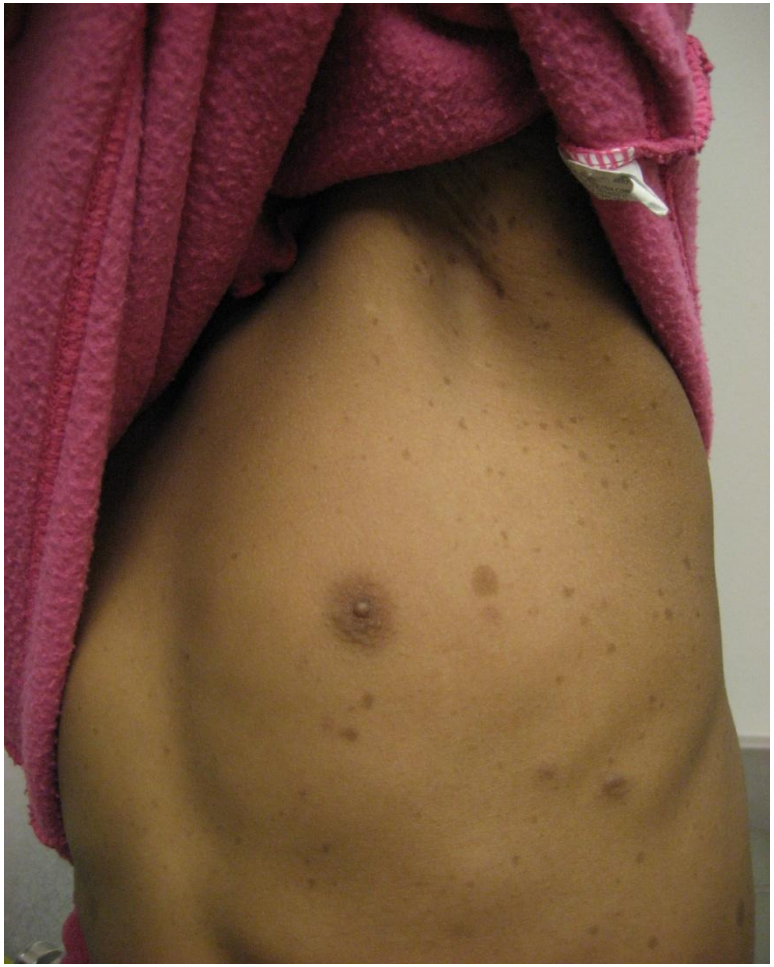






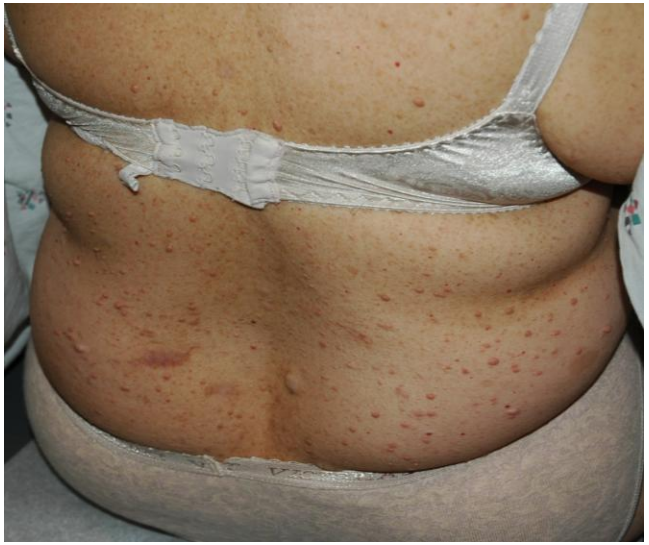
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

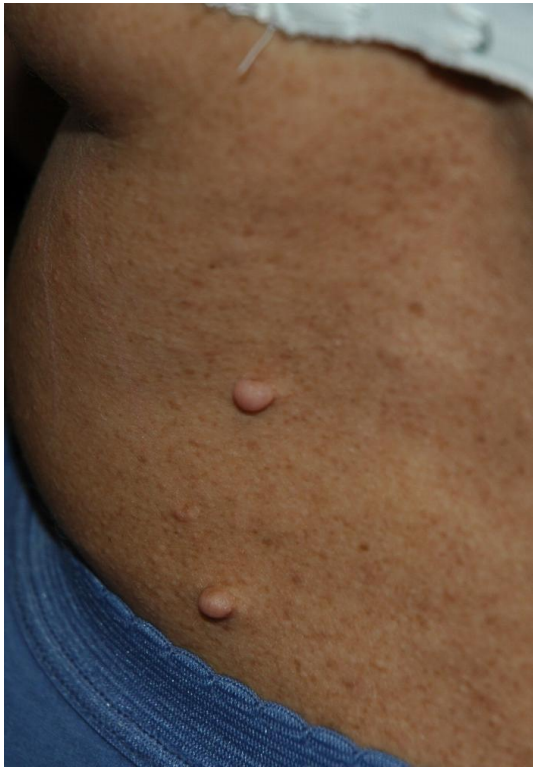
Freckling



Cutaneous Neurofibromas

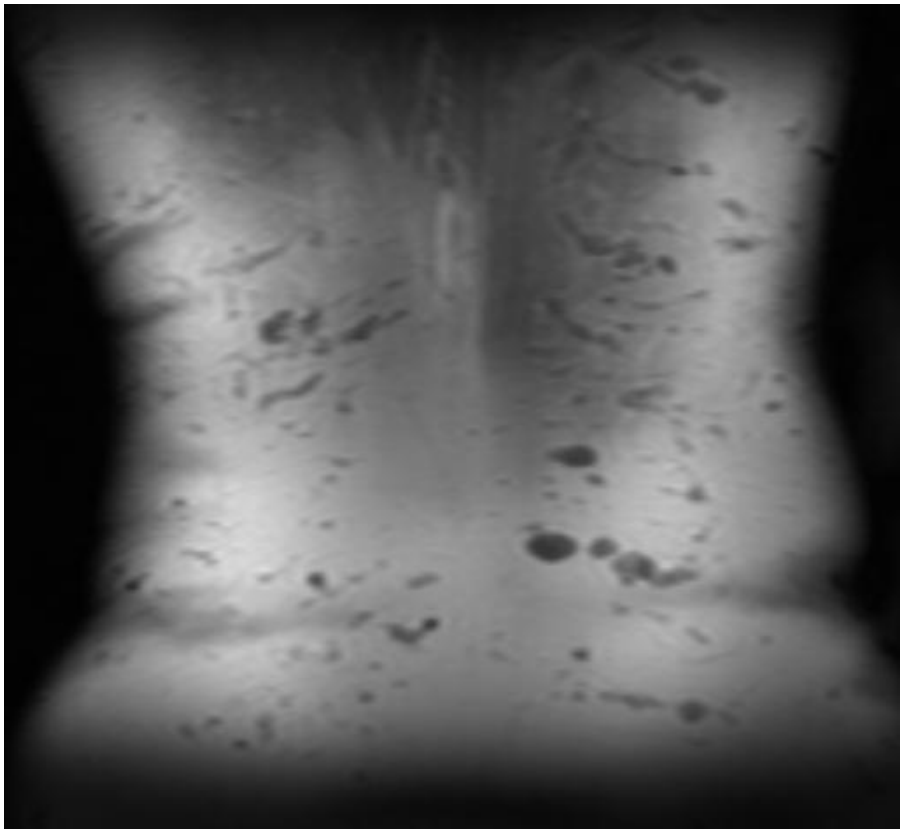
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

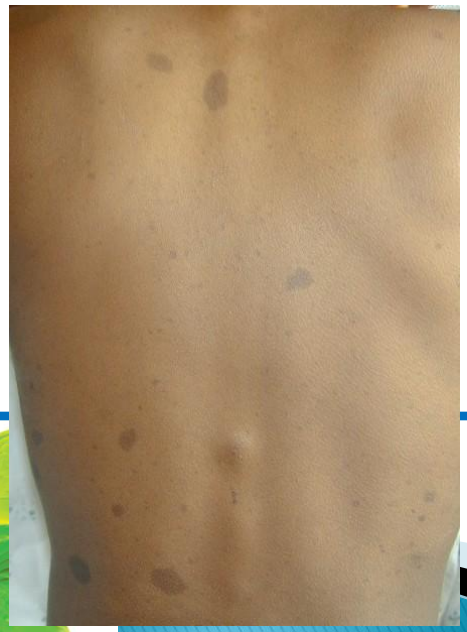




Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

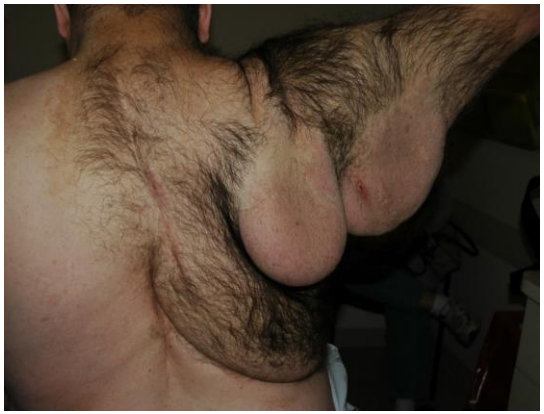
Subutaneous Neurofibromas

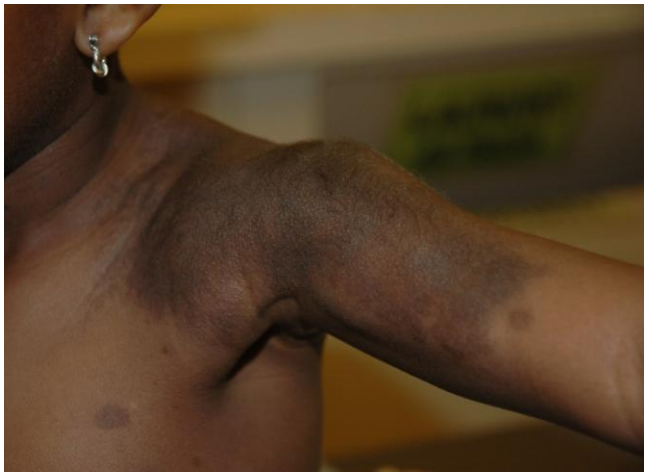


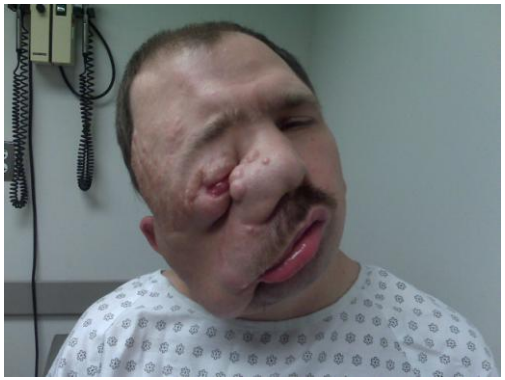


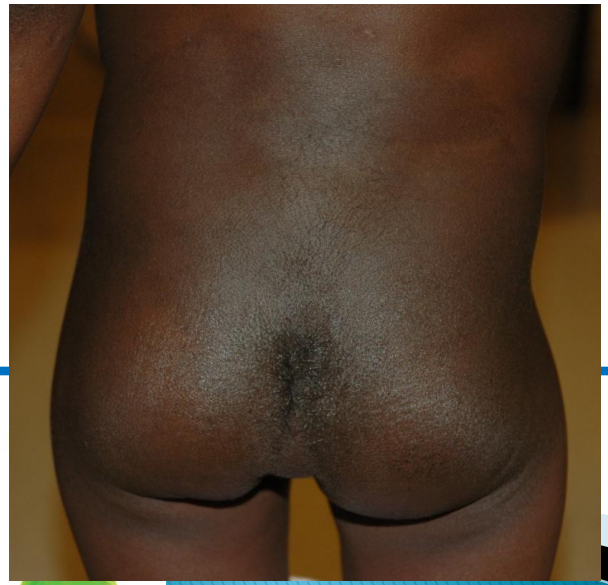
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

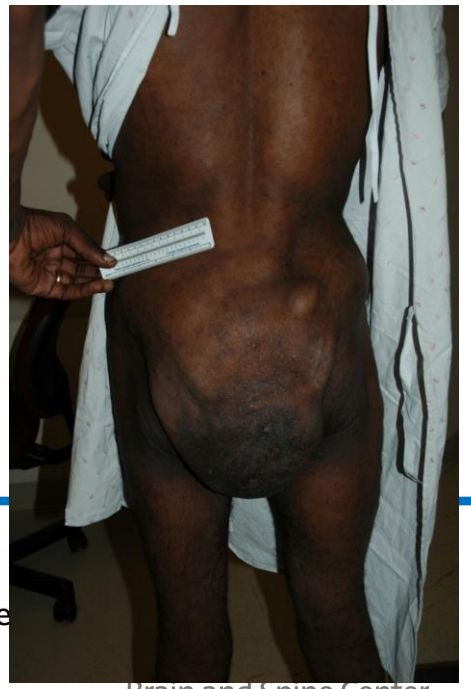
Plexiform Neurofibromas





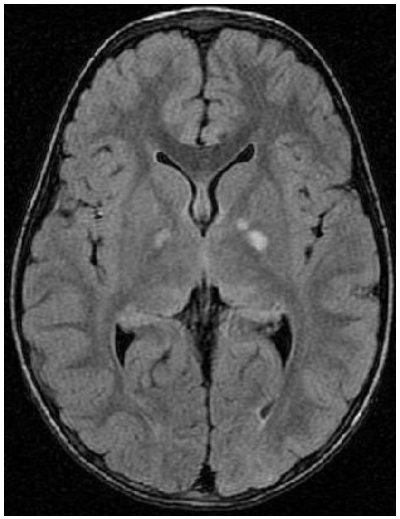


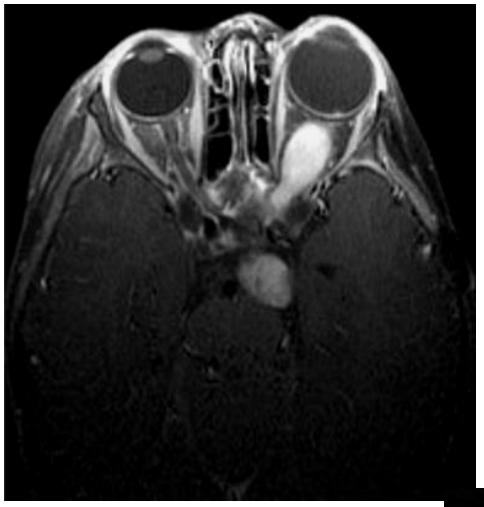


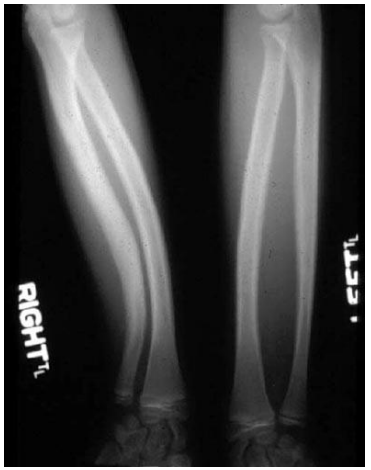


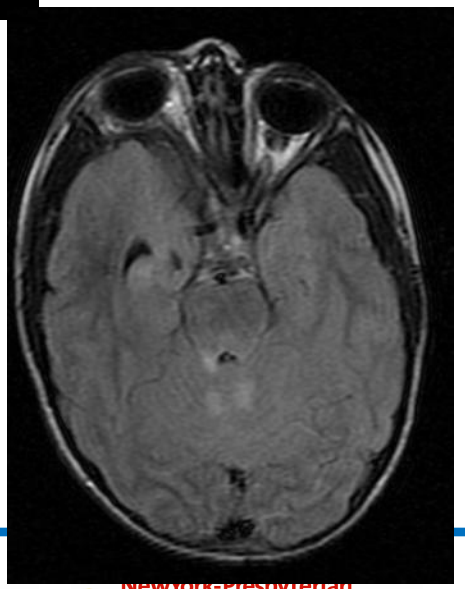
Other Manifestations

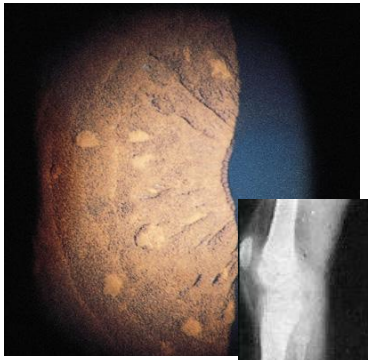
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

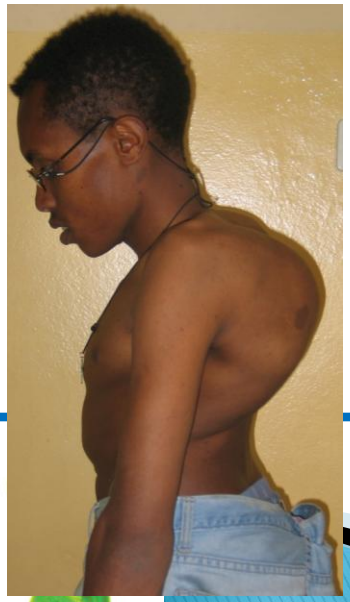


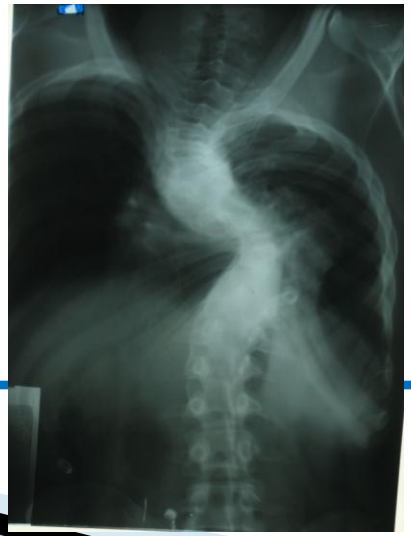


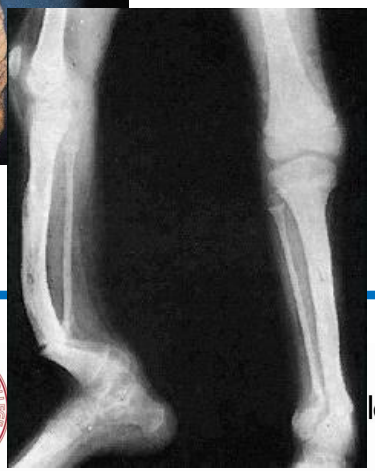






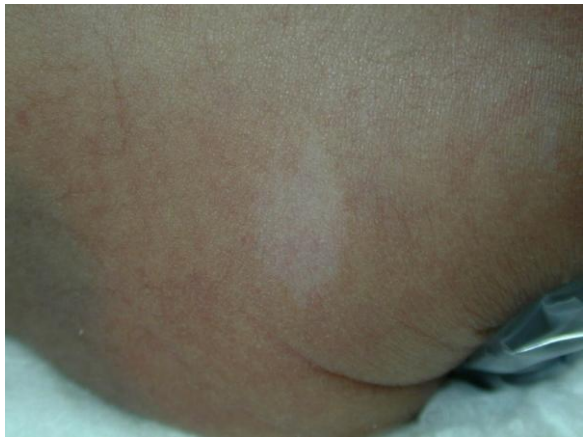


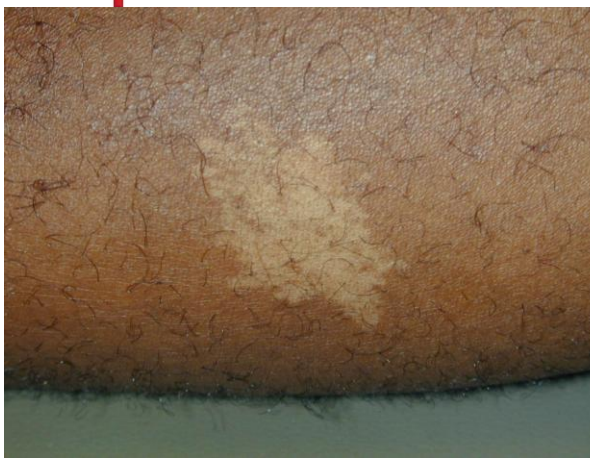


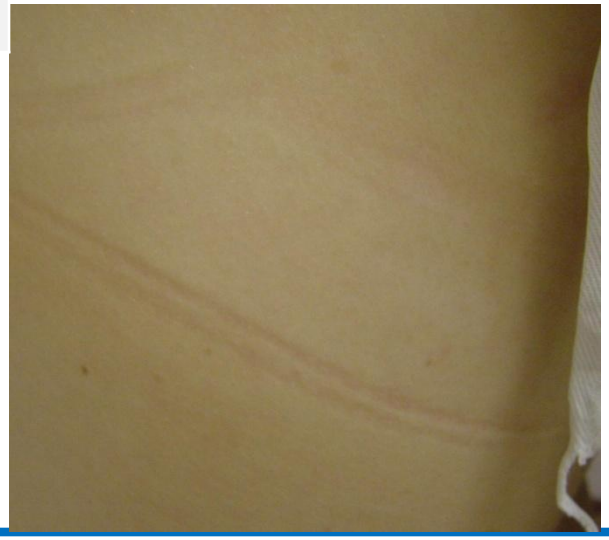


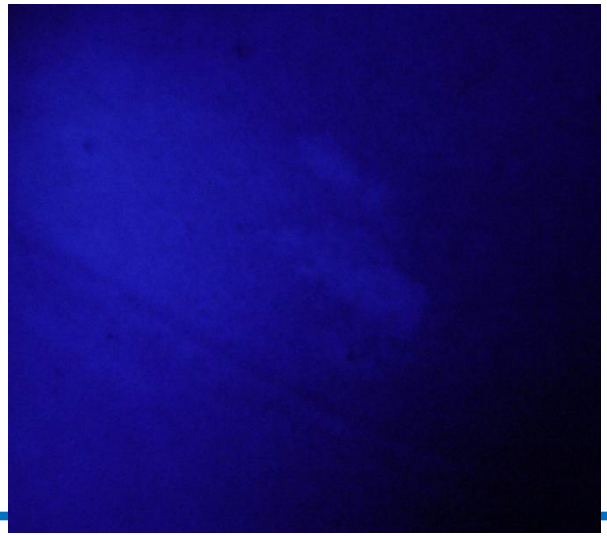
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

Ash Leaf Spots



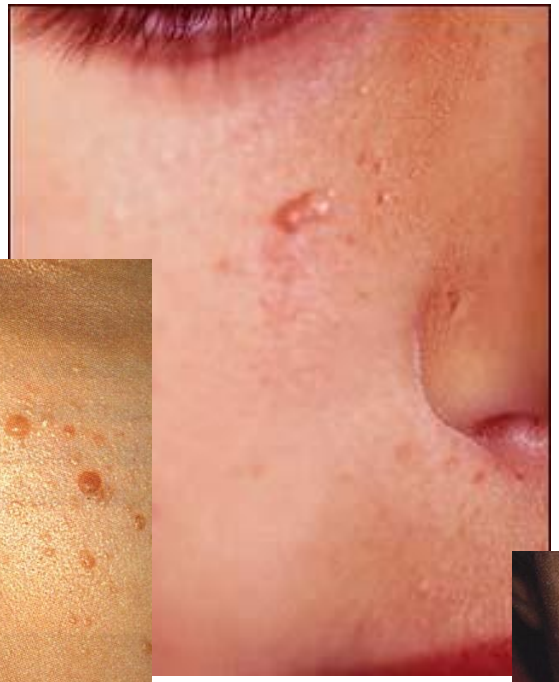


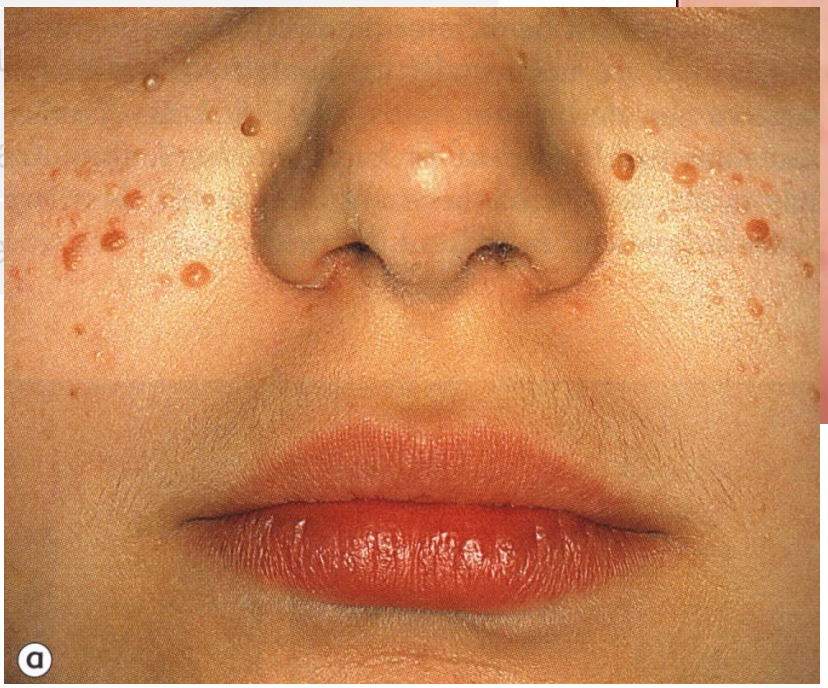


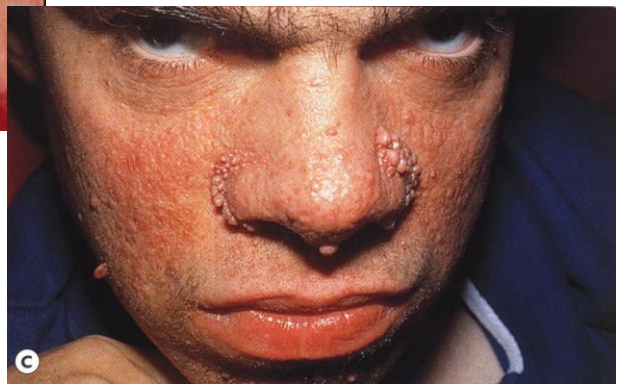


Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Stu
Ata
Ne

Angiofibroma



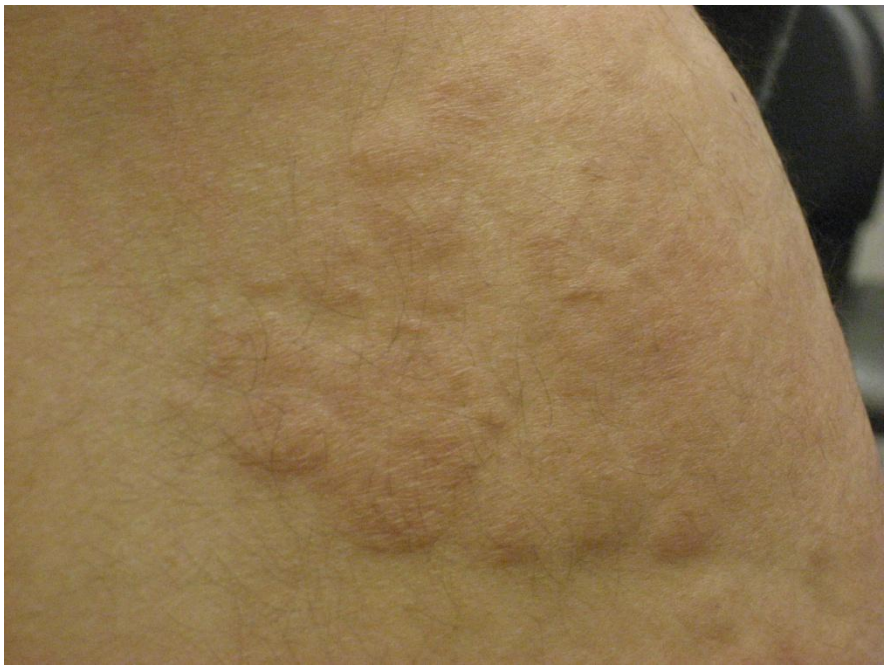




From: Pediatric Dermatology by Cohen, BA

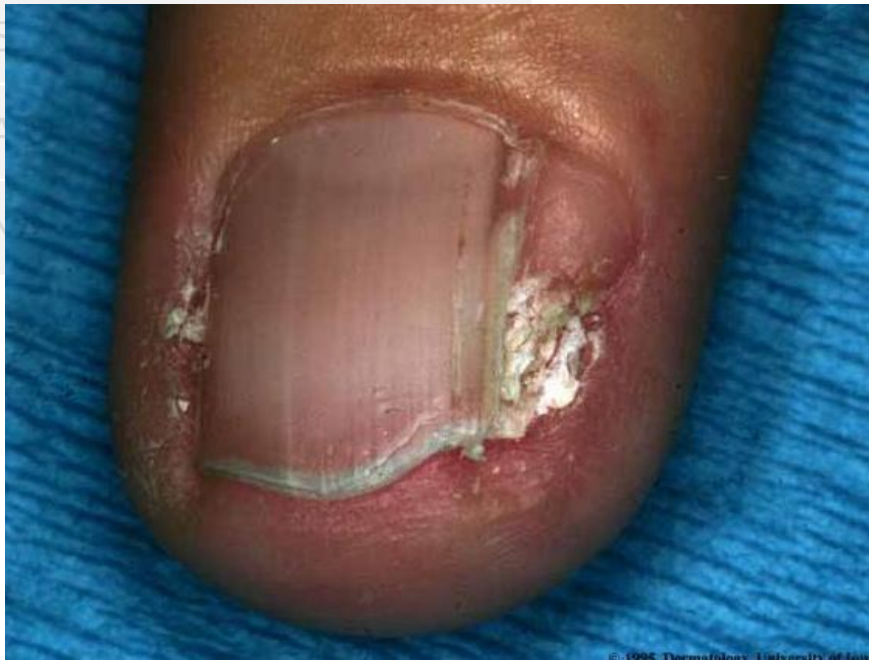
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

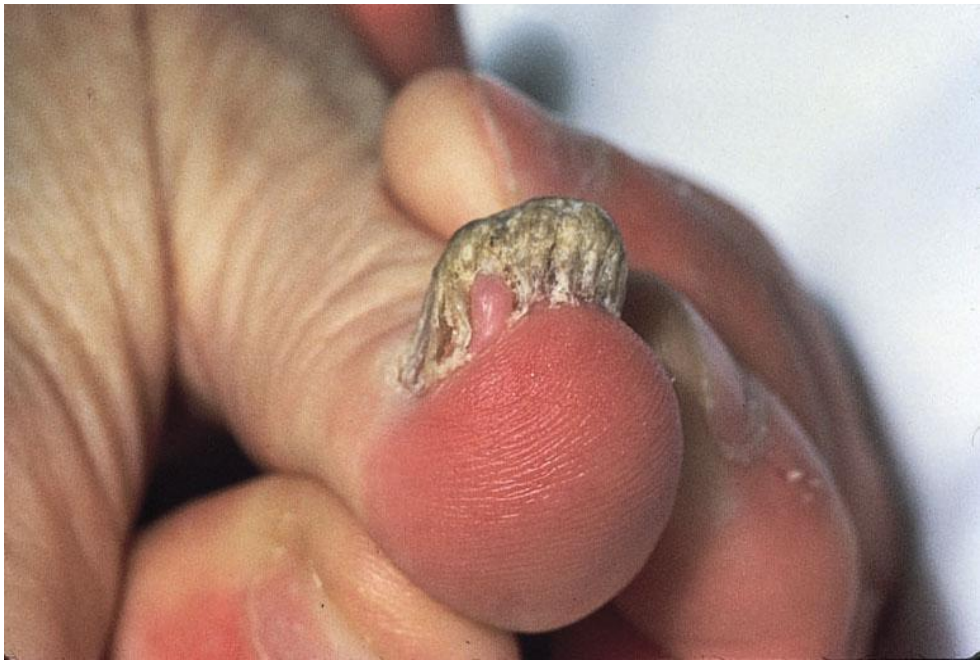
Shagreen Patch



Periungual and Ungual Fibromas

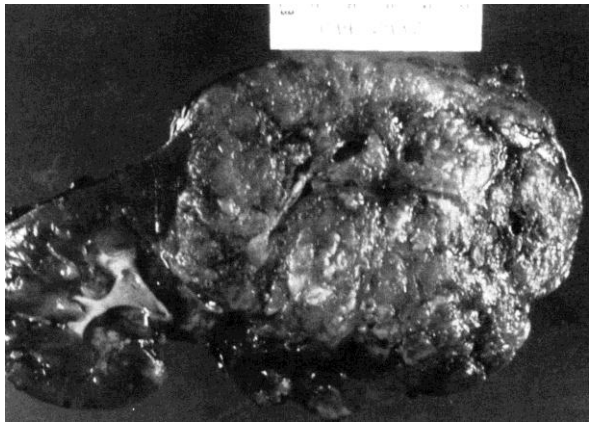
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2

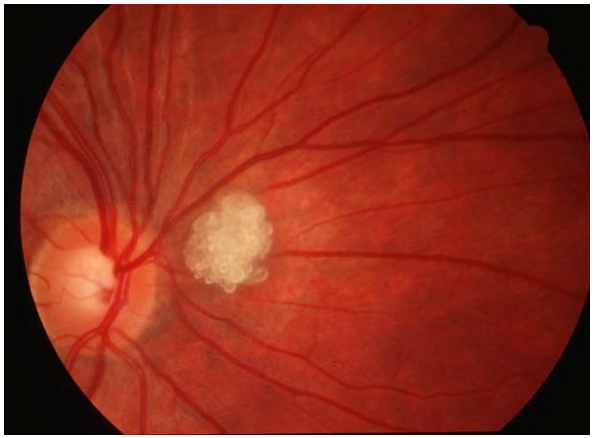


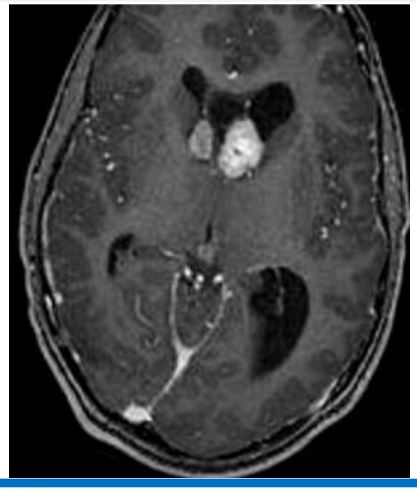


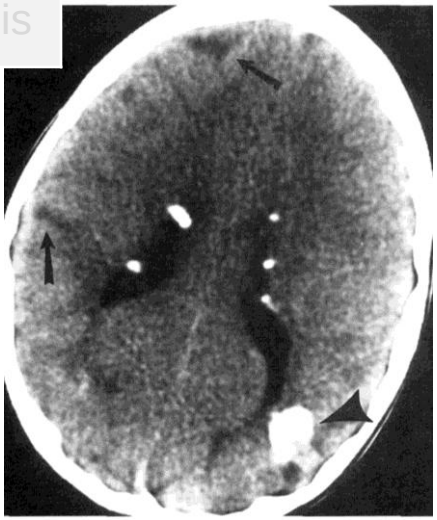
Other Manifestations

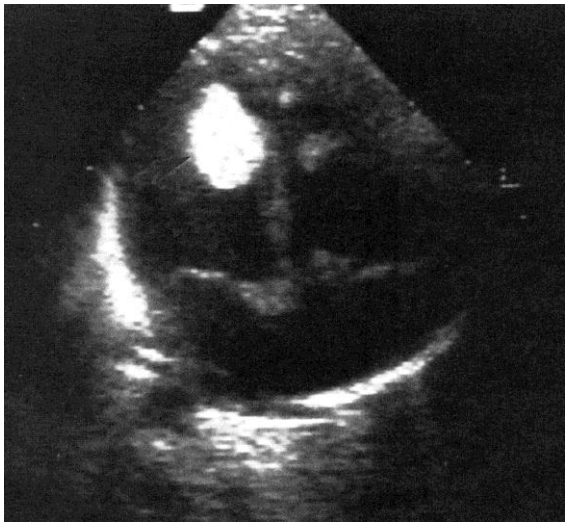
Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis

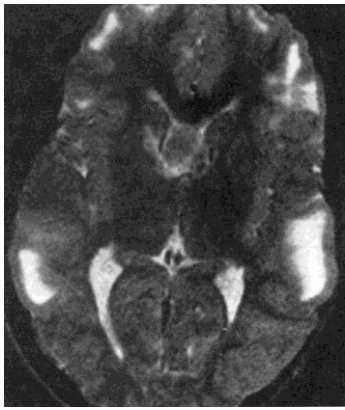






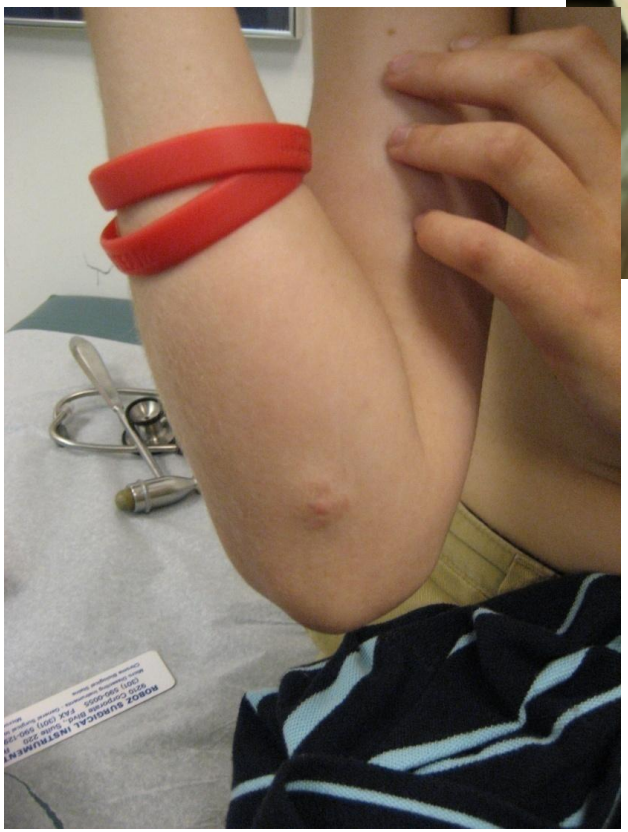


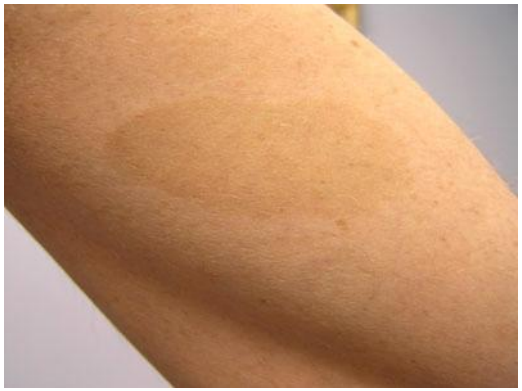


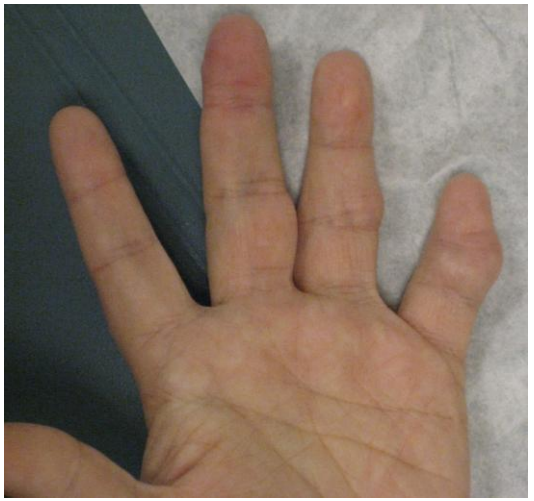


Cutaneous Manifestations

Syndrome
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis







Other Manifestations

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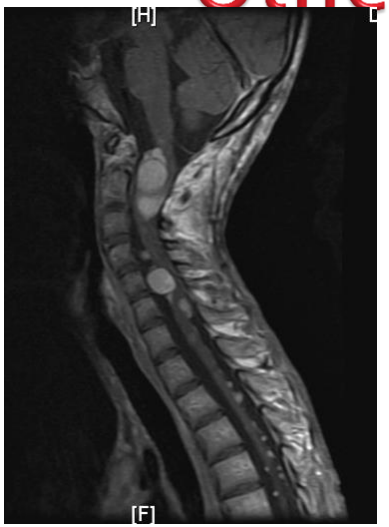
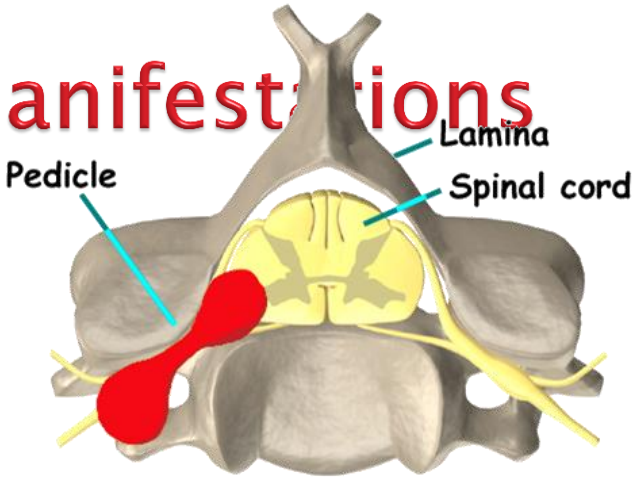
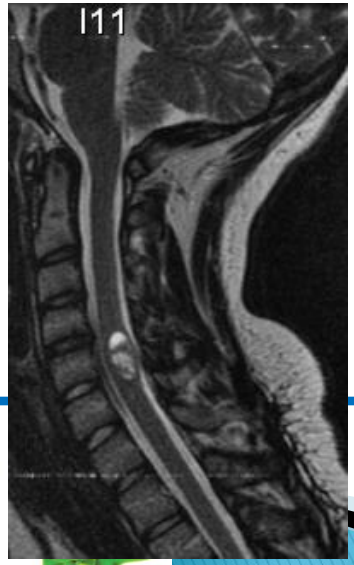
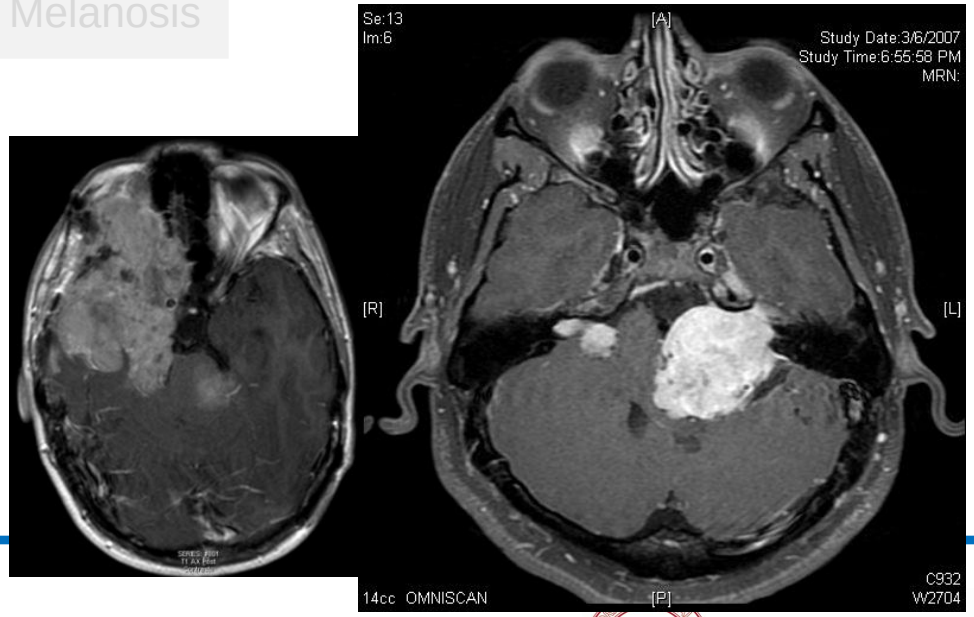
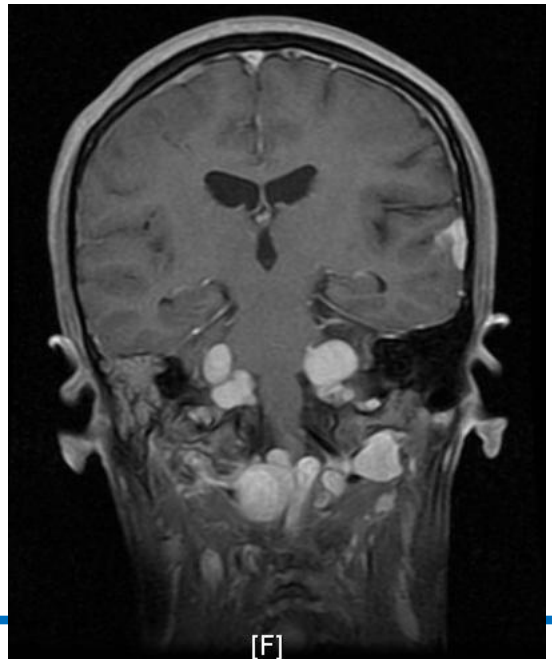
Neurofibromatosis Type 1
Tuberous Sclerosis
Neurofibromatosis Type 2
Sturge-Weber
Ataxia-Telangiectasia
Neurocutaneous Melanosis











Weill Cornell Medical College

NewYork-Presbyterian
Phyllis and David Komansky Center for Children's Health
Pediatric
Brain and Spine Center

Syndrome

- Neurofibromatosis Type 1
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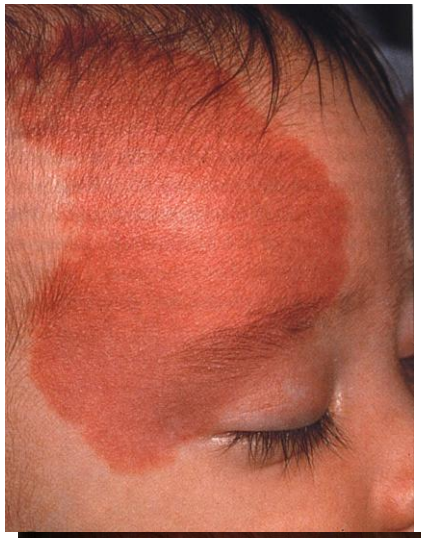
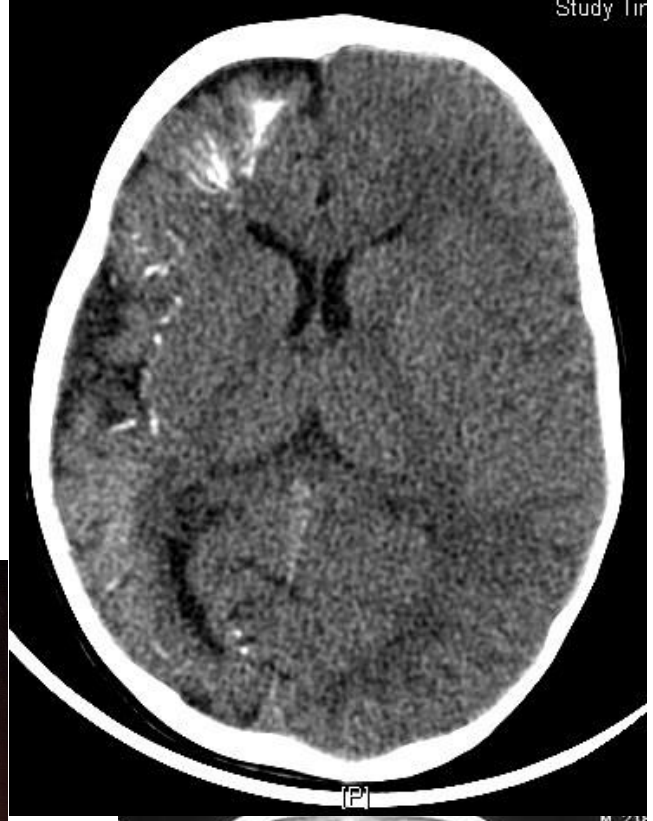
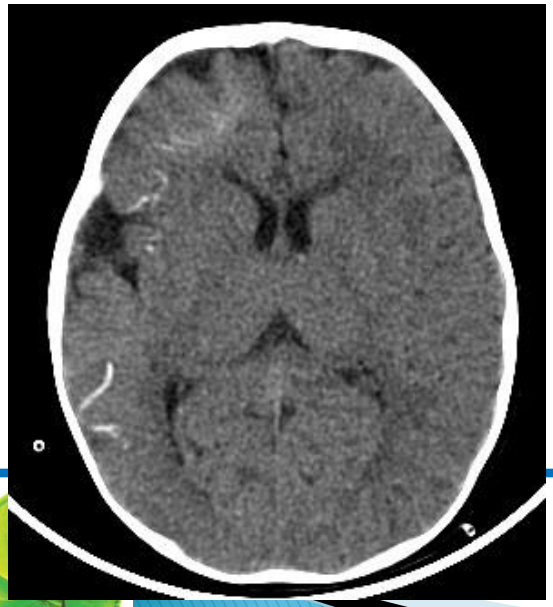
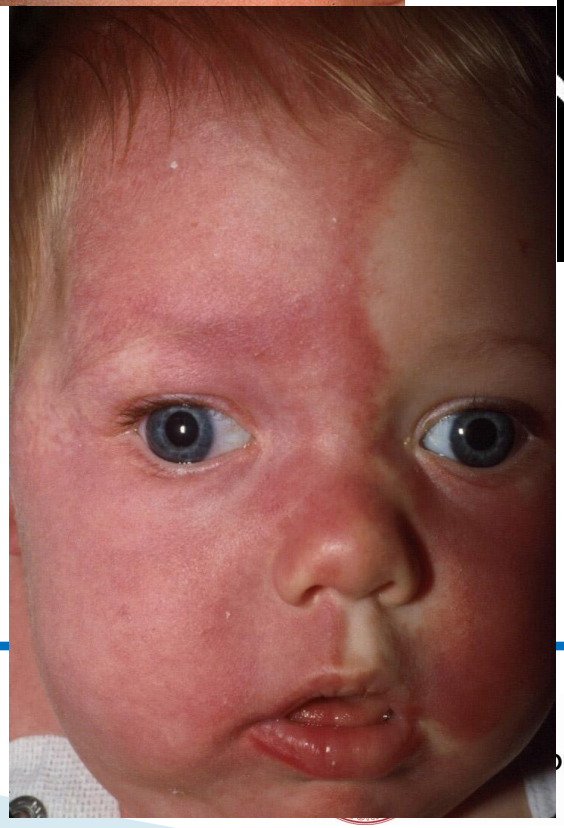
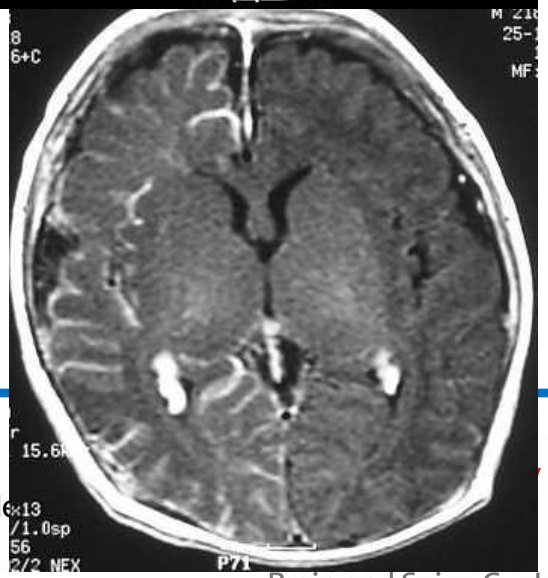
- Ataxia-Telangiectasia
- Neurocutaneous Melanosis











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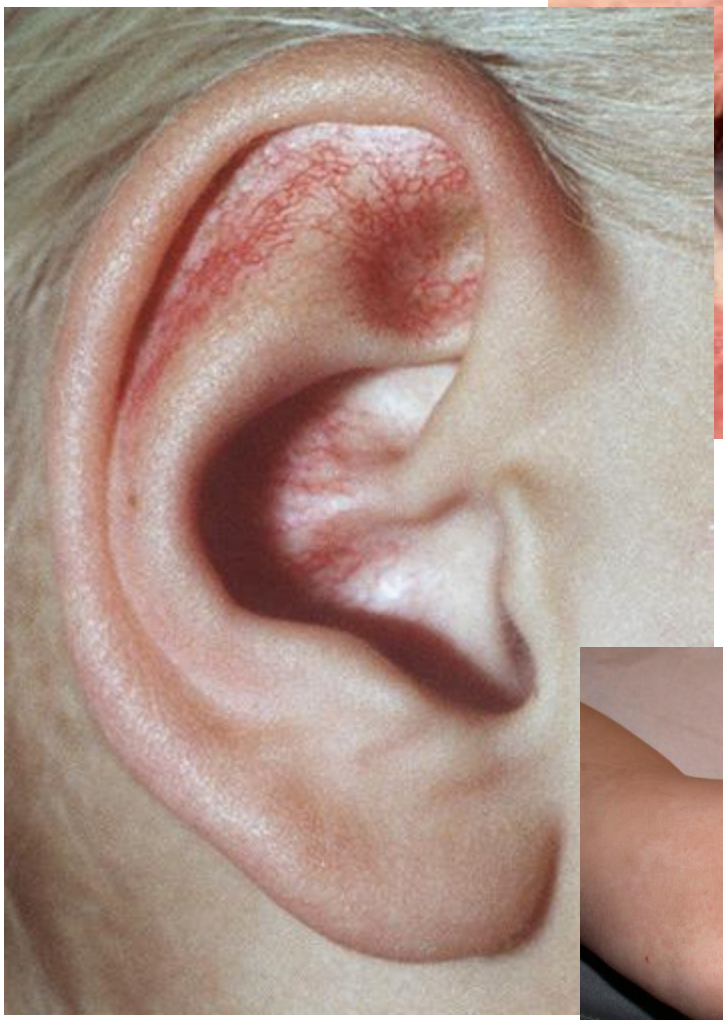

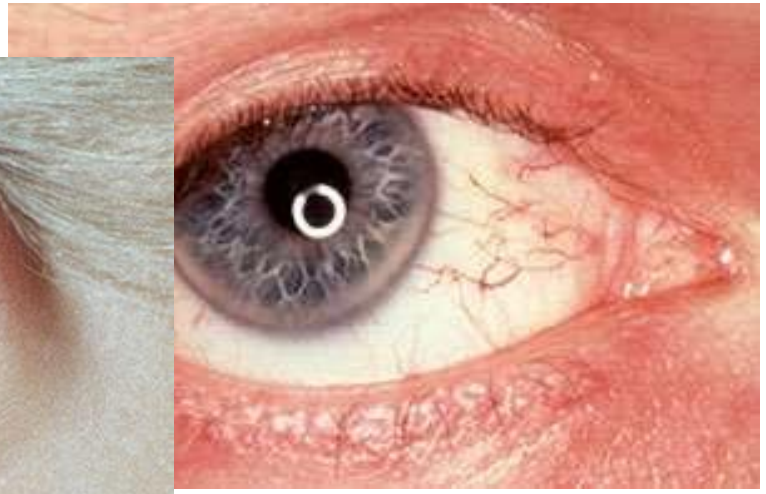

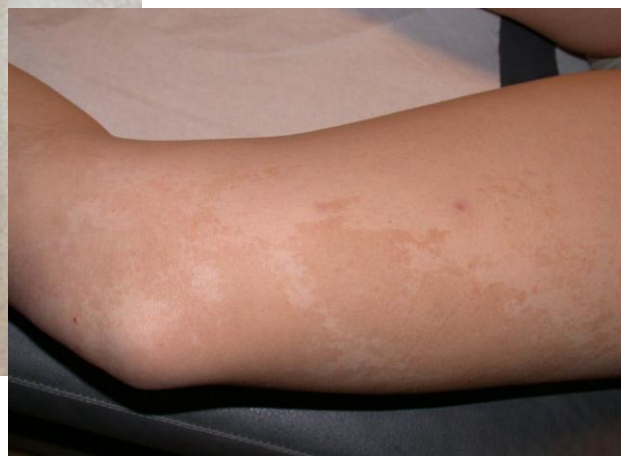

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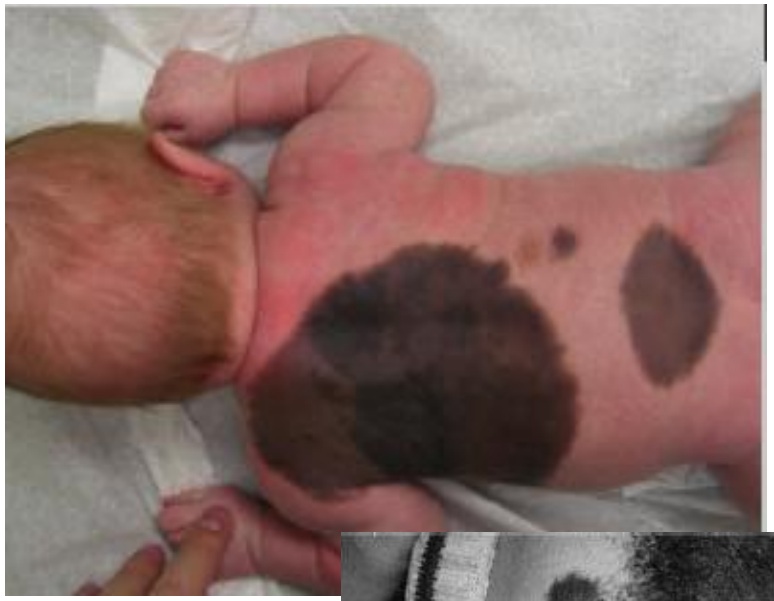

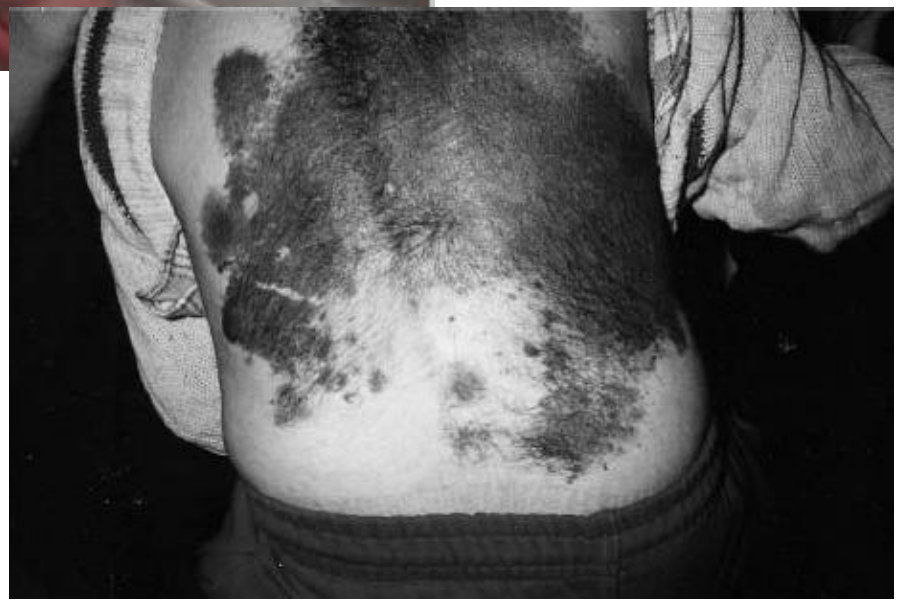

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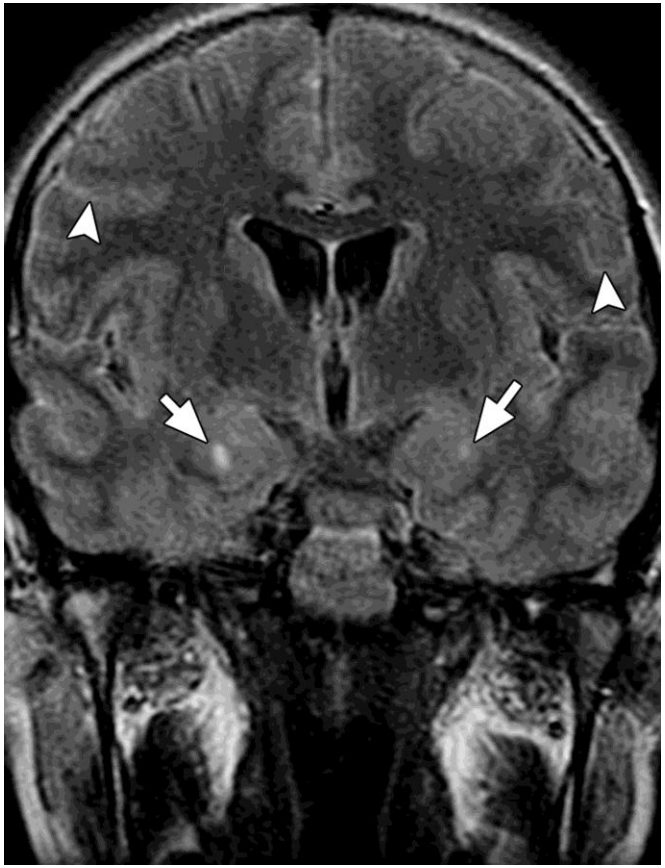

Smith A B et al. Radiographics 2009;29:1503-1524