

Neurocutaneous Syndrome

Neurofibromatosis

Neurofibromatosis 1

(von Recklinghausen's syndrome)

Mutation on chromosome 17

Neurofibromatosis 2

Mutation on chromosome 22

both types are **autosomal dominant** inheritance

- 1- Café-au-lait spots (at least 5 mm)
- 2- Axillary/groin freckles
- 3- Peripheral neurofibromas
- 4- Iris: Lisch nodules in > 90%
- 5- Macrocephaly, short stature, learning disabilities, feeding problems



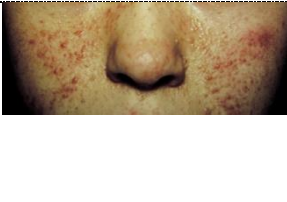


- 1- Ear: Bilateral acoustic neuromas
- 2- Eyes: Posterior subcapsular Cataracts

Tuberous sclerosis

Autosomal Dominant inheritance → the majority of features seen in TS are **neuro-cutaneous**

Cigarettes and coffee with a rough stupid person with a butterfly on his nose while he is dancing
Cigarettes (ash) - coffee (café-au-lait) - stupid (intellectual and developmental) - dancing (epilepsy)

Cutaneous features :

Depigmented 'ash-leaf' spots	Shagreen patches	Adenoma sebaceum	Subungual fibromata	Café-au-lait spots
which fluoresce under UV light	Roughened patches of skin over lumbar spine	butterfly distribution over nose	Fibromata beneath nails	may be seen
				

Neurological features :

- Developmental delay
- Epilepsy (infantile spasms or partial)
- Intellectual impairment

Also

- Brain: Gliomatous changes can occur in the brain lesions
- Retinal hamartomas: dense white areas on retina (phakomata)
- Heart: Rhabdomyomas
- Renal angiomyolipomata

Sturge-Weber syndrome

Cutaneous features :

- 1- **Port wine stains** (nevus flammeus) along the territory of trigeminal nerve (in V1 cranial nerve distribution)

Neurological features :

- 1- **Angiomatous** malformations of the brain
- 2- Seizures (focal or generalized)
- 3- Hemiparesis & Hemisensory disturbance

Eye: 1- Hemianopia 2- Ipsilateral Glaucoma




Treatment: 1- Evaluate for glaucoma: reduce IOP 2- Treat Seizures : Anti-convulsives
3- Argon laser therapy is successful in removing the skin lesions

Skull x-rays:

taken after the age of 2 years, reveal:
gyriform intracranial calcifications that resemble a **tramline**



Differential Diagnosis: Other Cutaneous Lesions

Diagnosis	Figure	Description	Association	Management
Mongolian spots		Blue/gray macules on: 1- presacral back 2- posterior thighs	Usually fade in first few years	Rule out child abuse
Erythema toxicum		Non-toxic neonate in 2nd day of life with : Erythematous papules & vesicles surrounded by patches of erythema and eosinophils in skin lesions		Benign, self-limited condition
Strawberry Hemangioma		Red, sharply demarcated, raised lesions. -Appearing in first 2 months - rapidly expanding, - then involuting by age 5-9 years	Consider underlying organ involvement with deep hemangiomas. - If it involves the larynx, it can cause obstruction. - May cause high output cardiac failure when large.	Treat with: 1- steroids or 2- pulsed laser if large or interferes with organ function.