Tuberous sclerosis complex clinical features

This rare multi-system genetic disease causes benign tumors to grow in the brain and on other vital organs such as the kidneys, heart, eyes, lungs, and skin. A combination of symptoms may include seizures, intellectual disability, developmental delay, behavioral problems, skin abnormalities, lung and kidney disease.

Classic clinical triad: seizures, mental retardation, and sebaceous adenomas; the full clinical triad is seen in < 1/3 of cases.

At least 50% of patients with tuberous sclerosis complex present with intractable epilepsy; for these patients, resective surgery is a treatment option.

The diagnosis of TSC is based on clinical features, but the variability of phenotype and age at symptom onset makes this challenging.

In the infant, the earliest finding is of “ash leaf” macules (hypomelanotic, leaf-shaped) that are best seen with a Wood’s lamp. Infantile myoclonus may also occur.

In older children or adults, the myoclonus is often replaced by generalized tonic-clonic or partial complex seizures, which occur in 70–80%. Facial adenomas are not present at birth but appear in > 90% by age 4 yrs (these are not really adenomas of the sebaceous glands, but are small hamartomas of cutaneous nerve elements that are yellowish-brown and glistening and tend to arise in a butterfly malar distribution, usually sparing the upper lip).

Retinal hamartomas occur in ≈ 50% (central calcified hamartoma near the optic disc or a more subtle peripheral flat salmon-colored lesion). A distinctive depigmented iris lesion may also occur.

Epileptic seizure in tuberous sclerosis complex

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