GENETICS AND GENODERMATOSES

A RARE BUT CLASSICAL AND DIFFERENT PRESENTATION OF STURGE-WEBER SYNDROME

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Background :- Sturge-Weber Syndrome is referred to as encephalofacial angiomatosis. It is one of phakomatosis and simultaneous involvement by developmental malformation of skin, eye and CNS. Incidence of Sturge-Weber Syndrome is 1/50,000 births.

Observation :- A 11 year female girl presented with sudden muscle weakness on the right side of body and skin lesion over left side of face since birth. On examination, Port wine stain seen over left side of face and left eye shows buphthalmos looks like ptosis. patient has no history of seizure, developmental delays and mental retardation but on examination her right sided body reflexes are slightly decreased. Ophthalmologist has diagnosed glaucoma over left eye.MRI of brain shows Rail track calcification over left temporo-occipital and parital region and ECG and CT Scan shows related findings.

Usually in Sturge-Weber Syndrome cutaneous finding is unilateral port wine stain. In most case neurological symptoms may find. Epilepsy occurs in 75-90% of cases and seizures is frequently associated with hemiplegia. Eye is involved in 50-60% of all cases. Treatment is symptomatic like laser treatment in used to lighten birthmark, anticonvulsant is used to control seizure. Educational therapy is prescribed for mentally retarted patient. Physiotherapy is useful for muscle weakness and prostaglandin or surgery used to reduce IOP.

Key Message :-Sturge-Weber Syndrome is not a hereditary disease but congenital malformation. The case is reported for its interesting clinical presentation and rarity.