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Letter to Editor

Skin-a Mirror of the Brain

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Keywords: Neurocutaneous Syndrome; Tuberous Sclerosis; Sturge-Weber Syndrome; Epilepsy; Adenoma Sebaceum; Hemangioma; Ash Leaf Macule

Abbreviations

TS: Tuberous Sclerosis; SWS: Sturge-Weber Syndrome; MRI: Magnetic Resonance Imaging; CT: Computed Tomography

The skin, because of its relationship with the sensory nervous system, autonomic and central nervous system, constitutes a neuroimmunoendocrine organ [1]. Therefore, many diseases also affect both the skin and the nervous system together; there are almost 300 of them. Among these are about 60 genetic diseases, which are called the 'neurocutaneous disorders' [2].

Case 1: A 12-year-old girl was brought with complaints of infantile onset epilepsy, increasing popular rashes over face since early childhood, poor scholastic performance and recent onset obsessive-compulsive behavior. On examination, she was noticed to have facial angiofibromas or adenoma sebaceum (Figure 1a) and multiple hypomelanotic macules (ash leaf macules) over skin (Figure 1b). Rest of the examination was within normal limits. MRI brain revealed multiple subependymal nodules (Figure 1c) and ophthalmological evaluation showed multiple retinal hamartomas. Renal, cardiac and pulmonary evaluations were non-contributory. A diagnosis of Tuberous sclerosis was made but genetic confirmation was not possible in view of non-availability.

Case 2: A 13-year-old developmentally normal girl presented with history of epilepsy since 1 year of age with current breakthrough seizure and right sided hemiparesis. There was also history of reddish rashes over face since birth. On examination, facial capillary malformation (Port-wine stain) was seen in the left paramedian region of the face (Figure 2a). CT scan of brain had tram-track calcification in the left occipital region (Figure 2b) suggesting the diagnosis of Sturge-Weber syndrome.

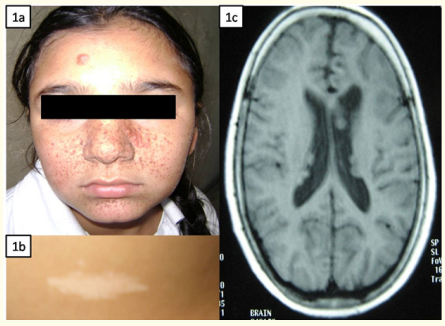

Figure 1: A 12-year-old girl with facial angiofibromas or adenoma sebaceum (1a), hypomelanotic macule (ash leaf macule) over skin (1b) and MRI brain showing multiple subependymal nodules (1c).

Figure 2: A 13-year-old girl with facial Port-wine stain (2a) and CT scan of brain showing tram-track calcification in the left occipital region (2b).

Tuberous sclerosis (TS) is an autosomal dominant condition with an estimated incidence of approximately 1 in 6000-10,000 live births. The two causative genes are TSC1 on chromosome 9q34 encoding for protein hamartin and TSC2 on chromosome 16 encoding for protein tuberin. The cutaneous lesions in TS includes hypomelanotic (Ash leaf) macule (90% present at birth or appear within first few years of life; 1 - 10 in number, 0.5 - 3 cm in diameter; disappear/ fade in adulthood and new lesions can appear at any age), facial angiofibromas (75 - 90% cases; onset at 2 - 5 years; concentrated over the alar grooves, extending symmetrically over the cheeks, nose and chin with relative sparing of upper lip and lateral face; may increase during puberty), fibrous facial plaque (2 - 40%; congenital of appears shortly after birth), shagreen patch (almost 50% cases; may present in infancy but can present later; initially looks like capillary hemangioma but there is palpable thickening of the dermis), ungular fibromas (usually found in adults), fibromas around the teeth or on the tongue, and molluscum fibrosum pendulum (skin tag like lesions, especially around the neck). The neurological manifestations are epilepsy (80 - 90% patients, often starting in infancy), subependymal giant cell astrocytomas (SEGAs) (10 - 15% cases), intellectual disability (approximately 50% cases), abnormalities in speech, autistic behavior and TSC-associated neuropsychiatric disorders. Other organs, such as lungs (lymphangioleiomyomatosis), heart (rhabdomyoma) and kidneys (angiomyolipoma) may also be affected [3].

Sturge-Weber syndrome (SWS) is caused by a sporadic somatic mutation in the gene GNAQ on chromosome 9q21; it affects 1 in 20,000 - 50,000 newborns. SWS is characterized by facial port-wine stain in association with ipsilateral leptomeningeal vascular anomalies with one or more symptoms of epilepsy, hemiparesis, intracranial calcifications and cerebral atrophy and glaucoma. Port wine stain is present at birth involving at least the first branch (Ophthalmic) of the V^{th} cranial nerve. The lesions are usually unilateral, but can be bilateral and can even have skip areas. MRI with contrast is the imaging modality of choice to detect leptomeningeal angiomas, whereas calcifications are best seen with CT scan of brain [4].

The above two cases illustrates that how in a patient with a disease with multisystem involvement, the diagnosis may get delayed due to lack of inter-specialty co-ordination. However, we hope that this report presenting such a late diagnosis of two neurocutaneous syndromes will encourage careful examination of the skin by the pediatrician, with consideration of referral to other specialties such as Dermatologist, for the early diagnosis of the patient.

Acknowledgements

Nil.

Conflict of Interest

Nil.

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