Subependymal Giant Cell Astrocytoma in Tuberous Sclerosis

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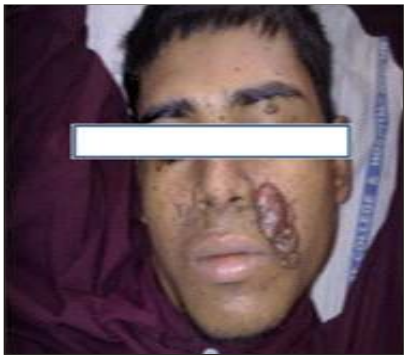

Figure 1 : **Adenoma Sebaceum**

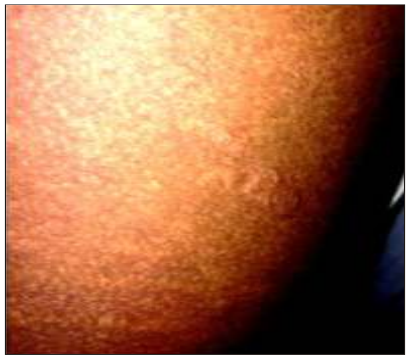

Figure 2 : **Ashleafs Macule**

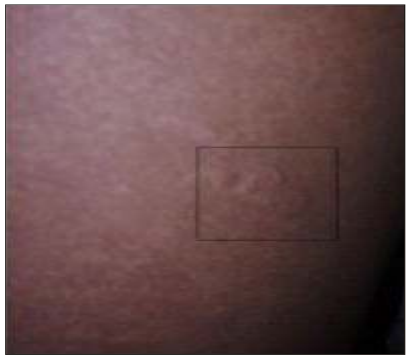

Figure 3 : **Shagreens Patch**

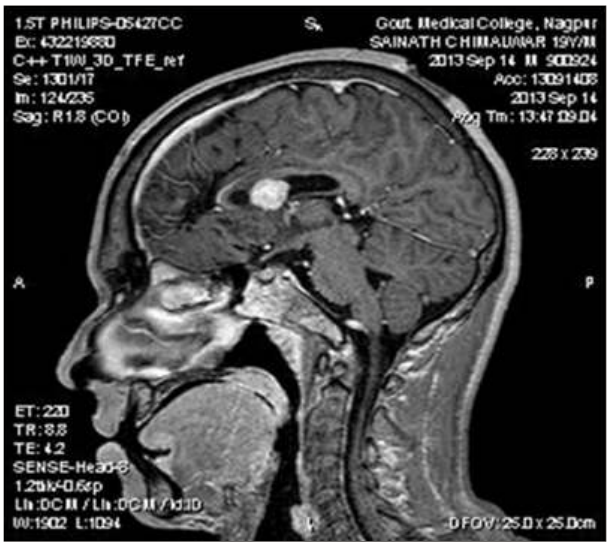

Figure 4 : **Subependymal Giant Cell Astrocytomas (SEGAS)**

19 years old male pt was admitted with complaint of generalized tonic clonic seizures 3-4 episodes since one day. Patient was a known case of mental retardation with seizure disorder on anticonvulsants since childhood. He had stopped anticonvulsant since 3-4 days.

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Examination showed adenoma sebaceum ***(Figure 1),*** Ashleafs macule ***(Figure 2)*** and Shagreens Patch ***(Figure3).*** His MRI was done which showed subependymal giant cell astrocytomas (SEGAS) an subependymal nodules (SENs) ***(Figure 4).***

Tuberous sclerosis complex (TSC) is a genetic disorder affecting cellular differentiation, proliferation, and migration early in development, resulting in a variety of hamartomatous lesions that may affect virtually every organ system of the body. Abnormal findings are seen in skin, nervous system, heart, kidneys, liver and gastrointestinal system, eyes, lungs, teeth and bones.

The best-known cutaneous manifestation of TSC is adenoma sebaceum ***(Figure 1),*** which often does not appear until late childhood or early adolescence. This lesion is an angiofibroma (ie, cutaneous hamartoma) and is not related to excessive sebum or acne. Flat, reddish macular lesions develop first, which can be mistaken for freckles early on. They become increasingly erythematous and papulonodular over time, occasionally with a friable surface that may bleed easily. Facial angiofibromas typically are noted first in childhood and exhibit progression during puberty and adolescence (see image above). Adenoma sebaceum may be disfiguring.

Other skin lesions consist of hypomelanotic (ie, ash leaf) macules ***(Figure 2),*** periungual or gingival

fibromas , and thickened, firm areas of subcutaneous tissue, often at the lower back (shagreen patch) ***(Figure 3)*** or forehead and face. Hypomelanotic macules are usually round or oval in shape and vary in size from a few mm to as much as 5 cm in length. Sometimes they have an irregular, reticulated appearance, as if white confetti paper had been strewn over the skin (confetti lesions). Hypomelanotic macules are a nonspecific finding and are not of themselves pathognomonic of TSC. Fibromas may occur in other locations. When present in the lumbar region they have been called a "shagreen patch." ***(Figure 3)*** The overlying skin may have an orange hue "Shagreen patches" are confined, however, to the subcutaneous tissue and are not associated with dysraphism, osseous lesions, or mass effects on neural structures.

Abnormal neurological findings result from the location, size, and growth of tubers and the presence of subependymal nodules (SENs) and SEGAs. ***(Figure 4)***

Tubers are noted most commonly in the cerebrum, without clear predilection for any particular lobe. They occur in the cerebellum as well, where they may be apparent only on microscopic examination. Rarely, they have been noted in the brain stem and spinal cord. The number, size, and location of tubers can vary widely from patient to patient. Depending on the location of tubers, neurological findings can include abnormalities in cognition (either global delays or specific location-related deficits like language delays), cranial nerves, focal motor / sensory / reflexes abnormalities, cerebellar dysfunction, or gait abnormalities.

SENs are noted about the wall of the lateral ventricles and may be either discrete or roughly confluent areas of firm, rounded hypertrophic tissue. SENs may occur anywhere along the ventricular surface, but most commonly occur at the caudothalamic groove in the vicinity of the foramen of Monro.

The generally benign SENs can degenerate into SEGAs in 5-10 % of cases. SEGAs can grow, often in an extremely indolent fashion, resulting in ventricular obstruction and hydrocephalus. Since this process occurs very gradually, patients may have marked hydrocephalus when they finally become symptomatic . In this situation, blindness or other permanent neurological deficit commonly ensues despite prompt neurosurgical intervention.

References :

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