STURGE WEBER SYNDROME: A CASE REPORT

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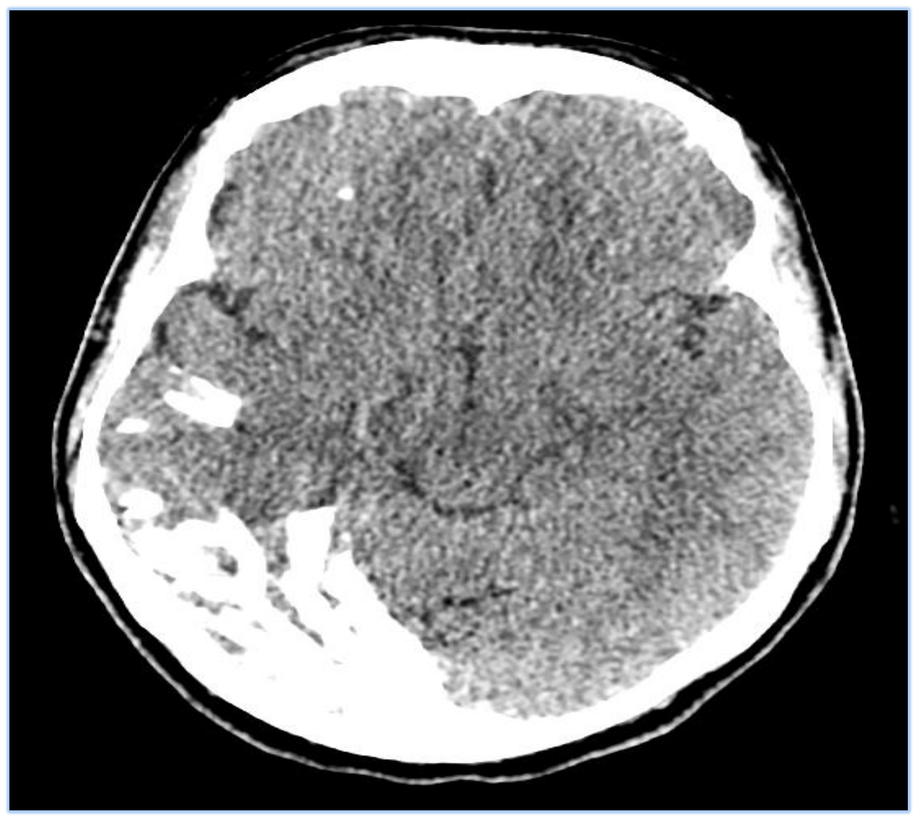
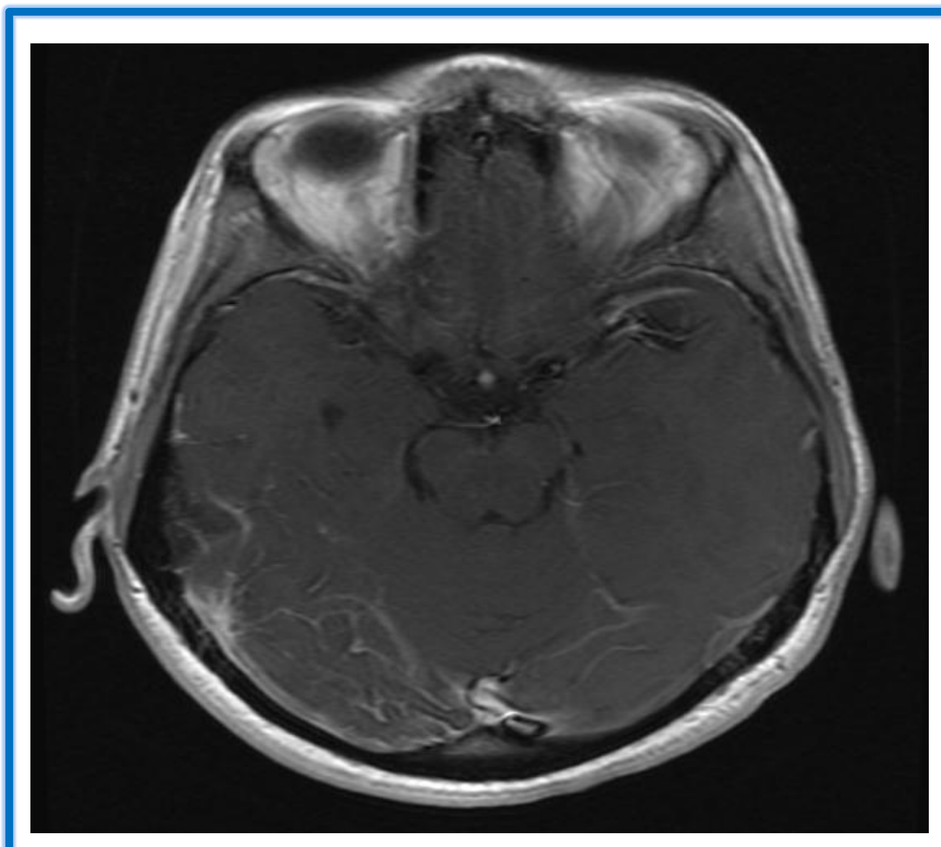
BACKGROUND

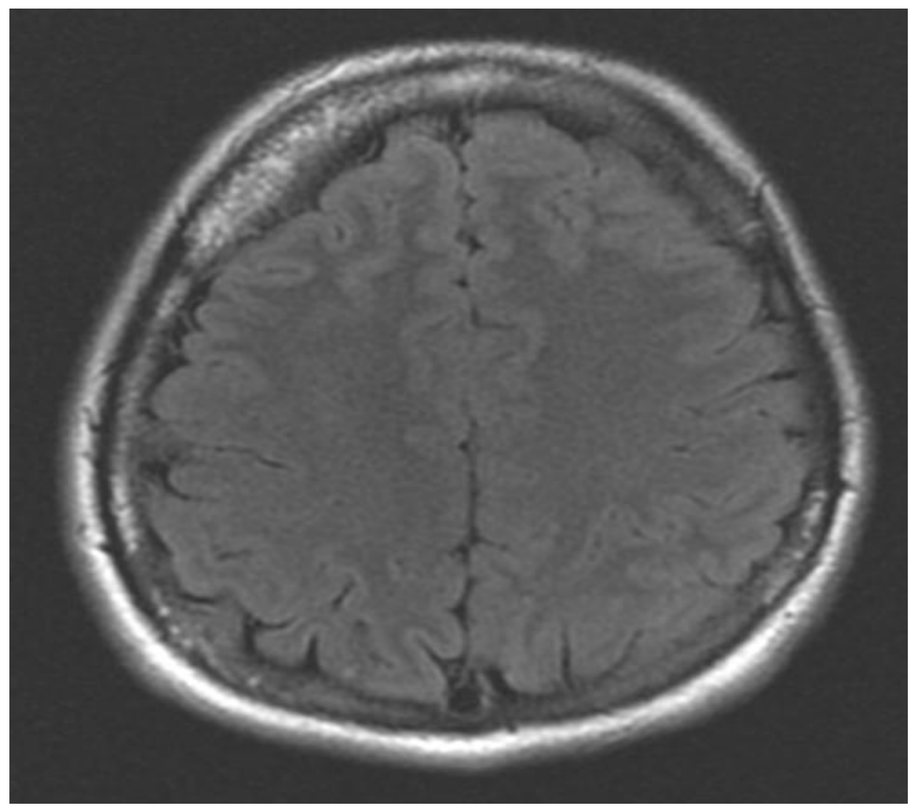
Sturge Weber Syndrome (SWS) or encephalo-trigeminal angiomatosis is a non-hereditary, non-familial, congenital and rare disorder of unknown aetiology. The condition belongs to the group of phakomatosis and is characterised by venous-capillary abnormalities involving the skin (facial port-wine stain, often localized in the distribution of the ophthalmic division of the trigeminal nerve), the central nervous system (leptomenigeal venous angiomas, often in the occipital and posterior parietal lobes) and the eye (glaucoma and choroidal hemangioma). Clinical neurologic manifestations are principally epilepsy, learning disabilities, motor and sensory deficits and headaches. According to the presenting symptoms, SWS is classified into three types (Roach Scale): neuro-oculo-cutaneous involvement (type I); facial angioma and glaucoma, no leptomeningeal angioma (type II); leptomeningeal angioma, no dermal and ocular involvement (type III).



CASE REPORT

A 27 years old Romanian female presented with a history of childhood-onset epilepsy. She had complex partial seizures, evolving sometimes to generalized tonic-clonic seizures. Even though she presented a normal cognitive development, she was affected by behavioural and personality disturbances since adolescence. Physical examination showed a port-wine stain involving the upper right part of the face. There were no focal deficit on neurologic examination. At the ophthalmologic evaluation there was no evidence of glaucoma, while ocular Indocianine Green angiography revealed the presence of choroidal hemangiomas on both sides. Cranial CT scan and brain MRI with gadolinium showed diffuse atrophy in the right hemisphere. CT scan revealed also cortical calcification in the right temporal and occipital areas. MRI with gadolinium demonstrated also the presence of leptomeningeal angiomas in the right temporal and occipital lobes and choroid plexus enlargement in the ipsilateral hemisphere. According to clinical and instrumental findings we made a diagnosis of SWS and antiepileptic treatment with levetiracetam was started.





CONCLUSION

SWS is an entity which occurs rarely and it may present with varied clinical features. SWS may have an incomplete and atypical presentation and the absence of classical triad of CNS, ocular and dermal impairment does not preclude the diagnosis. According to the Roach classification, our case was type I SWS, as the patient had neurological, cutaneous and ocular involvement. Despite the typical presentation, the impossibility to undergo specialized evaluation in Romania, caused a delay in the diagnosis. This case aims to underline the need to consider SWS also in adults.

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