A 40-year-old man with a diagnosis of Sturge–Weber Syndrome (SWS) was admitted to hospital with fever and urosepsis. The sequelae of this condition include seizures, intellectual disability, bilateral glaucoma, hypothyroidism, and diffuse port-wine stain. He was also noted to have several pyogenic granulomas and a large capillary malformation of the lower lip, with associated bleeding risk. Clinical images highlight the diagnostic cues and dermatologic manifestations that are associated with SWS. Management of patients with this condition requires multidisciplinary care of the neurologic, endocrine, and vascular complications of SWS, in addition to extensive psychosocial support.

Keywords: Sturge-Weber syndrome, dermatology, urology, pyogenic granuloma

A 40-year-old man with a diagnosis of Sturge–Weber Syndrome (SWS) was admitted to hospital with fever and urosepsis. The patient was diagnosed with Sturge–Weber Syndrome (SWS) and its sequelae, including history of seizures, intellectual disability, bilateral glaucoma, and diffuse port-wine stain (PWS). He was also noted to have several pyogenic granulomas and capillary malformation of the lower lip (Figure 1).

The patient had previously undergone surgical removal of pyogenic granulomas of the head and neck, which were associated with bleeding. Magnetic resonance imaging (MRI) revealed extensive bilateral leptomeningeal capillary-venous angiomatosis, a markedly atrophic right hemisphere, and prominence of bilateral choroid plexi (Figure 2). These
findings correlate clinically with left hemiparesis and associated joint contractures, secondary to encephalomalacia.

SWS is a rare, congenital, neurocutaneous syndrome that occurs due to cerebral vascular malformations during embryogenesis, resulting in a combination of facial PWS and vascular malformations of the central nervous system (leptomeningeal angiomatosis) with associated cortical atrophy.1

The disorder is associated with chronic cerebral hypoxia and complications such as seizures, progressive hemiparesis and hemiatrophy, intellectual disability, and glaucoma.1 MRI is the preferred diagnostic imaging modality and may reveal leptomeningeal angiomatosis with ipsilateral parenchymal atrophy and calcification, with bilateral involvement seen in 15% of cases.2

The differential diagnosis of SWS includes other syndromes, such as Klippel–Trenaunay–Weber and Osler–Weber–Rendu. Vascular malformations, including pyogenic granulomas as seen in this patient, can be associated with SWS and require a multidisciplinary approach given the risk of bleeding. SWS is also associated with an increased prevalence of growth hormone deficiency and hypothyroidism.3 This patient has confirmed hypothyroidism and short stature, but no prior diagnosis of growth hormone deficiency.

Management of SWS is focused on controlling neurologic and ocular sequelae of the disorder through medical and surgical techniques, as well as providing comprehensive psychosocial support. Vascular and dermatologic malformations may have a high associated bleeding risk. Bilateral leptomeningeal angiomatosis is associated with higher seizure risk and more severe intellectual and functional disability. Adult patients should also be evaluated for endocrinologic sequelae, as treatment of growth hormone deficiency and hypothyroidism can improve patient morbidity and quality of life.

Disclosures

The authors received no financial support for the research, authorship, and/or publication for this article. The authors have no competing interests.
References


Permissions

The authors have confirmed that informed consent was obtained from the patient and his or her substitute decision-maker for use of the images and information.

Acknowledgments

The authors would like to acknowledge Dr. Arun Mensinkai for providing input on optimal MRI image selection for the case.