Sturge-Weber Syndrome: An Extensive Cutaneous Presentation

Síndrome de Sturge-Weber: Uma Apresentação Cutânea Extensa

Regina CALDAS, Maria GUIMARÃES, Olga FERREIRA

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A 40-year-old woman presented with a congenital and generalized port-wine stain. Her medical history was relevant for bilateral glaucoma and seizures of infancy. The physical examination revealed multiple purple to red patches, with irregular, well-defined borders, spreading bilaterally from the face (Fig. 1) to the trunk, right upper limb (Fig. 2) and thighs, without body asymmetries. The magnetic resonance imaging showed slight atrophy of the temporo-occipital right lobe, leptomeningeal enhancement and hypertrophy of the choroid plexus. The clinical and imaging findings supported the diagnosis of Sturge-Weber syndrome.

The patient was referred for pulsed dye laser and maintained multidisciplinary follow-up.

Sturge-Weber syndrome is caused by a somatic mosaic mutation in the GNAQ gene, which induces malformations in the cephalic embryonic vasculature. It is characterized by a facial port-wine stain, often unilateral and rarely extra-facial, leptomeningeal angiomatosis and glaucoma. Skin involvement significantly affects the quality of life of patients. Nevertheless, neurological and ocular complications are the main prognostic determinants.

REFERENCES