

Encephalofacial Angiomatosis (Sturge-Weber Syndrome)

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Encephalofacial angiomatosis is more popular known as Sturge-Weber syndrome; it is also sometimes referred to as Kalischer syndrome, Parkes-Weber syndrome, and Sturge-Weber-Dimitri syndrome. Those with this congenital disorder have abnormal blood vessels, port-wine stains on their faces, glaucoma, and seizures. The other symptoms include intellectual disability, paralysis, developmental delay, and muscle weakness. This is specifically caused by the random mutation of the guanine nucleotide-binding protein G(q) (GNAQ) gene.

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