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Original Article

Leveraging a Sturge-Weber Gene Discovery: An Agenda for Future Research

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ABSTRACT

Sturge-Weber syndrome (SWS) is a vascular neurocutaneous disorder that results from a somatic mosaic mutation in GNAQ, which is also responsible for isolated port-wine birthmarks. Infants with SWS are born with a cutaneous capillary malformation (port-wine birthmark) of the forehead or upper eyelid which can signal an increased risk of brain and/or eye involvement prior to the onset of specific symptoms. This symptom-free interval represents a time when a targeted intervention could help to minimize the neurological and ophthalmologic manifestations of the disorder. This paper summarizes a 2015 SWS workshop in Bethesda, Maryland that was sponsored by the National Institutes of Health. Meeting attendees included a diverse group of clinical and translational researchers with a goal of establishing research priorities for the next few years. The initial portion of the meeting included a thorough review of the recent genetic discovery and what is known of the pathogenesis of SWS. Breakout sessions related to neurology, dermatology, and ophthalmology aimed to establish SWS research priorities in each field. Key priorities for future development include the need for clinical consensus guidelines, further work to develop a clinical trial network, improvement of tissue banking for research purposes, and the need for multiple animal and cell culture models of SWS.

Keywords: Sturge-Weber syndrome, GNAQ, somatic mutation, port-wine birthmark, glaucoma, seizures

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