



## Sturge-Weber Foundation Announces Major Leap Forward In Sturge-Weber Syndrome Research

### *Brain Vascular Malformation Consortium*

For Immediate Release

MT. FREEDOM, N.J./EWorldWire/Oct. 14, 2009 --- The Sturge-Weber Foundation (SWF) today announced it will actively participate in the Brain Vascular Malformation Consortium, which will focus on three related disorders - Sturge-Weber syndrome (SWS), Hispanic mutation familial cavernous malformations, and hereditary hemorrhagic telangiectasia.

The inclusion of Sturge-Weber syndrome and the SWF in this prestigious research effort marks a significant leap forward for SWS research. The foundation has maintained a database of natural history since 1987 which provided documentation for further studies on DNA and angiogenesis in SWS.

"This grant has been made possible because of many dedicated families, volunteers, physicians and crucial financial support of the SWF's endeavors to improve the quality of life for our members," said Karen Ball, president and CEO of the SWF. "With their enduring commitment and financial support, the SWF will continue to lead the fight on behalf of our members to increase the pace of discovery in our rare disease."

The consortium is part of the newly-announced expansion of the Rare Diseases Clinical Research Network (RDCRN), which is under the umbrella of the Office of Rare Diseases Research (ORDR) at the National Institutes of Health (NIH). NIH recently announced that 19 new and returning consortia will be awarded \$117 million for research.

The RDCRN was created to address the difficulties inherent in rare disease research. The RDCRN fosters collaboration amongst researchers, actively involves patient advocacy groups in the research process and creates a specialized infrastructure to support rare disease research.

The Brain Vascular Malformation Consortium primary research goal for SWS will be to determine whether certain substances called "biomarkers" found in the blood and urine of patients can be used to predict the progression of the syndrome. For SWS, the biomarkers in question are related to angiogenesis, the process by which new blood vessels are formed.

Researchers believe that dysregulated angiogenesis plays a significant role the development and progression of the neurological symptoms associated with SWS. Ultimately, this research may potentially lead to the development of a safe, screening tool that can predict which SWS patients will remain relatively stable and those who will have a more severe course of disease. This, in turn, can potentially enable doctors to know which patients may require more aggressive treatment earlier in the course of the disorder.

The inclusion of SWS in this important research network is the culmination of years of hard work and networking by Karen Ball with the directors of the NIH and National Institute of Neurological Disorders and Stroke (NINDS) clinical and scientific investigations and the executive directors of the Angioma Alliance and Hereditary Hemorrhagic Telangiectasia Foundation International - the patient advocacy groups who serve the other disorders being studied in the consortium.

## About Sturge-Weber Syndrome

Sturge-Weber syndrome is a rare, congenital but sporadic (non-inherited) disorder characterized by brain, ocular and cutaneous vascular anomalies. The specific symptoms of SWS are highly variable in each case but can potentially include a port-wine birthmark, leptomeningeal angioma, epilepsy, stroke-like episodes, headache, glaucoma, choroidal angioma, and developmental delays. The underlying mechanisms that cause this complex syndrome remain unknown.

## About the Sturge-Weber Foundation

The Sturge-Weber Foundation, founded in 1987, is an international organization serving individuals and families affected by Sturge-Weber syndrome and other port wine birthmark conditions with education, research, public information and friendly support.

Learn how to support the activities of the Sturge-Weber Foundation, visit [Sturge-Weber.org](http://www.sturge-weber.org) ('<http://www.sturge-weber.org>') or call 973-895-4445.

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