WE DID IT!

Researchers Uncover Cause of Sturge-Weber Syndrome

Discovery of Genetic Mutation Published Today

After 25 years of searching, the Sturge-Weber Foundation (SWF) is thrilled to join patients, physicians, researchers and government agencies in celebrating the identification of the gene mutation responsible for Sturge-Weber syndrome and port-wine birthmarks. The new research was published online today in *The New England Journal of Medicine*.

> We applaud researchers for working with us to bring new information and new hope to people affected by Sturge-Weber syndrome. We are excited to lead and support continued research to translate this science into new treatment approaches.
>
> - Karen L. Ball, president and chief executive officer of the SWF

The SWF played an early and critical role in getting this research off the ground. Researchers from the Brain Vascular Malformation Consortium (BVMC) identified a mutation in gene *GNAQ* on chromosome 9q21 in three individuals with Sturge-Weber syndrome, and confirmed this finding in 23 additional patients. The genetic mutation is somatic, which means it occurs after conception and is not inherited or passed on to children.

KUDOS

This discovery is a remarkable tribute to the Sturge-Weber community, starting with the patients who donated tissue samples for analysis. We are privileged to be part of this rare and brave community, and we are grateful to the researchers who worked tirelessly on this ground-breaking discovery. Together, we can do great things. Together, we will translate this research into new treatment approaches for Sturge-Weber syndrome.

LEARN MORE

Watch the video message to SWF members from Jonathan Pevsner, Ph.D., Director of Bioinformatics at the Kennedy Krieger Institute and co-senior author of the study.

Attend the13th Sturge-Weber Foundation International Conference July 25 – 27, 2013 in Denver to meet the researchers in person and discuss the research and its implications.

Read the SWF news release

Read the *NEJM* article

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