Suicide Screening in Sturge-Weber Syndrome: An Important Issue in Need of Further Study. Ped Neurol 2020 Mar. This study analyzed the results of suicide screening for patients seen at our SWS Center, as compared to general neurology patients seen at Kennedy Krieger. The study found that patients with SWS were more likely to report suicidality than were general neurology patients. These results emphasize the importance of this issue for patients with SWS, the need to screen for suicidality and urges further study of mood disorders and suicidality in SWS.

Identification of a Mosaic Activating Mutation in GNA11 in Atypical Sturge-Weber Syndrome. J Invest Dermatol 2020 Aug. About 90% of all SWS tissue samples in reported studies have the somatic mutation we discovered in 2013. We analyzed tissue samples and found another mutation associated with SWS: a somatic mutation in a gene called GNA11. GNA11 and GNAQ are similar in sequence and the proteins they code for have similar functions. This discovery adds to our understanding of the causes for SWS and provides further insights into the role of genetic testing in SWS.

Manuscripts for the following studies are being prepared:
- Medication in SWS
- Vitamins in SWS
- Pre-symptomatic treatment of SWS
- Protein expression SWS brain tissue
- Mouse Model of SWS

EDUCATIONAL EFFORTS:
Third Annual Family Symposium
This year the symposium was held virtually and was very successful; more than 80 participants registered for the webinar. Talks reviewed our recent publications; discussions on the first year after diagnosis; COVID-19 and school; our multidisciplinary clinic; mood disorders and Epilepsy; and fundraising. Next year we hope to be back in person combined with virtual access for participants.

We look forward to keeping you updated on our progress! If you would like information about research studies, please contact Dr. Comi at 443-923-9127 or via email at comi@kennedykrieger.org.