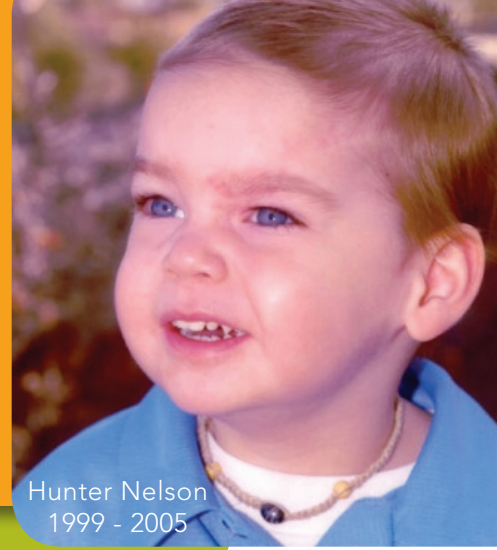


# Updates from THE HUNTER NELSON STURGE-WEBER CENTER



Hunter Nelson  
1999 - 2005

Dream. Discover. Cure.

## How Does a Multidisciplinary Visit Work?

The Hunter Nelson Sturge-Weber Center is dedicated to caring for patients with Sturge-Weber syndrome. The Center offers multidisciplinary visits to families that want to see our specialists. We are, therefore, fortunate to have Dr. Anne Comi, neurologist and director of the Hunter Nelson Sturge-Weber Center; Dr. Elizabeth Turin, behavioral specialist; Dr. Emily Germain-Lee, endocrinologist; Dr. Stacy Suskauer, medical rehabilitation specialist, Dr. Bernard Cohen, dermatologist; Dr. Eric Kossoff, epileptologist; Dr. Henry Jampel, ophthalmologist; and Dr. Andrew Zabel, neuropsychologist.

New patients' multidisciplinary visits are coordinated through the Center by the clinical coordinator. Several appointments may be scheduled over a couple of days to a week with the specific doctors that the family wants to see. Families have the opportunity to schedule a certain day and time to visit with doctors from the Kennedy Krieger Institute and Johns Hopkins Hospital in a few locations. With each doctor that they are scheduled to see, patients have an exam, acquire information, and receive recommendations.

Once all of the appointments have been confirmed with the families, a previsit packet of information is sent to the residence. The packet includes a customized appointment letter, map of hospital, local driving directions, local hotel list, and a new patient questionnaire. The customized appointment letter contains the dates and the times of the patient's upcoming appointments, doctors he or she will be seeing, locations of the visits, and what the family should bring to the appointments.

New patients are encouraged to complete the new patient questionnaire before they come for their appointment with Dr. Comi, and other doctors at the Center may also have questionnaires for new patients to complete. The information gathered from the new patient questionnaires gives the doctors an overview of the patient's medical history, family history, and current symptoms. Also, a review of the patient's neuroimaging, neurology, dermatology, ophthalmology records, and other records helps the doctors to make decisions about treatment and care.

## MAKING AN INTERNATIONAL APPOINTMENT

When a family overseas is interested in coming to the Hunter Nelson Sturge-Weber Center, the international patient is sent both consent and information forms for the family to sign and complete, in addition to a credit card authorization form. Families also receive an appointment request form, where they can enter their demographic information, medical information, who referred them, and dates they are available. The family then faxes the forms back to the Center at 1-443-923-9160. A contact from the Johns Hopkins International Office at the Kennedy Krieger Institute conducts the intake for international families. The clinical coordinator of the Hunter Nelson Sturge-Weber Center will coordinate Kennedy Krieger Institute appointments.

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During visits with Dr. Comi, new patients receive a "visit packet" that includes Dr. Comi's business card, a center description sheet, a Sturge-Weber syndrome care guide, and relevant information about the syndrome. During the consultation, Dr. Comi examines the patient, answers questions, and offers recommendations for the patient's treatment, so that patients and their families get an understanding of what their next steps should be. Dr. Comi and her research assistants also invite patients to participate in ongoing research studies. For more information about the Hunter Nelson Sturge-Weber Center and its research, please review the Center's website at [www.sturgetweber.kennedykrieger.org](http://www.sturgetweber.kennedykrieger.org).

Kennedy Krieger Institute provides care in accordance with all constitutional rights and without discrimination as to race, color, sex, age, national origin, religion, marital status, sexual orientation, genetic information, physical or mental disability, veteran status, or sources of payment for care. Additional safeguards include the preservation of personal dignity as well as cultural, psychosocial, spiritual, and personal values, beliefs, and preferences.



Hunter Nelson Sturge-Weber Center  
at Kennedy Krieger Institute

*Dream. Discover. Cure*

## A Conversation With Dr. Joshua Ewen

Dr. Ewen presented a co-authored Quantitative EEG (qEEG) paper at the February Sturge-Weber Center meeting. Dr. Ewen started his fellowship at the Kennedy Krieger Institute and Johns Hopkins Hospital in 2002, and began working with Dr. Comi at the Hunter Nelson Sturge-Weber Center in 2005. Here's what he had to say:

**Q: How did you become interested in helping with the Center?**

A: I was interested in electrophysiology, and Dr. Comi had a research opportunity for me to work on her qEEG study. Dr. Comi's qEEG study is part of her "Multidisciplinary protocol to address the pathophysiology of Sturge-Weber syndrome."

**Q: Describe your research.**

A: Quantitative EEG refers to using computers to process the EEG signal and give us numbers to assign to various aspects of the signal. Typically, EEGs are read by neurologists, whose experience and judgment goes into deciding whether the EEG is normal or abnormal.

We are looking at the use of qEEG to predict which babies with a port-wine birthmark will go on to develop Sturge-Weber syndrome brain involvement. We are also looking to see if qEEG will be helpful in predicting which babies will have a good response to medications, such as aspirin. If qEEG is shown to work, it will help us know which babies to give medication to, because the medication will help, and which babies not to give medication to.

**Q: Who do you see benefiting the most from your research findings and why?**

A: So far, it seems most likely that children under two years, who have not yet shown signs of Sturge-Weber syndrome, will benefit, if we can show reliably that qEEG can differentiate between those babies who will go on to develop Sturge-Weber syndrome and those who will not.

**Q: Where do you see your research in the future?**

A: There is a possibility that this line of research could be applied to other problems in Sturge-Weber syndrome as well as other clinical conditions.

*For more information, to arrange a clinical visit, participate in research, or see how else you can help, please contact Dr. Anne Comi at 443-925-9150 or email her at [comi@kenedykrieger.org](mailto:comi@kenedykrieger.org).*



## Summary Briefs: Recently Published Research or Review

Comi AM, Bellamkonda S, Ferenc LM, Cohen BA, Germain-Lee EL. Central hypothyroidism and Sturge-Weber Syndrome. *Pediatric Neurol.* 2008 Jul; 39(1):58-62.

Two (2.4 percent) out of 83 patients with Sturge-Weber syndrome (SWS) and brain involvement seen at the Hunter Nelson Sturge-Weber Center were found to have central hypothyroidism. We wrote a case series on those patients.

Subject 1 was a 10-year-old female with Sturge-Weber syndrome diagnosed at six years of age. She had a right side facial port-wine birthmark, left-sided weakness; right-sided brain involvement of the parietal, occipital, and temporal lobes; epilepsy, right-eye glaucoma, severe headaches; and a family history of hypothyroidism in maternal and paternal grandmothers. After levothyroxine replacement, her thyroid levels became normal. The patient was once again seizure free, her skin improved, her energy returned, her headaches improved, and her hair loss slowed down.

Subject 2 was a 12-year-old male with Sturge-Weber syndrome. He had a port-wine birthmark on the left side of his face and body, left eye glaucoma, and left side brain involvement. The subject experienced many seizures, stroke-like episodes, learning difficulties, and gross and fine motor problems. The patient's maternal grandmother was diagnosed with hypothyroidism. He had medically refractory seizures partially controlled with a vagal nerve stimulator and multiple seizure medications. After levothyroxine therapy, his thyroid

levels became normal. His seizures did not improve; however, he became more interactive and progress in school improved.

As part of clinical care, we recently started screening our patients for central hypothyroidism. Our patients may have central hypothyroidism due to the anticonvulsants that they take. Patients with Sturge-Weber syndrome are prone to growth-hormone deficiency, and this also could interfere with the hypothalamic-pituitary axis. It may be difficult to know what caused the patient's inattention, behavioral issues, sleepiness, headaches, and obesity. The symptoms may result from seizures, anticonvulsants, behavioral problems, and Sturge-Weber syndrome-related brain injury, in addition to possible central hypothyroidism. We recommend that healthcare providers annually check the thyroid function (blood for TSH and free T4) of their patients with Sturge-Weber syndrome.

*"We really appreciated the thoroughness of the doctors. They took the time to explain [procedures] and answer all our questions."*



# What Our Families are Saying

"Dr. Comi was very helpful in explaining the details of our child's diagnosis in a very caring manner and gave us clear direction for a future course of action."

"Meeting with the doctors gave my husband and me great peace of mind that we are doing everything we can for our daughter. Traveling to Baltimore was well worth the trip!"

"The doctors were warm, approachable, and friendly. They took their time with me and my son during our visits."

"Every physician and specialist went out of his or her way to make us feel valuable and welcomed."

"[The specialists] showed care for our family and communicated information about Sturge-Weber Syndrome thoroughly and with sensitivity."

"We appreciated meeting with all the doctors, who were not only knowledgeable, but nice and very concerned about our daughter's welfare."



## MAKING AN INTERNATIONAL APPOINTMENT continued

After the proper forms are completed and signed, the international office contact from the Kennedy Krieger Institute communicates with the International Office at Johns Hopkins Hospital, so that a patient services coordinator can be selected to coordinate the Johns Hopkins appointments. The patient services coordinator also assists with hotel accommodations, appointment scheduling, and language interpretation. Families send their medical reports and films to the Johns Hopkins Medicine International Office, and the office ensures that the Center receives copies. The patient services coordinator confirms appointments with the families.

The Center doctors see the patient with their appointed interpreters who are fluent in more than 20 languages. The doctors examine the patient and explain their recommendations and medical advice to the family through the interpreter, in addition to answering the patient's and family's questions.

International patients and their healthcare providers often wonder what the next step should be. Because many doctors in other countries may have never seen patients with Sturge-Weber syndrome, it is important that families share their new information with their doctors, so that the patients can be treated more effectively.

After the appointments are completed, if the family requests so, the International Office provides families with copies of the doctors' clinic notes from the Center and, if they are needed, follow-up appointments. On a very personal level, the Johns Hopkins International Office assists and accommodates international families in any way that is required.

