The Sturge-Weber Foundation
The stronger the wind, the tougher the trees

Sturge-Weber Syndrome: General Considerations

The Sturge-Weber Syndrome (SWS), which affects both sexes and all races, consists of the triad of vascular malformations of the skin, eye, and central nervous system. The authors Schirmer, Sturge, Weber and Dimitri have all contributed in various aspects to initial descriptions of the disorder. The intracranial malformation in SWS is a leptomeningeal venous angioma which is typically unilateral,but may (in 10%-15%) be bilateral. It can affect the whole hemisphere or may be restricted to a portion of the hemisphere. The neurological features of SWS may include epilepsy, hemiparesis contralateral to the port-wine stain, hemiatrophy, hemianopia and mental retardation. Glaucoma, dental abnormalities and skeletal lesions may also be present.

Port-wine stains occur with an incidence of 3 per 1,000 newborns, and are therefore not uncommon. When it involves the forehead and upper eyelid, the brain lesion (angioma) occurs in 10%-20% of cases. The incidence of intracranial involvement is much less when the port-wine stain involves only the lower face and trunk. Klippel-Trenaunay-Weber syndrome refers to those individuals with extensive somatic involvement of the limbs and trunk, such that there is enlargement of the involved extremities.

Glaucoma is seen in 30%-70% of SWS patients. Buphthalmos refers to a congenital enlargement of the globe resulting from glaucoma.

The first neurological manifestation in SWS is usually the emergence of seizures in various forms. Seizures eventually will affect 72%-80% of those with unilateral leptomeningeal angioma, and up to 90% of those with bilateral intracranial involvement. In those destined to have seizures, 75% will begin to have them in the first year of life, 86% by age 2 years, and 95% by age 5 years. About 50% of SWS patients have mental retardation, but this is far more likely to occur if the seizures are persistent and poorly controlled in the early years of life. In many cases, mental retardation can be prevented by aggressive management, including neurosurgical intervention, but the hemianopia and hemiparesis cannot be prevented. Hemiparesis can be slowly progressive or may occur in a series of stroke-like episodes which may or may not be associated with a cluster of seizures.

Early neuroimaging studies in SWS were performed mainly for diagnostic purposes, but recent advances have allowed the use of neuroimaging to be applied in how best to treat the disorder and to gain a better understanding of the mechanism of brain injury. Thus, CT, MRI, MRA, MRS, PET and SPECT scans have all been applied in the study of SWS.

Treatment is aimed mainly at control of seizures and glaucoma, but other supportive therapies (see below) are also used. Early neurosurgical intervention is indicated when the seizures are uncontrolled in order to prevent a declining cognitive state and mental retardation. The use of aspirin and other antiplatelet agents is controversial, and is not based on solid scientific data.

Several International Conferences on SWS emphasized the crucial involvement of basic scientists if the etiology and pathophysiology of SWS are to be elucidated. The Foundation needs to stimulate the interest and solicit the involvement of basic scientists who work in the areas of angiogenesis and neuroprotection, and provide these individuals with biological specimens and research funding. These areas of research must be pursued diligently in the hope of an eventual cure for SWS.

STURGE-WEBER SYNDROME: THE YOUNG PATIENT

Babies born with a port-wine stain involving the face should be checked for possible glaucoma. In the absence of neurological signs or symptoms, simply observing and examining the infant at regular intervals may be sufficient. If further evaluation is indicated, an EEG (to determine the presence of epileptiform activity and/or background asymmetry) and CT scan (to determine the presence of calcifications) will be informative.

The best method to diagnose a leptomeningeal angioma and to determine its extent is to perform an MRI scan with gadolinium, but the study may be falsely negative if it is performed too soon in infancy, and the test may have to be repeated after one year of age. In addition to revealing the location and size of the angioma, MRI may show an accelerated myelination pattern in the affected cerebral hemisphere. This finding is undoubtedly related to the pathophysiology of SWS, but the explanation for this curious finding is still unexplained almost two decades after its original description.

Perhaps related to the accelerated brain myelination pattern seen on MRI is the transiently increased glucose metabolism seen on PET scans and increased cerebral blood flow on SPECT scans in the affected hemisphere. Increased glucose metabolism and blood flow are not related to seizure activity during the scans because they are observed even before the onset of seizures. Gradually, as the affected brain regions calcify, decreased glucose metabolism and blood flow will be seen on the scans. The time course of these changes vary among patients.

We have recently observed, using PET scanning, that severe hypometabolism of the affected hemisphere occurring rapidly after birth is associated with a better cognitive and seizure prognosis than a persistent mild hypometabolism. Rapid demise of the affected hemisphere at an early age will trigger the opposite hemisphere to undergo reorganizational changes resulting in relatively intact cognition. In a sense, these children have undergone an ‘autohemispherectomy’. Mild hypometabolism, on the other hand, implies abnormal function that is persistent and continues to generate seizures or exert a negative influence on the normal hemisphere, preventing it from undergoing optimal reorganization. It is these children who are at highest risk for developing mental retardation, which can be prevented with aggressive management. Young children with SWS and intractable epilepsy should be evaluated for focal resection or hemispherectomy in order to prevent or minimize the mental retardation. If hemiparesis is advanced, hemispherectomy should be performed. However, if hemiparesis is mild, the motor cortex should be preserved even at the risk of a subsequent second surgery. Parents should insist that the resected tissue be banked and distributed to basic scientists involved in research on SWS.

We believe that children whose seizures are not all that intractable but who show progressive developmental delay should also be evaluated for surgery in order to preserve cognitive function by ‘forcing’ the opposite hemisphere to undergo reorganization. This is most likely to be successful in the first 3 or 4 years of life when brain plasticity is at a high capacity.

Physical, occupational and speech therapy should be prescribed when indicated. The choice of anticonvulsants should take into consideration potential cognitive and sedative side effects.

STURGE-WEBER SYNDROME: THE OLDER CHILD

The major issues to be dealt with in the older child with SWS are: glaucoma, hemiparesis or hemiplegia, seizure control, learning difficulties, self-esteem, self-care skills, and headaches. Surgery for epilepsy, when indicated, should have been performed already in the younger patient but, occasionally, focal resections are performed in the older child. These children typically have small angiomas. Anticonvulsant medications should be carefully selected in order not to affect school performance. The newer anticonvulsants hold great promise in this regard, but require further study.

Physical therapy should be offered with the goal of maximizing function of the hemiparetic side and preventing contractures. Occupational therapy should also be prescribed as indicated to promote self-care skills. Some children will require psychiatric consultation if low self-esteem or depression are evident. Social adjustment should be carefully monitored. Neuropsychological testing performed at regular intervals will allow ongoing assessment of cognitive status and assist in optimization of the educational approach on an individual basis. Higher education should be encouraged when appropriate.

Headaches seem to occur more commonly in SWS than in the general population, and these should be treated symptomatically. The etiology of headaches in SWS, and whether mechanisms unique to SWS are at play, is not clear and requires further study.

HARRY T. CHUGANI, M.D.
Professor and Chief, Division of Pediatric Neurology
Director, PET Center
Children's Hospital of Michigan

The Sturge-Weber Foundation
PO Box 418 - Mt. Freedom, NJ - 07970 - USA
800-627-5482 - www.sturge-weber.org