Sturge-Weber Syndrome

Definition

Sturge-Weber Syndrome is a neurocutaneous (brain-skin) disorder characterized by three features:

1. Port wine stain (cutaneous facial angioma)
2. Seizures and other neurologic complications associated with angioma of the leptomeningies
3. Glaucoma (increased pressure within the eyeball.)

Introduction

Sturge-Weber Syndrome (also called encephalotrigeminal angiomatosis) is an uncommon condition. It occurs in 1:50,000 to 1:60,000 births. The cause is unknown but some scientists think it is the result of abnormal development of the primordial vascular bed (blood supply) of the brain very early in the formation of the embryo. Most cases are sporadic but occasionally cases within families are reported. Males and females seem to be equally affected. It has been reported in individuals of White, Hispanic, African and Asian heritage.

The facial port wine stain is present at birth in 98% of individuals with Sturge Weber Syndrome. In general the port wine stain tends to be found in the area of the face enervated by the fifth cranial nerve (trigeminal nerve.) It tends to be unilateral (on one side or the other) but can be bilateral (on both sides.) Port wine stain may also be found on the trunk or extremities of some individuals with Sturge Weber Syndrome. Not all individuals with port wine stain have Sturge Weber.

Seizures and other neurologic complications are the result of leptomeningeal angioma (vascular malformations in the lining of the brain.) Leptomeningeal angioma are present in 100% of individuals with Sturge Weber Syndrome. Seizures occur in 83% of individuals with Sturge Weber Syndrome and may be extremely difficult to control. Seizures may have their onset any time from birth to adulthood. Individuals with seizures are more likely to have required special education for mental retardation or developmental delay, and to have emotional and behavior problems. Other neurologic complications include:

1. Mental retardation or developmental delay in 40-50% overall and 60-70% of individuals with seizures.
2. Hemiplegia (weakness on one side of the body) in 30%
3. Hemiatrophy (smaller size on one side of the body)
4. Headaches in 62% of adults. Headaches may be associated with aura, nausea/vomiting, slurred speech, dizziness or feelings of facial pulsation.
5. Emotional problems – 50% of individuals with normal intelligence and Sturge Weber Syndrome have emotional problems, primarily depression but also anxiety, low self-esteem, shame, emotional outbursts and isolation.
6. Behavior problems in 85% of individuals with mental retardation and Sturge Weber Syndrome have behavior problems that include violence or aggression toward others and self- injurious behaviors.

Glaucoma (increased pressure within the eyeball) is present in 60% of individuals with Sturge Weber Syndrome. It can be present at birth or occur anytime throughout the lifespan. It is unilateral (on only one side) 74% of the time and bilateral (on both sides) 26% of the time. Untreated, glaucoma can cause blindness. It can be extremely painful and may be a "silent" cause of behavior outbursts or self-injurious behavior in non- verbal individuals with Sturge Weber Syndrome. Regular eye examinations and a high index of suspicion between appointments are crucial for these individuals.

Diagnosis

Diagnosis is made by the presence of a facial port wine stain and evidence of leptomingeal angioma either by skull X-ray or CT scan that show intracranial calcifications.

Prevention and Treatment

No preventative measures are known.

Port wine stain may be camouflaged with make up or treated with laser surgery or cryotherapy.

Seizures may be very difficult to control. Anticonvulsant medication, one or a combination of two or more, may be prescribed by the PCP or by a consultant neurologist. In either case, the individual will need to be monitored for drug side effects and drug-drug interactions on a regular basis. A much less commonly used treatment method involves surgical removal of a large portion of the brain (hemispherectomy.) This is a drastic approach. In some cases improvement in both seizure control and behavior problems have been reported. However, other surgeons report major post-operative problems and little improvement.

Hemiparesis and hemiatrophy may require regular physical therapy and occupational therapy to maintain and/or improve function. Splinting or bracing may also be helpful.

Headaches may respond to simple treatment methods like Tylenol or ibuprofen. Vascular or migraine type headaches may respond to preventative measures (anti-depressants, beta-blockers, calcium channel blockers, ergots, anticonvulsants, periactin) or to abortive agents (NSAIDs, Midrin, opiods, Triptan, ergots, dopamine antagonists, steroids.)

Depression and other emotional problems may require antidepressant medication and/or psychotherapy.

Behavior problems may respond to behavior management programs. Remember, pain may be a driving force behind "behavior" problems. Common causes of pain, especially glaucoma in individuals with Sturge Weber Syndrome must be ruled out. Other common causes for pain include Gastroesophogeal Reflux Disease (GERD) headache (see above), sinus infections, ear infections, dental problems and undiagnosed bone fractures especially if the individual has osteoporosis.

Glaucoma can be treated with eyedrops, pills, laser surgery, eye operations or a combination of methods. The need to keep regular appointments with the individual's opthalmologist cannot be overstated.

Emergency Situations – What can go wrong?

Seizures can lead to:

- **Status epilepticus** – prolonged seizure activity such as a seizure that lasts for more than 10 minutes or several seizures that occur one after another for 20-30 minutes.
- **Injury** – including bruising, concussion, fractures or even drowning if the seizure occurs during a bath.
- **Trouble breathing** – individual's lips may turn blue.

What to Do:

1. Clear the area around the individual, stay with him/her to prevent injury. DO NOT put anything in his/her mouth.
2. Try to write down what happened before, during and after the seizure and how long the seizure lasts.
3. Notify agency nurse/supervisor as soon as possible.

4. Call 911 if the seizure lasts longer than 5 minutes, if individual is injured or if he/she stops breathing.
5. Begin rescue breathing if you are certified to do so.

The psychiatric condition most likely to lead to an emergency is

Depression and Suicide Attempt

What to Do:

1. Insure the individual's safety if possible.
2. Call 911.
3. Notify agency and individual's physician (psychiatrist and/or PCP) as soon as possible.

Acute Glaucoma

What to Do:

1. Notify the individual's opthalmologist immediately.

Conclusion

Sturge Weber Syndrome is a neurocutaneous condition characterized by facial port wine stain, seizures and other neurologic complications associated with angiomata of the leptomeninges, and glaucoma. It usually occurs sporadically although it occasionally is found in families. Males and females are equally affected. It has been reported in individuals of all races and in every socioeconomic group. Seizures may be very difficult to control. 60-70% of individuals with Sturge Weber Syndrome have associated mental retardation or developmental delays. Other neurologic conditions include hemiparesis, hemiatrophy and headaches. Emotional problems, especially depression, and behavioral problems are common. No method of prevention is available. Treatment is aimed at seizure control, prevention and treatment of glaucoma, recognition and management of emotional and behavior problems with medication, psychotherapy and behavioral therapies, symptomatic treatment of headaches, and cosmetic attention to port wine stain. Physical and occupational therapy may also be beneficial.

References

Sujansky and Conradi, "Outcome of Sturge-Weber Syndrome in 52 Adults" American Journal of Medical Genetics 57:35-45 (1995.)