PORT WINE BIRTHMARK PRESENTATION

Port wine birthmarks (capillary vascular malformations) of the face are the hallmark of the Sturge-Weber syndrome.

WOUNDS IN PORT WINE BIRTHMARKS

- Port wine birthmarks are comprised of a massive excess of capillary blood vessels. Therefore, wounds in port wine birthmarks tend to bleed profusely.
- Compression bandages are important before and after suturing wounds in port wine birthmarks.
- Local anesthesia is the same as for any wound.
- Extensive electrocoagulation of bleeding dermal blood vessels should not be done in order to avoid cutaneous necrosis.
- Deeper wounds require dermal suturing to obtain perfect dermal alignment. Resorbable 4.0 sutures should be used.
- Cutaneous sutures should be non-resorbable, 4.0 or 5.0 and removed in 7-10 days.
- Patients should wear a compressive bandage for 24 hours following suturing.
- Patients should be seen in follow up the next day.

PYOGENIC GRANULOMAS IN PORT WINE BIRTHMARKS

- Pyogenic granulomas are neovascular growths that are seen in normal skin, but also occur commonly in port wine birthmarks.
- They have a very friable surface and bleed frequently.
- Local anesthesia is the same as for any skin injury.
- The simplest and best single treatment for pyogenic granulomas is cauterization or laser photocoagulation of the bleeding papule.
- Bleeding from the subsequent wound is best controlled by pressure and gentle electrocoagulation.
- Recurrence of the lesion can occur requiring re-treatment.
- Excisional surgery should not be performed because it is more difficult, will lead to a larger scar and may also induce the development of satellite pyogenic granulomata.
- For lesions less than 5 mm, pulsed dye laser treatment if available, is simple and may be effective.

OTHER DERMATOLOGIC CONDITIONS OF THE SKIN INVOLVED WITH A PORT WINE BIRTHMARK

- Common skin conditions such as eczema, acne, impetigo and the majority of skin diseases may be treated the same as in patients without port wine birthmarks.

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**The information contained herein is specific to the United States. We recognize that emergency procedures in other countries may differ.* 2013

STURGE-WEBER SYNDROME EMERGENCY ROOM GUIDE

NEUROLOGIC PRESENTATIONS

The major neurological features of patients with Sturge-Weber syndrome include seizures, headache, focal neurologic deficits and cognitive and psychological impairment.

SEIZURES

- Seizures develop in 70 to 80% of patients with Sturge-Weber syndrome.
- The risk of developing seizures is highest in the first two years.
- 75% of those with seizures have the initial event during the first few months of life often in conjunction with hemiparesis or other focal deficits.
- The natural course of Sturge-Weber syndrome depends on the presence, persistence and resistance to treatment of seizures. Onset of seizures before 1 year of age, and the occurrence of episodes of status epilepticus predict an unfavorable outcome.
- Seizures are usually focal motor or secondarily generalized tonic clonic seizures, but infantile spasms, myoclonic seizures and atonic seizures may also occur.
- Many patients have frequent and repeated seizures, but some children have only occasional seizures. In some patients, seizures may cluster.
- EEG studies document decreased amplitude and frequency of electrocerebral activity over the affected hemisphere. Diffuse, multiple and independent spike foci are commonly present.
- CT scan demonstrates calcification, atrophy of the brain, and enlargement of choroids plexus on the side of the pial angioma. MRI of brain with gadolinium enhancement is the preferred imaging modality to evaluating the extent and severity of intracranial involvement. Brain MRI may be occasionally negative during the first few months (or even years) of life despite showing abnormalities subsequently.
- When a child presents with a new-onset seizure, a neurologic consultation should be sought. Routine studies including blood glucose, electrolytes, serum calcium and magnesium and CBC should be obtained to evaluate for precipitating factors. EEG and neuroimaging studies such as CT or MRI scan of brain should also be obtained. Aggressive antiepileptic regimen should be established from the first seizure.
- Arrest of status epilepticus is of utmost importance. Immediate management includes assessment of vital functions, establishment of intravenous lines, blood sampling for investigation of possible metabolic or infectious causes and IV benzodiazepines such as lorazepam. IV benzodiazepines may be repeated if necessary. If seizures are not controlled within minutes of giving benzodiazepines, IV fosphenytoin or phenobarbital should be given in adequate doses. For resistant status epilepticus, close EEG monitoring and supervision by specialized medical and nursing personnel in intensive care unit are required. General anesthesia with IV barbiturates or other agents, neuromuscular blockade and respiratory support may be needed.
- Approximately half of the patients achieve complete control with standard antiepileptic agents and a significant proportion of the remaining patients achieves partial control.
- In medically-refractory cases, focal cortical resection or hemispherectomy can improve seizure control and may prevent cognitive decline.

HEADACHE

- About half of the patients complain of headaches. Migraine represents the most frequent type of headache.
- Headaches commonly occur after epileptic seizures.
- In a few patients headaches may be due to glaucoma.
- Headaches should be managed as in other persons. However due to increased risk of focal neurologic deficits, vasoconstrictor agents such as triptans and ergots should probably be avoided.
- When a patient presents with severe headache for the first time, neurologic consultation should be obtained to establish a diagnosis. Neuroimaging studies may be necessary to evaluate for intracerebral or subarachnoid hemorrhage. Cerebrospinal fluid analysis may exclude subarachnoid hemorrhage or central nervous system infection.
- Acute presentation of recurring severe headaches may be treated with oral or parenteral analgesics and antiemetics (e.g. IM ketorolac/IV prochlorperazine). Neuroimaging studies are indicated for recurring headaches that do not fulfill the strict definition of migraine or other primary headache disorder. For recurring migraine-type headaches, preventive therapy may be needed.

FOCAL NEUROLOGIC DEFICITS

- Hemiplegia occurs in at least one-third of the cases and is localized to the side opposite to the facial port-wine birthmark.
- Hemiplegia commonly first appears after an episode of seizure and may become more severe with the recurrence of seizures.
- Hemianopia is present in majority of patients, either alone or in association with hemiparesis.
- Transient hemiplegias not following an epileptic attack and sometimes accompanied by migraine-like headaches are observed in many cases of Sturge-Weber syndrome. These hemiplegic episodes are apparently not of epileptic nature and may be a consequence of vasomotor disturbances within or around the angioma.
- Intracranial hemorrhage is quite rare but has been documented in a few cases.
- When a child presents with acute focal neurologic deficit, determination of the cause is essential in guiding therapy. Neuroimaging studies such as CT or MRI scan of the head should be performed and a neurologic consultation should be obtained.
- Low dose aspirin has been used by some physicians theoretically to prevent recurrent venous thromboses that are supposed to cause stroke-like episodes and focal deficits. However, lack of controlled clinical trials and the clinical variability of the syndrome make it impossible to determine whether aspirin is helpful.

OPHTHALMIC PRESENTATIONS

Ophthalmic abnormalities are part of the Sturge Weber syndrome (SWS). The information below is designed to offer Emergency Room physicians insight into the common eye findings unique to SWS and the special ocular problems that patients with SWS may experience.

COMMON “typical” SWS EYE FINDINGS

- Port wine birthmark involving the eyelids.
- Differentiate from erythema by lack of tenderness, presence of sharply demarcated edges often respecting the midline, and lack of warmth to touch.
- Red eye (prominence of conjunctival and episcleral vessels).
- Differentiate from ocular inflammation by lack of discharge, lack of pain, clearly visible tiny vessel complexes, tortuous and teleangiectatic in appearance, and absence of symptoms.
- Retina and red reflex appearing redder than usual or than in the uninvolved contralateral eye.
- Due to choroidal hemangioma (choroid lies underneath retina).
- Differentiate from retinal hemorrhage by absence of distinct blood spots and appearance of diffuse redness. Particularly obvious in unilateral SWS when compared to unaffected eye.

COMMON SWS EYE PROBLEMS

- Lid swelling.
- Most often occurs on waking.
- Differentiate from cellulitis by history of long recumbent period, recurrence, absence of fever, absence of tenderness, lack of true erythema (see above), lack of warmth to touch, and resolution within hours with no treatment.
- Glaucoma.
- May present, especially in children < 5 years old, with ipsilateral enlarged eye, photophobia, tearing, cloudy cornea, decreased red reflex, and enlarged optic nerve cup.
- All signs and symptoms not required for diagnosis.
- Requires urgent referral to ophthalmologist if any suspicious signs or symptoms.
- Glaucoma in older children usually presents with no symptoms or signs except enlarged optic nerve cup but may have some of the findings above.
- Intraocular pressure significantly higher than in the contralateral uninvolved eye.
- Be sure to check vision in both eyes, with glasses on and/or with pinhole to get best corrected visual acuity measurement.
- Visual field defects.
- Especially in children with seizures and those who have had hemispherectomy, most common is one sided hemianopsia.
- Decreased peripheral vision common either due to brain involvement or glaucoma.
- Retinal detachment and other complications from choroidal hemangioma are very rare.
- Refer to ophthalmologist if new discovery of decreased vision in one eye and absent or white (leukocoria) pupillary “red reflex test”.

SPECIAL CONSIDERATIONS

- All newborn babies with SWS and a port wine mark affecting the eyelids and/or brain involvement should see an ophthalmologist in the first few weeks of life. Children with port wine birthmark involving lids may be at life long risk of glaucoma and require periodic eye examination, often under anesthesia or sedation in the first years of life until able to be done awake.
- Any child who has had prior eye surgery and presents to the Emergency Department with a red eye (above baseline), pain, or any other ocular concern (including conjunctivitis) should be urgently referred to their ophthalmologist.

Children with glaucoma are often on multiple medications. In general:

Red top drops = pupil dilators (sympathomimetics or parasympatholytics).
Green top drops = pupil constrictors (parasympathomimetics).
Purple top drops = alpha agonists.
Orange top drops = carbonic anhydrase inhibitors.
Others: topical prostaglandin analogues, antibiotics, steroids and other anti-inflammatory drugs may be in use. May also use be using oral steroids or oral carbonic anhydrase inhibitors for eye problems.

- Some bottles may contain more than one agent (combination preparation).
- Children with SWS on certain alpha agonists (e.g. brimonidine) may be more prone to sedative side effects that can be acute and dramatic. In infants or children with severe SWS, bradycardia, apnea, and hypothermia may also be seen even if only one drop given.
- If a child has had a glaucoma drainage tube implant (also called seton or tube), there may be a visible whitish square at the edge of the cornea in one quadrant. This is tissue used to cover the plastic tube.
- If a child has had glaucoma surgery, he or she may have a cystic appearing mass (bleb), either vascular or avascular, in one or more quadrants beyond the edge of the cornea. This is the expected area of fluid drainage.