PORT WINE STAINS AND STURGE-WEBER SYNDROME

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It is important for general practitioners to recognize cutaneous port-wine stains as these could signify important association with Sturge Weber syndrome, where neurological and ocular complications are likely. As importantly, advances in technology in the past two decades have made laser the standard of care for the management of port-wine stains. Port-wine stains are venous ectasias that are present at birth, but changes colour from pale to pink in the newborn to a deep red or dark purple colour (as in port wine) later. As port-wine stains can be progressive, laser treatments should be done early to prevent thickening and nodularity of the skin.

Contents

- What are Port Wine Stains?
- Options of Treatment for Port Wine Stains
- What is Sturge-Weber Syndrome (Encephalofacial Angiomatosis)?
- Clinical Features of Sturge-Weber Syndrome
- Neurological Complications of Sturge-Weber Syndrome
- Diagnosis
- Classification of Sturge-Weber Syndrome
- Management
- Prognostic Factors
What are Port Wine Stains?

Cutaneous port wine stains (PWS) are present from birth and the colour changes with age as the infant grows. In the newborn, the PWS are often flat and pale pink in colour, but darkens with age to a deep red or dark purple colour (as in port-wine) as well as thicken and become more nodular. They were in the past erroneously referred to as capillary hemangiomas, but are in fact venous ectasias. They appear sporadically in 3 per thousand births with no sex predilection. Although often depicted to involve the first division of the trigeminal nerve on the forehead, it could also extend and be present in other parts of the body e.g. trunk and the limbs.

Options of Treatment for Port Wine Stains

Facial PWS are unsightly resulting in psychological problems such as low self-esteem, lack of self-confidence, and difficulties in social interaction. Thus, options for treatment need be considered despite the possibility of incomplete response or resolution. For older children and adults with PWS, one of the obvious methods is covering up using camouflage make-up. Some people may achieve the desired effects through experimenting with over-the-counter cosmetics.

Advances in technology in the past two decades have made laser the standard of care for the management of PWS despite unanswered questions on the treatment results and long-term benefits. As PWS can be progressive, laser treatments should be done early to prevent thickening and nodularity of the skin. Majority of PWS lighten significantly with treatment using pulsed dye lasers capable of selective photo-thermolysis that destroys the ectatic vessels. Although EMLA had been tried, general anaesthesia is preferred for pain control in paediatric patients. Only about 15 to 20% of PWS clear completely and they could still recur after treatment. Thus, the individual may need to have maintenance laser treatments for life.

In the paediatric population, the best results of laser treatments were in the youngest children (less than one year old) and the greatest decrease in size of the port-wine stains were in the smaller lesions (< 20 cm²) and those located over the bony areas of the face such as the central forehead as opposed to on the face peripherally.
What is Sturge Weber Syndrome (Encephalofacial Angiomatosis)?

Sturge-Weber syndrome (SWS) is a non-familial neurocutaneous syndrome characterized by facial PWS and occipital leptomeningeal angiomatosis with resulting neurological abnormalities. Other features that are often associated with SWS include eye manifestations and rarely internal organ involvement. Nevertheless, each case of SWS is unique and exhibits the characteristic features to varying degree. Though rare, there had also been reports of SWS without any facial naevus. Extending this concept, the clinical spectrum of SWS could also include children with encephalofacial angiomatosis sparing the occipital lobe and without the facial naevus as being sufficient to fulfil the criteria for SWS.

Clinical Features of Sturge-Weber Syndrome

1. Cutaneous Manifestation
   Although the presence of port-wine stain in a newborn brings on the strong suspicion of SWS, on the whole only about 8% of these will go on to develop SWS. If the PWS involves both the first and second trigeminal nerve distribution (i.e. both the upper and lower eyelids are involved), the risk of leptomeningeal angioma (and thus SWS) increases to 90%.

2. Leptomeningeal Angioma
   The leptomeningeal angiomatosis is most often ipsilateral to the facial nevus. The occipital and parietal lobes are usually involved, though other areas more anteriorly could also be affected. In SWS, histopathologic studies reveal large tortuous venous structures in the thickened and discoloured leptomeninges. The underlying cerebral cortex is often atrophied, with calcified deposits.

3. Ocular Features
   Other features that are often associated with SWS include eye manifestations such as glaucoma (30% to 70%) and buphthalmos (up to 50% of newborns with SWS). The risk of glaucoma is highest in the first decade and most of these patients (92%) too have the PWS affecting the dermatomes of both the first and second division of the trigeminal nerve. The majority of patients with unilateral facial PWS had ipsilateral glaucoma. With bilateral facial PWS, 45% had bilateral glaucoma. The other ocular findings that may be present in SWS include choroidal angioma, episcleral/conjunctival angiomas. Less often, there could be heterochromia of the iris, optic atrophy and dilated retinal veins.
Neurologic Complications of Sturge-Weber Syndrome

Besides the clinical features above, other important clinical manifestations of SWS include neurological complications such as seizure, focal neurological deficit and mental retardation.

1. **Epilepsy**
   About 75% of patients of SWS develop seizures, and this is more common in the presence of bilateral PWS. Seizures generally start either in infancy of early childhood, with 95% presenting before 5 years of age. The seizures are primarily partial motor seizures that often become secondarily generalized tonic-clonic convulsions. Almost half the patients go on to have convulsive status epilepticus, occurring as prolonged clonic seizures. Less commonly, infantile spasms and myoclonic seizures could also occur.

2. **Hemiparesis**
   Up to half of the patients with SWS are hemiparetic, with a third also showing hemiatrophy that are usually contralateral to the facial and intracranial lesions. Hemiparesis often develops acutely with the onset of seizures. The postictal paralysis (Todds paralysis) requires progressively longer periods to recover till permanent weakness results. On the other hand, some children develop step-wise deterioration with progressive hemiparesis that is thought to follow a series of stroke-like episodes.

3. **Visual field defects**
   Visual field defects are common when the occipital lobe or optic tracts are affected. Homonymous hemianopsia is difficult to be determined in young children, but is estimated to occur in about a quarter of patients with SWS.

4. **Mental disability**
   Most children with SWS develop normally for a few months, and then exhibit developmental delay. Intellectual disability is worse in patients with bilateral cerebral involvement. Ultimately mental handicap is present in about 60% of patients, with severe mental disability in a third. Not surprisingly, behaviours associated with attention deficit hyperactivity disorder, oppositional defiant disorders and conduct problems were much higher in patients with SWS. The behavioural and psychological problems were associated with poorer cognitive function and frequent seizures.
Diagnosis

The diagnosis of SWS is most commonly made when an infant or child with PWS develop neurological complications, and the magnetic resonance imaging (MRI) of the brain with contrast studies demonstrate the presence of leptomeningeal angiomatosis and accompanying cortical abnormalities. If MRI is unavailable, computed tomography scan of the brain may demonstrate brain calcification that typically assume a linear, parallel configuration (“tram” sign) or sometimes a convolutional pattern in the parieto-occipital regions. During infancy, leptomeningeal angiomatosis may not be reliably detected by neuroimaging and consequently a negative scan is non-informative. Thus, for infants presenting with only PWS, intracranial involvement cannot be excluded with certainty in the first year of life.

Classification of Sturge-Weber Syndrome

Presently there are clear differences for the minimum diagnostic criteria required for SWS. Thus, the diagnosis of SWS could be made based on the presence of the common triad of cutaneous facial angioma, leptomeningeal angiomatosis and ocular involvement, or just based on the presence of intracranial leptomeningeal vascular malformation. To address some of the differences in the definition of SWS, ES Roach had classified the encephalofacial angiomatosis into three types in 1992 based on the varying degrees of involvement as previous reported.

**Type I (Classic) Sturge-Weber syndrome**
This is the most commonly described form, with both facial and leptomeningeal angiomas. Seizures usually occur in the first year of life, and ocular involvement, most commonly glaucoma, is likely to be present.

**Type II Sturge-Weber Syndrome**
This type manifests with facial angioma and the possibility of glaucoma, but with no evidence of intracranial disease.

**Type III Sturge-Weber syndrome (forme fruste)**
This type only involves leptomeningeal angioma, with no facial nevus and usually no ocular manifestation of glaucoma.

In both types II and III SWS, the natural course of the disease and progression over time is not known for certain and additional research is necessary.
Management

Presently there is yet any specific treatment for SWS, and the management of the clinical manifestations and complications is still far from adequate.

Even for children without glaucoma, regular ophthalmologic surveillance is necessary till adulthood as some may develop raised intra-ocular pressure much later. In rare instances, the glaucoma could develop as late as the 4th decade. If glaucoma is present, topical treatments alone using beta-blockers and carbonic anhydrase inhibitors are often ineffective, and combination with surgical treatment, such as cryo-coagulation of the ciliary body, may be more effective.

Epilepsy is the most common and often the first neurological complication of SWS. Unfortunately, the success of seizure-control with anti-epileptic drugs (AEDs) is variable and unpredictable. About two-thirds of patients have seizures that are resistant to treatment with the conventional AEDs. Only in one-third of patients is the epilepsy easy to control. Nevertheless, less than 10% of patients with SWS could be successfully weaned off medication after being seizure-free. For children with drug-resistant seizures, epilepsy surgery need be considered early rather than late when cognitive impairment sets in.

Prognostic Factors

The prognosis for SWS obviously depends on the extent of the leptomeningeal angiomatosis. Besides this, the most important prognostic factor is the absence or presence of seizures, and if present the age of onset of the seizures. Of children who do not have seizures in the first 2 years of life, only 14% developed epilepsy later on. Children with later onset of seizures showed lower risk for developmental delay and the need for special education. Overall, almost 60% of children with SWS showed early developmental delay and required special education. This risk increased to 83% if seizures started before one year of age. On the brighter side, only 6% of children without epilepsy had developmental delay and only 11% required special education.