# Port-wine Birthmarks: Update on Diagnosis, Risk Assessment for Sturge-Weber Syndrome, and Management

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## EDUCATION GAP

Large forehead or midline port-wine birthmarks (PWBs) carry the highest risk of Sturge-Weber syndrome. The gold standard treatment for PWBs is the pulsed dye laser (PDL), which is the most effective laser for PWBs and has the longest safety history in children. PDL is most effective when started early in childhood, ideally within the first year after birth.

## LEARNING OBJECTIVES *After completing this article, readers should be able to:*

1. Identify facial port-wine birthmarks (PWBs) at highest risk for Sturge-Weber syndrome.
2. Differentiate a facial PWB from other vascular birthmarks.
3. Based on initial evaluation, determine appropriateness of referrals to ophthalmology, neurology, and/or dermatology.
4. Describe the natural progression of the PWB and the risks, benefits, and optimal timing of pulsed dye laser as a treatment option.
5. Provide necessary psychosocial support and resources to children with Sturge-Weber syndrome and their families.

## ABSTRACT

Sturge-Weber syndrome (SWS) is a neurocutaneous disorder that classically presents with a triad of vascular anomalies affecting the skin, eyes, and brain. Previously, the trigeminal nerve distribution of a port-wine birthmark (PWB) of the face was used to identify risk of SWS. However, recent evidence has demonstrated that PWBs are vascular, not neurologic, in embryologic

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**ABBREVIATIONS**

| | |
|---|---|
| AVM | arteriovenous malformation |
| IH | infantile hemangioma |
| MRI | magnetic resonance imaging |
| NS | nevus simplex |
| PDL | pulsed dye laser |
| PWB | port-wine birthmark |
| SWS | Sturge-Weber syndrome |

origin, and facial PWBs at highest risk for the brain involvement of SWS involve the forehead location. Furthermore, a PWB involving the upper or lower eyelid carries a risk of glaucoma, which requires lifelong monitoring. The gold standard of treatment for PWB is the pulsed dye laser, which has many advantages when started as early as possible in infancy. In this review, we discuss the locations of facial PWBs at risk for neurologic and ophthalmologic complications, the differential diagnosis of facial vascular birthmarks, recommendations for patient referral(s) when needed, and the advantages of early laser therapy when desired for the PWB. We also provide additional resources for pediatricians to support patients and their families.

## BACKGROUND

Sturge-Weber syndrome (SWS) is a neurocutaneous disorder that classically presents with at least 2 of a triad of vascular anomalies affecting the skin, eyes, and brain. Often, the first clue to SWS in a newborn is the presence of a facial port-wine birthmark (PWB), previously referred to as a port-wine "stain," with "birthmark" now the preferred term due to the more positive connotation, or the medical term "capillary malformation." A PWB is a red, well-demarcated patch composed of capillaries that may affect the skin anywhere on the body but when associated with SWS involves the face. (1) A somatic activating mutation in *GNAQ* (G protein subunit alpha q) is the most common cause of SWS and PWBs. (2)(3)(4) *GNAQ* affect the RAS (rat sarcoma) pathway, which plays an important role in vascular cell growth and angiogenesis. *GNAQ* inhibits cellular apoptosis and increases cell proliferation via the RAS pathway, leading to uncontrolled vascular growth in affected tissues. (5)

## DIAGNOSIS

For a diagnosis of SWS, children typically have at least 2 of the following 3 criteria: a characteristic facial PWB, vascular malformations of the eye, and brain magnetic resonance imaging (MRI) evidence of vascular malformations specific to SWS, termed leptomeningeal angiomatosis. (6) Although children with SWS have a facial PWB, there are rare reports of SWS in the absence of a PWB. (7)(8)(9)

## RISK OF SWS

Recent literature has demonstrated that the location and extent of a facial PWB are key to predicting risk of SWS. (5)(6) Of note, the trigeminal nerve distributions V1, V2, and V3 are no longer used to describe the location of PWB on the face because it is now known that PWBs are not related to the trigeminal nerve in any capacity but are instead vascular in origin, with a cutaneous distribution corresponding to embryologic vasculature development. (5) The best independent predictor of SWS is a PWB located on the forehead. (5) The forehead location is defined by an inferior border drawn from the outer canthus of the eye to the top of the ear, including the eyelid. Furthermore, forehead PWBs at risk for SWS must be either lateral on the forehead and large (involving greater than one-half of the hemi-forehead) (6) or, more rarely, located over the midline forehead and of any size (Fig 1A). (10) Furthermore, the more the PWB extends beyond just the forehead, the even greater the risk of SWS. (11) For example, a child with a PWB affecting the entire hemiface or bilateral face is at higher risk than a child with a PWB affecting only the forehead. (12)(13) Notably, the cutaneous forehead shares the same vasculature origin as the cerebral cortex and eye, explaining the classic SWS triad. (5)

## EVALUATION OF AN INFANT AT RISK FOR SWS (FIG 2)

A pediatric dermatologist or other vascular birthmark specialist can help confirm the diagnosis of PWB, determine risk for SWS, and discuss the option of pulsed dye laser (PDL) therapy. A newborn diagnosed as having a facial PWB at risk for SWS should also be evaluated by an ophthalmologist and neurologist experienced in the management of SWS. Ophthalmologic evaluation is urgent in the newborn period because the glaucoma associated with SWS can be congenital. Furthermore, any individual with a PWB that involves the periorbital skin, including either the upper or lower eyelid, is at risk for acquired glaucoma and thus needs regularly scheduled eye examinations throughout his or her lifetime (Fig 1B), even if initial screens are negative. (14) Notably, an infant with an upper eyelid PWB that does not involve the forehead is at low risk for SWS. At-risk infants for SWS should also be referred to neurology for assessment and potentially for brain MRI with and without contrast. Although controversial and variable among pediatric neurologists and between institutions, some advocate for earlier imaging even in asymptomatic patients, (5) especially in infants with extensive facial PWBs at highest risk for SWS, given the potential for prophylactic treatment with antiepileptics (15) and aspirin. (16)(17) However, there is not a consensus on timing of

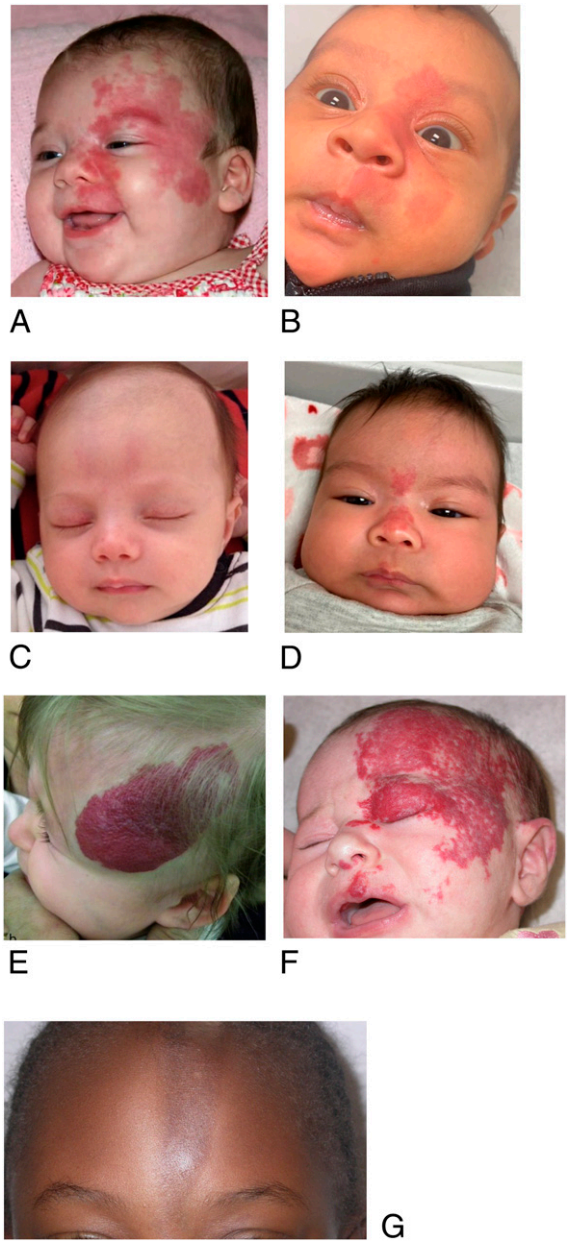


**Figure 1.** Differential diagnosis for port-wine birthmark (PWB) at risk for Sturge-Weber syndrome (SWS). A and B, Examples of PWBs. Note the solid, sharply demarcated erythematous patch. A, Hemi-forehead PWB defined as large (involving greater than one-half of the hemi-forehead and therefore high risk for SWS). B, Unilateral PWB defined as small (involving less than one-half of the hemi-forehead and therefore low risk for SWS). Note, however, that this infant is still at risk for acquired glaucoma because the PWB involves the periorbital skin. Examples of nevus simplex (NS) on the midline forehead with characteristic involvement of the upper eyelids (C) and on the glabella, nasal dorsum, nasal tip, and lip (D). Compared with PWBs, NS is lighter in erythema, is patchy, and has more ill-defined borders. A newborn with glabellar NS will often have NS evident in other common locations, such as the upper eyelids, perinasal skin, philtrum, and occiput. E and F, Examples of segmental facial hemangiomas at risk for PHACE; will be raised and palpable, segmental, and (when at risk for PHACE syndrome) greater than 5 cm in any distribution in any location over the face or scalp. G, Morphea consists of a more well-demarcated, erythematous or violaceous patch compared with PWBs. Over time, morphea develops an atrophic, bound-down, scar-like appearance. Morphea is acquired and not present at birth and can be associated with alopecia, when the condition extends into a hair-bearing area.

imaging because normal MRI findings early in infancy do not preclude the possibility of neurologic involvement because results can be falsely negative when performed early in life. (5)(18) Anesthesia risks must also be considered with early imaging. However, in some experienced centers, MRI can be performed in young infants without general anesthesia, instead using a "feed and wrap" technique with the child fed and swaddled to induce sleep. (19)

## MANAGEMENT OF SWS

SWS is a spectrum of disease, with some children having more severe sequelae and others having milder symptoms. Management of SWS focuses on treating the dermatologic, neurologic, and ophthalmologic manifestations of the disease. Children with SWS and brain involvement may experience significant neurologic morbidities, including headaches, epilepsy, strokelike episodes, developmental delays, and encephalopathy. (20) Psychiatric comorbidities, most often anxiety and depression, are also common. (21)(22) Neurologists thus perform an essential role for at-risk infants by providing seizure education to caregivers, interpreting imaging results, and managing epilepsy and other neurologic complications. In addition, children with SWS should also have regular eye examinations to monitor for glaucoma. If intraocular pressures are found to be high, topical agents are considered first line to reduce aqueous humor by decreasing production, increasing outflow, or a combination of the 2 methods. (21) However, given the anatomical abnormalities of the eye in children with SWS, topical agents may prove ineffective, in which case, surgical intervention may be necessary. (21)(23)(24)

## DIFFERENTIAL DIAGNOSIS OF PWB (TABLE 1)

A PWB, especially when located on the midline forehead, must be differentiated from nevus simplex (NS), commonly known as angel's kiss. NS that involves the midline forehead is markedly more common than a PWB in the same location that would be at risk for SWS, which is rare. In contrast to PWB, NS tends to be a lighter red color and poorly demarcated with ill-defined borders (Fig 1 C and D). In addition, a newborn with glabellar NS will often have NS evident in other common locations, such as the upper eyelids, perinasal skin, philtrum, and occiput. Also in contrast to PWBs, which persist throughout life, NS is most often transient in nature, typically fading over the first year, although NS localized to the glabella or occiput can sometimes persist. (5) In cases of uncertainty, referral to a vascular birthmark specialist is advised for definitive diagnosis before implementing an evaluation for SWS.

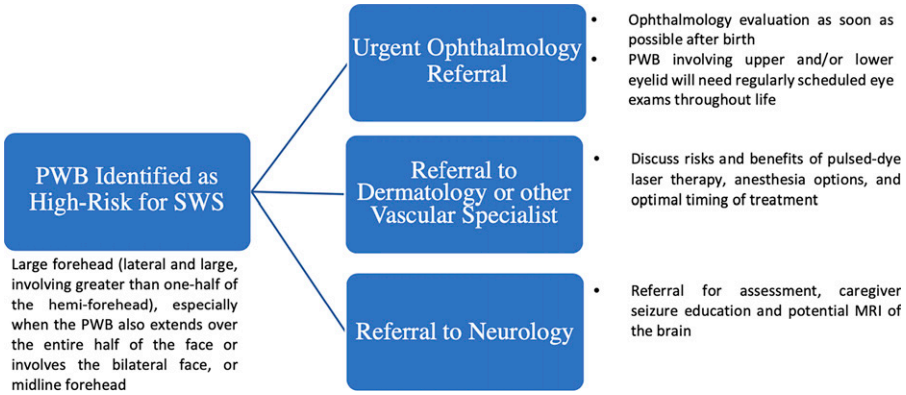


**Figure 2.** Flowchart for identification and management of a port-wine birthmark (PWB) at risk for Sturge-Weber syndrome (SWS).

The other important differential diagnosis for PWB in a newborn is infantile hemangioma (IH), particularly when the IH is large, plaquelike and patterned, also known as "segmental." Segmental IHs can present very similarly to PWBs at birth but are often lighter in erythema and more telangiectatic, referring to the presence of scattered, dilated superficial blood vessels, as opposed to a solid and well-defined erythematous patch (Fig 1E). In addition, IHs will show evidence of proliferation within the first 2 to 4 weeks after birth, with the development of raised, textural changes, whereas PWBs will not. Also note that segmental IHs will sometimes show only subtle proliferation, also known as IH with minimal or arrested growth (Fig 1F). Last, pediatricians should be aware that segmental IH of the face and neck can be associated with a similar but distinct vascular syndrome from SWS known as PHACE (*p*osterior fossa anomalies, large, segmental *h*emangiomas of the head and neck, cerebrovascular *a*rterial anomalies, *c*ardiovascular anomalies, and *e*ye anomalies). Neurovascular and cardiovascular imaging and an ophthalmologic examination are necessary to establish the diagnosis of PHACE syndrome, for which diagnostic criteria have been published. (25)

Although rare, 2 other entities that can be initially misdiagnosed as PWB include arteriovenous malformation (AVM) and morphea (Fig 1G). AVM can be congenital or acquired and may affect the forehead. However, over time an AVM will develop characteristic features, including increased warmth, pulsatility, swelling, and/or draining veins, that will distinguish this as a high-flow vascular birthmark distinct from PWB. Screening for an AVM can be performed with Doppler ultrasonography. Morphea is an inflammatory skin condition that may present on the forehead similar to PWBs in a linear pattern but is typically an acquired disorder compared with PWBs, which are present at birth. Over time, morphea evolves into a scarlike appearance with bound-down, firm skin, atrophy, dyspigmentation, and hair loss when the condition extends into the scalp or brow. The diagnosis of morphea is usually made clinically but can be confirmed by pathology.

## NATURAL HISTORY AND TREATMENT OF PWBS

Untreated, the natural history of PWB involves a deepening of the color from red to violaceous, skin thickening, and the potential development of pyogenic granuloma–like lesions prone to bleeding (Fig 3). (26)(27) Such changes are most common when the PWB affects the face, although these changes may occur on other parts of the body as well. These changes typically do not occur until puberty or adulthood. (28) Soft tissue hypertrophy, especially when the PWB affects the middle and lower half of the face, can also lead to overgrowth of gingival tissue and the underlying mandible with resultant dental and orthodontic complications, likely due to the underlying *GNAQ* mutation. (26)(29) PDL therapy can be used to improve the appearance of the PWB and, particularly when initiated early in childhood before significant hypertrophy of the birthmark occurs, can prevent at least some of the progressive changes observed in the skin. (30)(31)

## LASER TIMING AND EFFICACY

The gold standard treatment for PWB is PDL, (30)(32)(33) which targets hemoglobin species contained in the cutaneous blood vessels. (34) Although several lasers are used for vascular lesions of the skin, PDL is the most efficacious and has the longest history of safety, particularly in children. (27)(30)(31) Although generally well-tolerated, risks of PDL include ocular damage if appropriate eye protection is not used, skin dyspigmentation that is generally temporary, blistering, and rarely, scarring. (33)(35)(36) Skin discomfort after the procedure is usually minor, and prescription analgesics are rarely required. Postoperative

**Table 1.** Differential Diagnosis for Facial PWBs at Risk for SWS

| | PWBs AT HIGH RISK FOR SWS | MIDLINE FOREHEAD NS | SEGMENTAL FACIAL IH AT RISK FOR PHACE |
|---|---|---|---|
| Onset | Present at birth | Present at birth | May be minimally present or absent at birth |
| Skin appearance | Solid, sharply demarcated erythematous patch | Lighter in erythema, ill-defined borders | May initially be a telangiectatic or solid segmental plaque, but will become raised and palpable |
| Distribution | Forehead: lateral + large (involving greater than one-half of the hemi-forehead) OR involve the midline forehead (any size)<br>Additional extent over the face beyond the forehead portends even higher risk of SWS | A newborn with a glabellar NS will often have NS evident in other common locations, such as the upper eyelids, perinasal skin, philtrum, and occiput | Segmental and >5 cm in any distribution over the face or scalp |
| Natural progression | Grow proportionally with child, do not involute<br>Do not proliferate in early childhood, but in adolescence or adulthood often progress with darkening of erythema, thickening, development of vascular blebs | Most are transient, fading over the first year, although glabellar or occipital NS may sometimes persist | Proliferation apparent within the first 2–4 wk after birth; on average, most proliferation occurs by age 6 mo<br>Sometimes shows only subtle proliferation, known as infantile hemangioma with minimal or abortive growth<br>Involution typically starts between the first and second year and continues until age 4–5 y |
| Brain | Leptomeningeal angiomatosis | NA | Cerebrovascular (most commonly of the internal carotid artery) and structural brain (most commonly of the posterior fossa) |
| Eyes | Potential congenital or acquired glaucoma or other ophthalmologic complications | NA | Several rare ocular anomalies may occur |
| Other | NA | NA | May also have cardiovascular anomalies, most commonly of the aortic arch, and midline ventral developmental defects of the skin ranging from a pit or scar to complete sternal agenesis |
| Evaluation | Referral to pediatric dermatology or other vascular birthmark specialist to confirm the diagnosis and determine risk of SWS<br>Ophthalmology examination at birth and regularly scheduled eye examinations thereafter if PWB involves the periorbital skin<br>Referral to neurology for brain MRI and epilepsy management, when indicated | No further evaluation indicated | Referral to pediatric dermatology or other vascular birthmark specialist to confirm the diagnosis of IH and determine risk for PHACE syndrome<br>MRI/MRA of the head and neck to evaluate for cerebrovascular anomalies, with neurology referral when indicated<br>Echocardiogram to evaluate for cardiovascular, particularly aortic arch, anomalies, with cardiology referral as indicated<br>Ophthalmology evaluation |

IH=infantile hemangioma, MRA=magnetic resonance angiography, MRI=magnetic resonance imaging, NA=not applicable, NS=nevus simplex, PWB=port-wine birthmark, SWS=Sturge Weber syndrome.

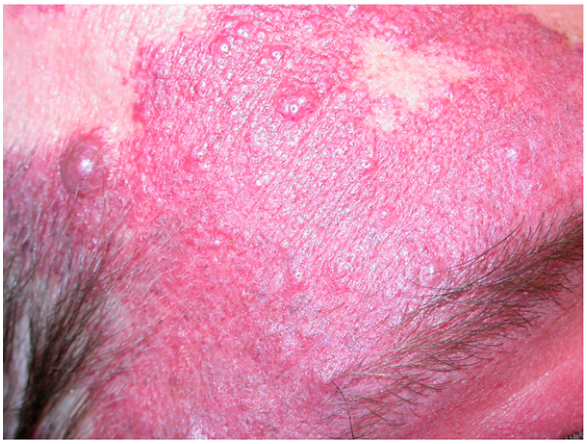

**Figure 3.** Progression of a port-wine birthmark. Adult showing increased violaceous discoloration, thickening, and the development of vascular blebs.

purpura occurs immediately after PDL, and this typically lasts 1 to 3 weeks (Fig 4). (37) Fortunately, this purpura, which looks like "bruising," is most often painless. Additional risks of laser are those related to anesthesia because the procedure itself is uncomfortable, often described as a repeated "rubber band snap" to the skin. Risks and benefits of anesthesia options should thus be thoroughly discussed with the family.

The decision to start PDL for a PWB is a commitment because a series of treatments is required and that number varies greatly, ranging from less than 5 to 30 or more. (38)(39) In general, smaller PWBs that involve peripheral areas of the face, such as the lateral forehead, respond better to laser and may require fewer treatments. (39) Conversely, PWBs involving the midface, particularly when more extensive, as is often the case with SWS, are typically more challenging to treat. (39)(40) PDL treatments are generally spaced 1 to 3 months apart to allow time for adequate healing. For facial lesions, the ideal time frame of PDL treatment is 3 to 6 weeks apart due to the recurrence of vessels. (41) Parents must also be appropriately educated to the fact that laser treatments can lighten the PWB but are very unlikely to permanently remove it. Thus, treatment is considered complete either when the patient and family reach a desired cosmetic end point or there seems to be no further lightening with subsequent sessions. However, even after a successful series of laser treatments with satisfactory lightening of the birthmark, a rebound in erythema often occurs later, such that future "maintenance" laser treatments are anticipated over a person's lifetime. (30)(42) The decision of when to stop the initial series and transition to maintenance varies between families but typically occurs by school age.

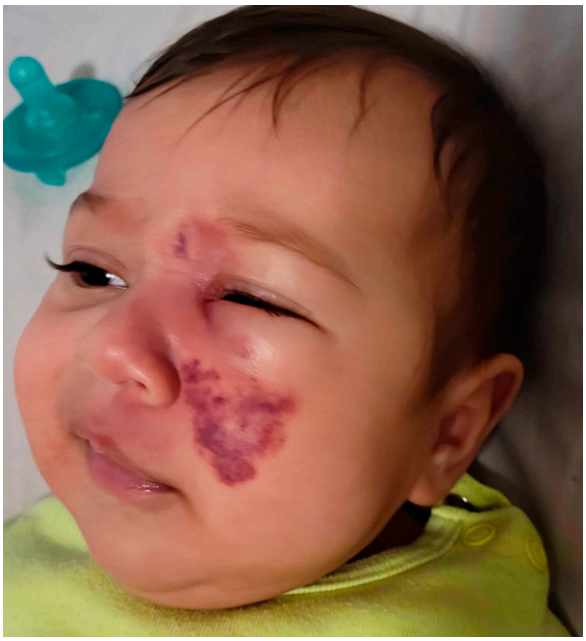

**Figure 4.** Typical purpura 1 week after pulsed dye laser. During healing, the purpura will be more noticeable than the original birthmark.

Per recent consensus recommendations, PDL is most effective for PWBs when started early in childhood, ideally within the first year after birth. (26)(30)(37)(43)(44)(45)(46) There are several advantages to these recommendations. The first is efficacy. The relatively thin nature of infant skin, the thin nature of the PWB itself, the smaller and more superficial nature of the blood vessels in the PWB, and the lesser amount of melanin in infant skin all allow for optimal laser penetration. (30)(39)(43)(45) Newborn infants also have increased fetal hemoglobin levels, in addition to increasing amounts of hemoglobin A, thus potentially providing an additional chromophore target. The second advantage of early laser is the potential avoidance of general anesthesia before toddlerhood because PDL can often be performed relatively quickly in an infant, especially when the PWB involves only a small surface area. (21)(46) This is particularly important given concerns raised by the US Food and Drug Administration (FDA) regarding potential adverse effects on neurodevelopment in children who have had repeated exposure to general anesthesia before age 3 years, although this risk has not been substantiated in recent publications addressing this issue in infants undergoing PDL for PWB. (47)(48)(49) Another advantage of completing the initial series of laser treatments early, optimally before a child starts kindergarten, is to optimize psychosocial development before stronger social interactions occur. Early laser treatments also avoid multiple school absences due to the procedure itself and the child having to return to school with postlaser

purpura. Strict sun protection is also of utmost importance before and after PDL, which is also more difficult to ensure in a school-age child. Sun protection is essential to avoid hyperpigmentation of the laser-treated skin during the healing stage. In addition, because the PDL targets melanin, albeit to a lesser degree than hemoglobin, tanned or pigmented skin may decrease the effectiveness of the laser by competing with the vascular target.

## LASER OF THE PWB IN CHILDREN OF COLOR

PWBs in children of color may also be treated with laser, but the risks of scarring and dyspigmentation are higher with PDL in children of color due to laser light absorption by melanin. (50)(51) This risk can be minimized with laser modifications in the hands of an experienced laser surgeon. As in all children, the risks and benefits of laser must be carefully weighed.

## HOPE FOR THE FUTURE

Permanent removal of a PWB with PDL can rarely be achieved for several reasons. Variations in vessel size and depth between patients (and even within the same PWB) make it challenging to target those vessels accurately with the laser without tissue sampling, clearly not ideal for a child with a facial birthmark. However, it is hopeful that advanced imaging technology, applied to the skin surface, will improve this capacity in the future. In addition, the PDL has a cutaneous depth of penetration of only 1.2 mm, thus often missing deeper vessels. Although alternative lasers with deeper penetration exist, they have a significantly higher risk of scarring and are thus not generally recommended in children; however, it is anticipated that advances in laser technology will continue to show improvements in both efficacy and safety. Last, given the use of targeted gene therapies in oncology and the recent development of such therapies for other vascular birthmarks, it is optimistic that medical therapies targeted to *GNAQ* will be available in the future for PWBs.

## PATIENT AND FAMILY SUPPORT

Providing necessary support resources for children with SWS as well as their families is a vital aspect of patient care. As with any chronic childhood medical condition,

**Table 2.** Family Support Resources

| | |
|---|---|
| Nonprofit organizations | • The Sturge-Weber Foundation (https://sturge-weber.org/): long-standing national organization dedicated to supporting research collaboration, patient education, and other advocacy efforts for SWS and can be a tremendous resource for families. Efforts are being made to provide medical guidance and other support to older teens with SWS transitioning to adulthood that will be available through the foundation.<br>• AboutFace International (https://rarediseases.org/organizations/aboutface-international/): organization that provides education and emotional support for individuals with facial differences and their families. |
| Educational resources | • The Society for Pediatric Dermatology (https://pedsderm.net/): provides patient handouts for many common skin conditions, including PWB and PDL therapy. These handouts are available to the public and can be useful first steps.<br>• Vascular Birthmarks Foundation (https://birthmark.org/): organization that provides information and support for patients and resources for providers. |
| Camps for kids | • Day or weekend camps specifically dedicated to children with facial differences are available<br>• Camp Discovery (https://www.aad.org/public/public-health/camp-discovery) and Camp Wonder (https://www.csdf.org/camp-wonder): camps supported by the American Academy of Dermatology with no cost to families.<br>• Such opportunities, which allow kids to "just be kids" while giving a child a chance to meet another with a facial PWB, perhaps for the first time, can be life changing. |
| Children's books | • *Sam's Birthmark:* written by a child with a PWB, which aims to teach children to embrace their own and each other's differences.<br>• *I am Unique!:* a story about self-love and sharing your unique talents.<br>• These books can be useful for parents to read at home and can also be read to the child's classroom. |

PDL=pulsed dye laser, PWB=port-wine birthmark; SWS=Sturge-Weber syndrome.

the stressors of having a child with SWS can adversely affect the family dynamics and be a significant source of financial and emotional strain. Parents of affected children often feel guilt that they somehow played a causative role in their child's PWB/SWS and may experience additional guilt over the decision to pursue (or not pursue) laser. Parents should be counseled about the genetic nature of the condition and that there is no action or lack of action that resulted in the PWB or SWS. Finally, as with any facial birthmark, the child must "wear" his or her birthmark, which can be an additional source of psychosocial and emotional distress for not only the child but family members as well. (52) Table 2 contains a list of organizations that provide information on SWS for patients as well as providers and resources for support for children with SWS and their families.

## Summary

- Based on consensus, for a diagnosis of Sturge-Weber syndrome (SWS), children typically have at least 2 of the following 3 criteria: a characteristic facial port-wine birthmark (PWB), vascular malformations of the eye, and brain magnetic resonance imaging evidence of vascular malformations specific to SWS, termed leptomeningeal angiomatosis. (6)
- Based on consensus, the differential diagnosis of a PWB includes nevus simplex, segmental infantile hemangioma (which may be associated with PHACE syndrome), arteriovenous malformation, and morphea.
- Based on recent research evidence, children with large PWBs involving greater than one-half of the hemi-forehead or involving the midline forehead of any size are at highest risk for SWS. (5)(6) That risk increases with PWB extent, such that children with a forehead PWB plus extension of the birthmark to the entire unilateral or bilateral face portend a risk that is even higher. (11)(12)(13)
- Based on consensus, children at risk for SWS should be referred to a pediatric dermatologist or other vascular birthmark specialist as soon as possible to definitively diagnose the PWB when necessary, determine risk of SWS, and discuss risks and benefits of pulsed dye laser (PDL) therapy.
- In addition, based on consensus, newborns at risk for SWS should be referred urgently to ophthalmology for evaluation for potential congenital glaucoma and to neurology for assessment and brain magnetic resonance imaging, caregiver seizure education, and epilepsy management when indicated.
- Based on research evidence, if PDL treatment is considered, there are multiple advantages to starting the series of laser treatments as early in infancy as possible.
- Based on strong evidence, PDL is safe to use in children and is the gold standard of treatment for PWBs. PDL is most effective at fading the PWB when initiated early in infancy, preferably within the first year of life. (26)(28)(31)(32)(33) Initiating treatment at this age also optimizes psychosocial development, minimizes school absenteeism, and minimizes the need for general anesthesia during early treatments.
- Based on consensus, providing necessary psycho social support and resources for children with SWS and their families is also key.
- Based on strong evidence, future medical therapies targeted to genetic vascular pathways, specifically *GNAQ* in the case of PWB/SWS, are hopeful.

### Acknowledgment

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*References and teaching slides for this article can be found at*
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# PIR QUIZ

1. A 1-week-old infant is brought to the clinic by her parents to establish care after being discharged from the newborn nursery. She was born at 39 weeks' gestation without complications during pregnancy or delivery. After delivery she was noted to have a red patch with a well-demarcated outline on her face. The family was informed that the baby has a port-wine birthmark (PWB). After searching the Internet, the parents are concerned about possible Sturge-Weber syndrome (SWS) and inquired whether their baby is at high risk. Of the following facial distributions, which one puts the baby at highest risk for SWS?

   A. Jaw line.
   B. Left eyelid.
   C. Left forehead.
   D. Right cheek.
   E. Vertex scalp.

2. A 2-week-old infant diagnosed as having a right eyelid PWB is being evaluated at her routine visit. After reviewing newborn care and ensuring she is feeding and growing well, you discuss the next clinical steps recommended for the baby regarding her birthmark. Which of the following recommended subspecialist referrals is the most urgent to complete at this time in this patient?

   A. Dermatology.
   B. Neurology.
   C. Neurosurgery.
   D. Ophthalmology.
   E. Otolaryngology.

3. A 4-week-old infant is brought to your clinic by his parents to establish care. Before you enter the examination room the nurse tells you that the baby is overall well-appearing with appropriate weight gain but that she noticed a birthmark on the baby's face. Which of the following clinical descriptions is most likely consistent with a PWB?

   A. Dark red, raised plaque.
   B. Dark red, well-circumscribed patch.
   C. Light red mark with ill-defined margins.
   D. Light red patch with red papules at the periphery.
   E. Brown, indurated plaque.

4. A 6-month-old is seen in the clinic for her health supervision visit. She has a PWB above the lateral aspect of her right eyebrow that extends to 1 cm under the hair line. You review with the family the recommended precautions and follow-up visits related to her birthmark, including possible treatment with pulsed dye laser. Which of the following represents the most optimal age at which to begin treatment with pulsed dye laser?

   A. Younger than 1 year of age.
   B. Between 2 and 5 years of age.
   C. Between 6 and 10 years of age.
   D. Between 11 and 15 years of age.
   E. Between 15 and 18 years of age.

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5. A 1-week-old infant is brought to the clinic by his parents to discuss the diagnosis of PWB. The mother is in tears and concerned that she caused the birthmark. You discuss her pregnancy and note that she was taking a selective serotonin reuptake inhibitor for depression and acetaminophen for headaches intermittently. She also consumed 1 to 2 caffeinated beverages daily, including soda and coffee. She denies any alcohol or illicit drug use. The infant was born at 38 weeks' gestation without any complications. There is no family history of any significant birthmarks or any neurologic abnormalities. The mother asks you what might have caused the birthmark. Which of the following is the most appropriate response about the potential cause of the birthmark in this patient?

   A. Acetaminophen exposure.
   B. Caffeine exposure.
   C. Environmental factors.
   D. Genetic abnormality.
   E. Selective serotonin reuptake inhibitor exposure.