Irregular Erythematous Patch on the Face of an Infant

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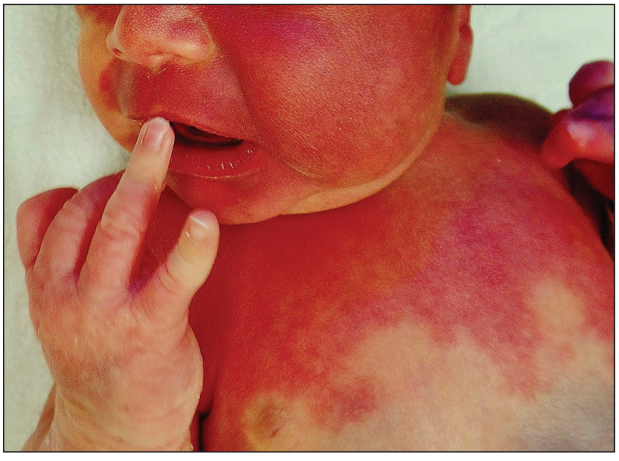

A newborn presented with an irregular and well-demarcated erythematous patch on the face, trunk, buttocks, and toes on the left foot. Another red patch was present on the right side of the face, while a slate gray patch covered the flanks and back. The limbs appeared symmetric and he exhibited no gross deformities. On close physical examination, he was noted to have a cloudy left eye. An ophthalmology consultation revealed a choroidal hemangioma and congenital glaucoma in the left eye.

WHAT'S THE **DIAGNOSIS?**

a. blunt trauma
b. cellulitis
c. congenital nevus
d. ecchymosis
e. phakomatosis pigmentovascularis with Sturge-Weber syndrome

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The authors report no conflict of interest.
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THE **DIAGNOSIS:** Phakomatosis Pigmentovascularis With Sturge-Weber Syndrome

The erythematous patches were identified as capillary malformations (port-wine stains) and the slate gray pigmentary changes as dermal melanocytosis (Mongolian spots)(Figure). In fact, the diagnosis of phakomatosis pigmentovascularis (PPV) type II requires dermal melanocytosis and capillary malformation with and without nevus anemicus.[1] In one case series, 46% (7/15) of patients with PPV had nevus anemicus[2] but our patient did not.

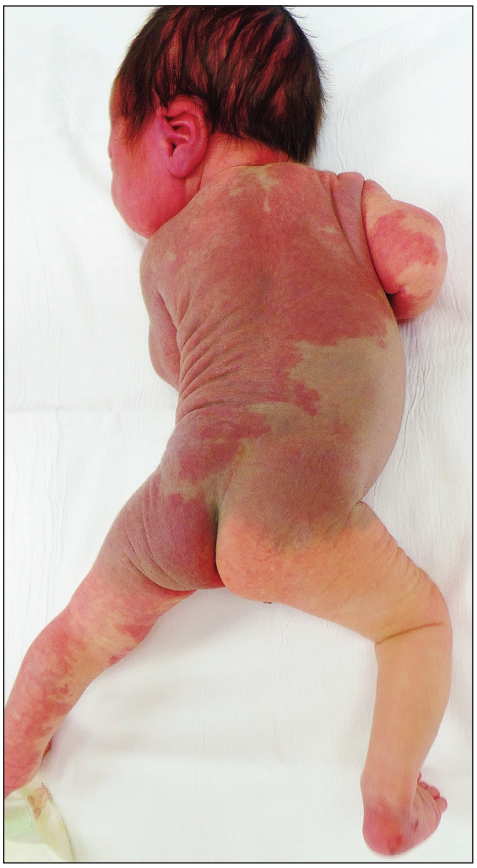
Phakomatosis pigmentovascularis was divided into 4 types in 1985,[3] then later 5 types.[4] Subcategories of the 5 types include type A, which denotes a lack of extracutaneous involvement, and type B, which is used when internal manifestations have been exhibited. Since 1947, approximately 222 cases of PPV have been described in the literature.[2]

A case of PPV associated with Sturge-Weber syndrome (SWS) was reported in 1997.[5] Since then, PPV occasionally has been linked with SWS,[5-9] though there have been other syndromic associations including Klippel-Trenaunay-Weber syndrome and melanosis oculi.[2] The incidence and prevalence of overlap of PPV and SWS is unknown but is likely to be rare. In our case, magnetic resonance imaging of the patient's brain did not reveal the characteristic tram-track appearance of SWS; however, the diagnosis of SWS type II only requires facial angioma with or without glaucoma.[9,10] Most cases of PPV originate from Japan, Argentina, and Mexico.[2] Interestingly, our patient's parents were both of Mexican ancestry. Phakomatosis pigmentovascularis type IIb is the most common, followed by type IIa.[2] Most cases have been described as sporadic, though our patient's mother also exhibited a port-wine stain on the right neck, suggesting a possible genetic association.

The etiology of PPV has been postulated as twin spotting or didymosis (Greek for twin), most commonly seen in plants and animals. A previous review defined twin spotting as 2 mutant tissues situated adjacent to one another and unique from the normal tissue surrounding both of them.[2] When the cell loses its heterozygosity, this phenomenon appears. An alternative etiology supplants that a drug or virus toxic to the nervous system causes aberrant angioblasts and melanoblasts.[11,12] The etiology of SWS also is unknown, though vasomotor instability has been postulated as a cause.[6,13]

It is important to exclude associated internal organ involvement with both of these syndromes because approximately 50% of PPV cases have extracutaneous organ involvement.[2,14] In fact, PPV is known to involve the brain, skeletal system, and eye, potentially manifesting as deafness, hydrocephalus, extremity overgrowth, scoliosis, cataracts, and more.[2] Patients with SWS often exhibit brain and eye symptoms including seizures.[1] To screen for extracutaneous involvement, multiple imaging studies should be performed. In our patient, an echocardiogram revealed a patent foramen ovale and normal cardiac anatomy for his age. Brain imaging revealed a hypoplastic left sigmoid and transverse sinus without venous thrombosis and unremarkable appearance of the brain. An ultrasound of the liver, spleen, kidneys, and pancreas revealed no evidence of solid, cystic, or vascular lesions, though the gallbladder exhibited hyperechoic areas.

To manage the skin lesions, some authors recommend Q-switched lasers for pigmented lesions and pulsed dye



Dermal melanocytosis (Mongolian spots) on the flanks and back.

lasers for capillary malformations.[15] Paller and Mancini[1] cited evidence that pulsed dye laser treatment before the age of 1 year may offer a psychological advantage, while other views have been offered.[16] Some physicians believe that no urgent treatment of capillary malformations is needed unless internal organs are involved.[2,15]

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