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Concurrent Sturge-Weber Syndrome, Facial Infantile Hemangioma, and Cutis Marmorata Telangiectatica Congenita



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PRACTICE POINTS

- This case highlights that several vascular malformations of different origins can occur in the same patient.
- Epigenetic factors along with genetic factors can lead to development of complex vascular conditions.
- Close collaborations of different medical specialties is necessary to make an accurate diagnosis and to follow up to achieve optimal long-term outcomes for patients with complex medical conditions.

We present a unique case of 3 vascular malformations—Sturge-Weber syndrome (SWS), facial infantile hemangioma (IH), and cutis marmorata telangiectatica congenita (CMTC)—with dermatologic manifestations presenting in the same patient. This case highlights the possibility of occurrence of multiple vascular malformations in the same patient; the potential role of epigenetic factors; and the importance of a multidisciplinary approach to diagnose, treat, and manage this complicated interplay of vascular abnormalities to achieve the best outcome.

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Sturge-Weber syndrome (SWS) is a disease of dermatologic, neurologic, and ocular significance.¹ The most distinctive manifestation is facial capillary malformation, commonly referred to as a port-wine stain or nevus flammeus. The dysregulated angiogenesis, caused by somatic mutations of the G protein subunit alpha Q gene, *GNAQ*, also affects the central nervous

system.² Seizures, intellectual disability, and glaucoma are common consequences.¹ Not all port-wine stains are associated with SWS.³ Distribution in the ophthalmic dermatome is associated with increased risk for SWS, with 8% of patients with port-wine stains also having SWS.⁴ The disease is more serious when bilateral lesions are present.⁵ Diagnosis is clinical based on dermatologic, nervous system, and ophthalmologic findings.⁶ The disease is nonheritable because the mutation is found only in the somatic cell lines.² The possibility of epigenetic influence on disease development has to be investigated. The treatment is aimed at managing complications, as there is no cure.⁷

Infantile hemangioma (IH) likewise represents a disruption in the process of vascular development but without the widespread consequences of SWS. The pathogenesis of hemangioma development has not been fully elucidated, with presence of GLUT1 (glucose transporter 1) protein implicated in lesions.⁴ Facial infantile hemangiomas have an incidence of approximately 5 in every 100 births, and the prevalence decreases with age. Most hemangiomas undergo growth followed by an involution process, with most lesions vanishing by 5 years of age.⁴ They typically are seen at 2 to 3 weeks of age, growing rapidly for the first 6 months, which is a contrast to the static nature of nevus flammeus. Infantile hemangiomas are regarded as sporadic, though autosomal-dominant inheritance patterns have been observed.⁴ Our patient demonstrated facial IH at birth, which is a rare and interesting finding suggesting that some epigenetic factors influenced this modification of the disease course in this patient.

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Cutis marmorata telangiectatica congenita (CMTC) is a rare cutaneous vascular condition found in newborns. Its extraordinary infrequency is reflected in the fact that only 300 cases have been reported worldwide.⁸ At birth, CMTC manifests as a pinkish reticulated pattern all over the body mimicking cutis marmorata; however, unlike cutis marmorata, the lesions do not improve with warming.⁹ The lesions of CMTC gradually lighten as the patient ages.⁸ Limb asymmetry is the most common extravascular complication of CMTC and, similar to SWS, glaucoma also can occur.¹⁰ Cutis marmorata telangiectatica congenita has been known to occur simultaneously with SWS or IH, but the combination of all 3 conditions in our patient is unique. Due to the scarcity of cases, the pathophysiology and treatment is poorly understood, with appropriate monitoring for sequelae recommended.⁹

Case Report

The patient was born at 39 weeks' gestation following an uncomplicated pregnancy and delivery. She weighed 2950 g, her length was 19 in, and her head circumference was 13.25 in, correlating to the 10th, 50th, and 25th percentiles, respectively. Her Apgar score was 8/9 at 1 and 5 minutes. Her parents were nonconsanguineous and in good health. The patient's family lived in poverty, which led us to conjecture about the role that toxins played in the epigenetics of the patient and her family. It was the mother's third pregnancy; both prior pregnancies resulted in healthy children. The patient was breastfed. No family history of heritable vascular disorders was reported.

On the first day of life during the newborn examination, dark red pigment changes were noticed under the nose and erythematous pigmentation over the whole body was observed (Figure). On examination, 2-toned reticular lesions identified as extensive nevus flammeus were found bilaterally over the distribution of the ophthalmic division of the trigeminal nerve. A separate erythematous plaque over the maxilla also was recognized. The pediatrician suspected SWS and facial IH. The patient

was discharged after 3 days with a referral to pediatric dermatology, and appropriate follow-up with a pediatrician was scheduled. The patient returned for these appointments and the significance of SWS was explained to her parents. Consultation with pediatric dermatology at 2 weeks of age confirmed the diagnosis of SWS as well as facial IH.

Upon further follow-up with pediatric dermatology at 2 months of age, the patient received an additional diagnosis of CMTC. These exceedingly rare lesions were located over the back, trunk, arms, and legs. The patient's parents were counseled about the management of these conditions and appropriate follow-up.

Comment

This case describes 3 different vascular malformations in the same patient. Cutis marmorata telangiectatica congenita is rare and yet is described in this patient along with 2 other notable endothelial abnormalities. The clinical interest of this case is heightened by the presence of CMTC.

The causative factor of SWS is a well-documented mutation of the *GNAQ* gene, but there is considerable variability in how it affects the patient. Unlike in SWS, no single factor can be attributed to the development of IH. This case shows that these 3 diseases are not mutually exclusive and can present with unusually severe features when they occur concomitantly. The embryologic basis of SWS traces its roots back to the first trimester during vascular development, where lack of regression and development of the primitive cephalic venous plexus occur.¹⁰ The presence of a large IH on the patient's philtrum that demonstrated markers of pericyte and neural crest cells illustrates that the developmental origins of one neurocutaneous disorder do not necessarily interfere with the development of other cutaneous conditions.¹¹

The severity of the SWS in our patient was highlighted by the extensive nevus flammeus. These lesions occurred over the face, trunk, arms, and legs. The port-wine stain



A diffuse purple and pink reticular pattern over the entire back can be observed in a patient with concomitant Sturge-Weber syndrome, facial infantile hemangioma, and cutis marmorata telangiectatica congenita (A). On the face, an infantile hemangioma and distribution of nevus flammeus over the frontal bone denoted Sturge-Weber syndrome (B).

with dermatomal distribution of the ophthalmic nerve was the most concerning feature regarding the development of neurologic complications in this patient. Although the developmental delays associated with SWS can be serious, early intervention is important and can improve long-term outcomes. The facial IH arising at birth was contrary to the typical presentation. All of these factors will be kept in mind as the patient progresses and patient-centered care is provided. Because this patient's presentation differed from other patients with IH, we will be more vigilant in providing close follow-up and monitoring for other medical problems involving other organs (eg, the brain); for instance, we will monitor for seizures and developmental delay.

Conclusion

In our patient, a unique pattern of SWS, facial IH, and CMTC are described in a pediatric patient. Many disciplines are involved in the treatment. In the patient's first days of life, extensive collaboration between pediatrics and dermatologists was pivotal, with ophthalmology, pathology, and radiology consultations at hand. This case highlights that several vascular malformations of different origins can occur in the same patient. Epigenetic along with genetic factors likely contributed to this fascinating presentation. The importance of parental education and maintaining appropriate follow-up for this patient is crucial for a favorable outcome.

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