Acquired Port-Wine Stain (Fegeler Syndrome): A Case Report and Literature Review

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Abstract

Acquired port-wine stains are a type of capillary malformation rarely reported in the literature. Most documented cases are idiopathic in nature or caused by physical trauma. We describe a case of a 61-year-old man with an acquired port-wine stain in the left V1 distribution with ipsilateral ophthalmic findings, and hereby recommend an ophthalmologic exam for patients who present with acquired port-wine stains in the V1 trigeminal distribution.

Introduction

Port-wine stains (PWSs) are cutaneous capillary malformations, also known as nevus flammeus, nevus simplex or salmon patch, and are usually considered congenital vascular lesions. First described by Fegeler in 1949, acquired PWSs are exceedingly rare but have been previously reported and documented.1 A recent literature search revealed that fewer than 100 of these lesions have been described.2 Most cases are idiopathic, but trauma may be a precipitating factor.3-5 Whether congenital or acquired, PWSs usually present as irregularly bordered, violaceous-to-red patches and plaques, many of which follow the V1 or V2 distribution. Congenital PWSs result from abnormal vessel development during embryogenesis, with histopathology revealing an increased number and ectasia of blood vessels in the dermis.6 Herein, we present a case of an acquired port-wine stain, also known as “Fegeler syndrome,” and a review of the literature.

Case Report

A 61-year-old Caucasian male presented after being referred for ongoing rosacea around his left forehead, eye and nose. The patient gave a history of the “rash” appearing suddenly one morning about 19 years ago. He could not account for any sort of precipitating factor such as trauma to the area, recent infections, or new medications. He described the lesions as being occasionally pruritic and slightly painful, which became more noticeable with sweating. The patient had no history of shingles. Dermatologic exam showed a patchy and somewhat coalescing, red-pink, vascular-like lesion that extended from the left eyebrow to the distal left nasal tip and involved the inferior left eye and cheek region (Figure 1a). Previous ophthalmologic examinations revealed periorbital hemangiomas, with vision changes consistent with increased intraocular pressure; and blurred vision and brown tint in the left eye for approximately eight months. Also described were benign, age-related ophthalmologic findings. His surgical, family and social history were non-contributory.

A punch biopsy from the left nasal sidewall was taken, and histopathologic exam revealed a vascular lesion with associated vascular ectasia in the surface (Figure 2, H&E). The vessels were lined by flattened endothelial cells, some of which showed slight hyperchromasia, while others showed a thickened vascular wall. No infiltrative pattern of the lesion was seen. No mitotic figures were present. A CD34 immunostain was positive in the endothelial lining of the vessels. D2-40 was negative in the lesional cells, ruling out the presence of lymphatic cells. A diagnosis of a vascular proliferation consistent with a port-wine stain was made.

Our patient successfully underwent two treatments with a V-beam pulsed dye laser (PDL) set at: spot size: 7 mm², fluence: 13J/cm², pulse duration: 1.5 ms. He showed much improvement (Figure 1b) after two treatments and is scheduled for two more PDL sessions.

Discussion

The pathogenesis of acquired PWS is not completely understood. A history of trauma is given in approximately half of all documented cases. Our patient denied any prior trauma. Whether idiopathic, trauma-related, or from other proposed causes, the exact reason these vascular malformations become chronic and sometimes lifelong lesions has yet to be elucidated. One hypothesis points to non-proliferative vascular ectasia due to a defect in nerve fibers associated with these blood vessels, resulting in decreased sympathetic tone.7 They may also be associated with malformations at the post-capillary venule.8 Other studies suggest abnormalities in blood-vessel connective tissue and associated nerve supply.9

While it is known that patients with certain vascular lesions, such as infantile hemangiomas, may have other organ systems involved (e.g., hepatic), little is known about systemic involvement in acquired PWSs. There are many known syndromic diseases featuring congenital PWSs with a constellation of other organ systems involved (Sturge-Weber syndrome, Klippel-Trenaunay syndrome, Proteus syndrome, phakomatosis pigmentovascularis, and possibly tuberous sclerosis), but none so far are associated with acquired PWSs. The patient described in this case did have some ocular involvement, as his PWS involved the lower eyelid. He also had ipsilateral ophthalmic findings, including a posterior vitreous detachment (PVD) and an epi-retinal membrane (ERM). PVD is insidious
and asymptomatic, but it may lead to more serious macular and optic disc disease.\textsuperscript{10} ERM is also a benign ocular condition but may lead to visual impairment and necessitate retinal surgical intervention.\textsuperscript{11} While both of these can be normal age-related eye conditions, the ipsilateral nature and timing of both cutaneous and ophthalmic findings is a conspicuous association.

PWSs in the V1 distribution can be a strong predictor of neuro-ocular involvement.\textsuperscript{12} In our case, the patient’s PWS appeared several years before his ophthalmologic findings, and while benign in nature, our patient did state some increasing left-side blurriness. As such, we recommend an ophthalmologic exam for any patient presenting with an acquired PWS involving the V1 or V2 distribution.

Some common entities that could be included in the differential diagnosis of PWS include various forms of hemangiomas (e.g., glomeruloid hemangiomas), tufted angiomas and Kaposi sarcoma. Other considerations for differential, hemangiomas), tufted angiomas and Kaposi sarcoma. Other considerations for differential, PWSs including the V1 or V2 distribution.

A more recent study by Parsa et al. has suggested that PWSs are due to intracranial circulation abnormalities and may result in cutaneous findings, implying that SWS is a product of “acquired venous obstruction rather than neural dysfunction.”\textsuperscript{15} Other studies suggest abnormalities in blood vessel connective tissue and associated nerve supply.\textsuperscript{16-17} Histopathological evidence supports the neural mechanism theory of venous ectasia as documented by decreased nerve density within cutaneous biopsy specimens.\textsuperscript{15} Considering the aforementioned, it is plausible to say that acquired PWS may be due to an occlusive event in the cutaneous vasculature, whether traumatic or thrombotic in nature, and that the precise etiology on a molecular level may be neural or strictly vascular.

Genetic studies have indicated the presence of a GNAQ somatic activating gene mutation that encodes p.Arg183Gln amino acid substitutions in skin and brain tissue from patients with Sturge-Weber syndrome as well as those with non-syndromic PWS.\textsuperscript{19} The gene makes a Gq protein whose cell surface receptors, when activated by ligand, bind and hydrolyze GTP. This initiates an intracellular MAPK signaling cascade. The mutation locks Gq into a mildly activated state. This supports the long-standing hypothesis that SWS and acquired port wine stains are caused by the same underlying somatic mutation dependent on when and where in development the somatic mutation occurs.\textsuperscript{20-21} Given this genetic activating mutation, it can be postulated that the venodilatation observed in port wine stains is due to the increased GTP, which leads to smooth muscle relaxation in walls of post-capillary venules.

**Conclusion**

Acquired PWSs are much less common than their congenital counterparts. While not cataloged or classified as being part of any syndromic condition, their presence near the eye or in the V1 or V2 distribution warrants an ophthalmology workup to rule out any associated malignant or potentially serious sequelae. PWSs, whether acquired or congenital, may respond well to PDL laser therapy. Our patient was fortunate to respond well after only two treatments, as facial and distal-limb PWSs can be more resistant to laser therapy. Other treatment modalities include embozolization or skin grafting, but these options require an extensive multi-disciplinary approach.

**References**


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