Capillary Malformations (Portwine Stains) of the Head and Neck

Natural History, Investigations, Laser, and Surgical Management

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KEYWORDS

• Capillary malformation • Port-wine stain • Natural history • Laser
• Surgical treatment

KEY POINTS

- Facial (capillary malformation) CM often occurs with a quasidermatomal distribution according to the sensory trigeminal nerve distribution.
- With time, CM darkens progressively, and soft tissue hypertrophy, bony hypertrophy, and/or nodule formation can develop.
- The mainstay and gold standard therapy for facial or aesthetically sensitive CM is still the pulsed dye laser (PDL) treatment.
- In patients with associated soft tissue/bony hypertrophy, surgical management is helpful in restoring the normal anatomy and in re-establishing a symmetric contour.

CLINICAL FEATURES

Capillary malformations (CMs), also known as port-wine stains or nevus flammeus, are the most common type of congenital vascular malformations. They are present at birth and persist throughout life.[1–3] These lesions are initially flat and bright pink, red, or violaceous and typically affect the face (90%), followed by the neck, trunk, leg, arm, and hand.[4–6] They often seem to lighten significantly over the first few months of life. This is not indicative of spontaneous resolution, but it is probably caused by a drop in circulating blood hemoglobin concentration.[7] In contrast to other similar

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birthmarks, most CMs become darker, thicker, and more nodular over time. This is particularly true of facial lesions.[8] The incidence rate is reported as 0.3% in newborns with an equal sex distribution, occurring spontaneously within the population.[9] In most affected individuals, CMs occur as a sporadic unifocal lesion and are not associated with any underlying abnormalities. However, CMs are sometimes associated with other underlying syndromes, such as Sturge-Weber syndrome; macrocephaly-CM syndrome; CM-arteriovenous malformation (AVM) syndrome; and overgrowth syndromes, such as Klippel-Trénaunay syndrome.[10,11]

The pathogenic mechanism of CM is still unknown. Shirley and colleagues[12] identified a somatic mutation in *GNAQ* with isolated CMs, disrupting vascular development. *GNAQ* encodes a guanine nucleotide-binding protein G(q) subunit alpha that mediates signals between G-protein-coupled receptors and downstream effectors. It has also been identified that the subclass of CMs associated with AVMs has an autosomal-dominant mutation in the *RASA1* gene, encoding Ras1, a GTPase-activating protein involved in cell growth, proliferation, and differentiation during angiogenesis.[13–16] Recently, Frigerio and colleagues[17] reported that novel somatic variants were found in *GNAQ* and *RASA1*.

Histologically, CMs are initially composed of a normal capillary network in the papillary dermis with no evidence of cellular proliferation; ectatic vessels with a small venular morphology in the papillary and occasionally reticular dermis become more evident over time.[2,18]

Facial CMs initially appear as a faint pink macule; however, some patients may develop soft tissue hypertrophy, bony hypertrophy, and/or nodule formation during adulthood.[5,6,19,20] Depending on the size and location, these changes can cause functional deficits in vision, speaking, or eating and significant psychological distress related to the resulting stigmatization or disfigurement.[19] The mucosa of the oral cavity, gingiva, tongue, larynx, nasal mucosa, soft tissue of the neck, and even the parotid gland have been demonstrated as possible sites of manifestation, leading to severe conditions, such as macrocheilia, painful cervical or parotid swelling, globus pharyngeus, dysphonia, dysphagia, gingival bleeding, epistaxis, and nasal and upper airway obstruction.[21]

Although multiple treatments have been reported, the mainstay and gold standard therapy for facial or aesthetically sensitive CMs is still the flashlamp-pumped pulsed dye laser (PDL) treatment.[22,23] However, laser therapy has a limited benefit for long-standing CMs with soft tissue hypertrophy, and a more satisfactory result is achieved after surgical intervention.[5,24]

Local complications of CMs include pyogenic granulomas and eczematous dermatitis occurring within the stain.[25–27] Pyogenic granulomas need to be excised or treated with electrodessication and curettage because they do not resolve spontaneously and are complicated by repeated bleeding and discomfort.

The differential diagnoses of CMs include other vascular lesions, including early infantile hemangioma, vascular stain associated with AVMs, and other vascular malformations. Doppler assessment may be helpful in differentiating between CMs and AVMs, based on a bruit and thrill associated with AVMs. Sometimes, it is difficult to distinguish between early CMs and infantile hemangiomas located on the face during infancy; the distribution (dermatomal in CM vs segmental in infantile hemangiomas) is a clue.[28] The endothelial cells of CMs do not stain for GLUT-1, a specific marker for infantile hemangiomas.

NATURAL HISTORY

Facial CMs often occur with a quasidermatomal distribution according to the sensory trigeminal nerve distribution: V1, the ophthalmic region, including the forehead and