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Phakomatosis Pigmentovascularis Type V

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An otherwise healthy 11-week-old boy presented with multiple skin abnormalities that had been present since birth. He had been born at 39 weeks of gestation and had required intubation after delivery, but he recovered well and had been discharged 2 days later.

Physical examination. At presentation, cutaneous examination revealed 3 notable findings: (1) a pink flat patch on the left face involving the upper cutaneous lip (with sharp midline cutoff), left cheek and nasal sidewall, and left lower eyelid (**Figure 1**); (2) reticular pink patches and macules in livedo configuration involving all 4 extremities and the torso (**Figures 2 and 3**); and (3) large faint gray-blue patches on the buttocks and central midback (**Figure 4**).

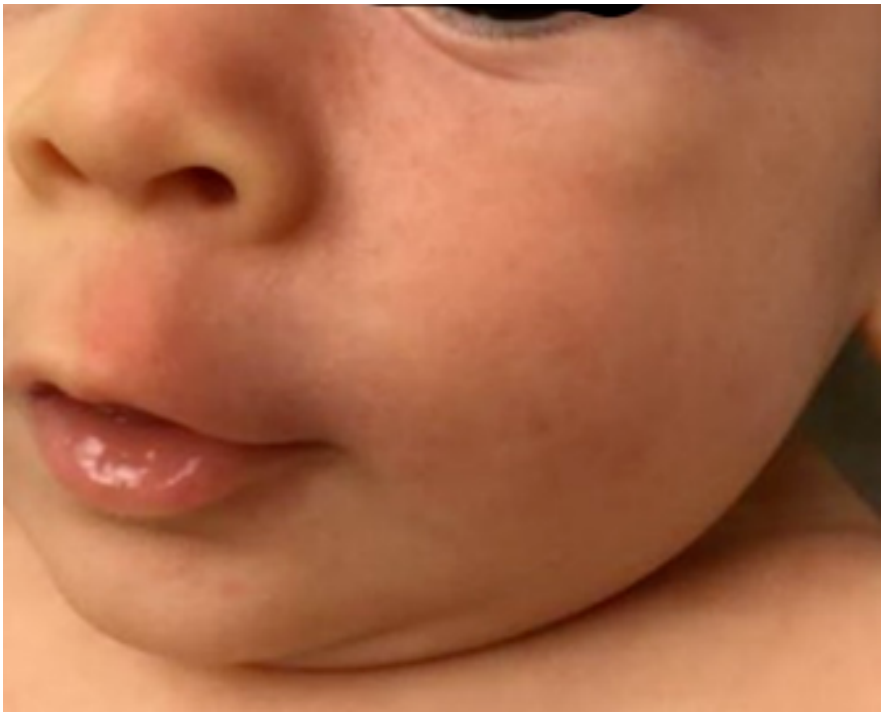

Figure 1. *Facial capillary malformation on the left face.*

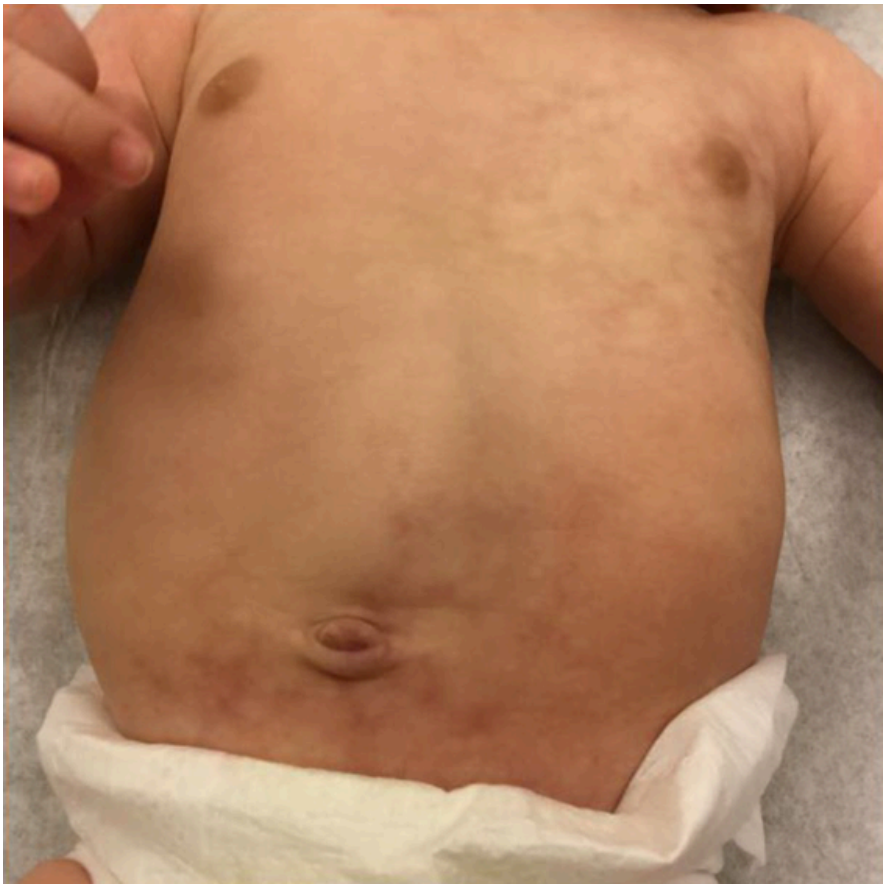

Figure 2. *Cutis marmorata telangiectatica congenita on the trunk.*

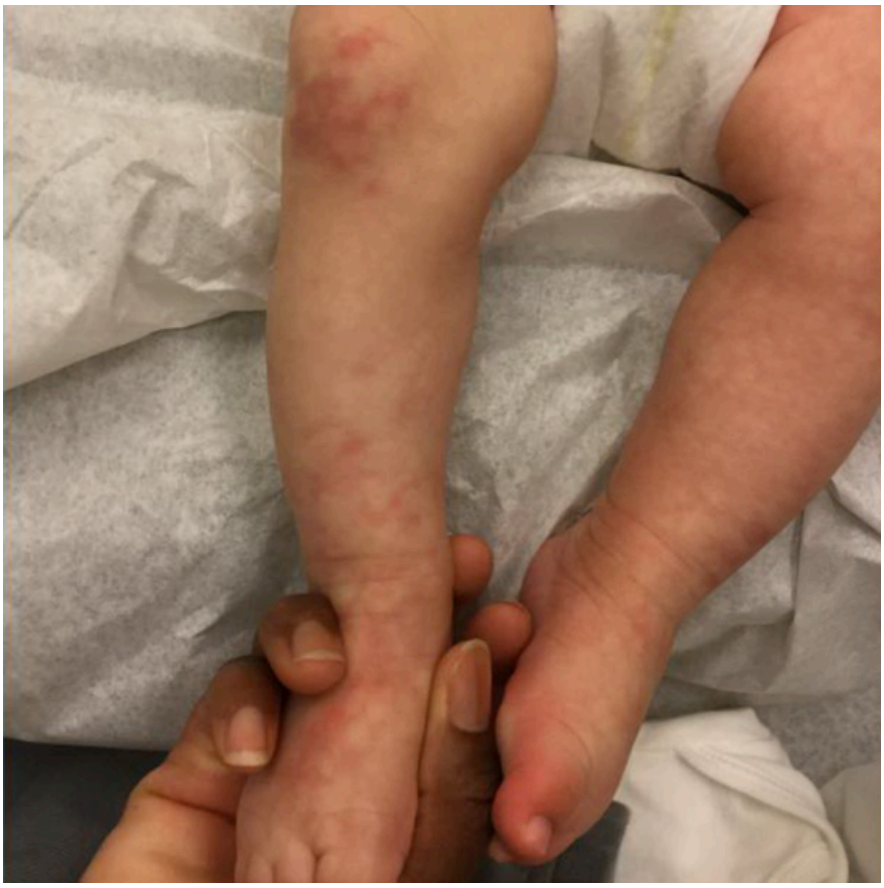

Figure 3. *Cutis marmorata telangiectatica congenita on the extremities.*

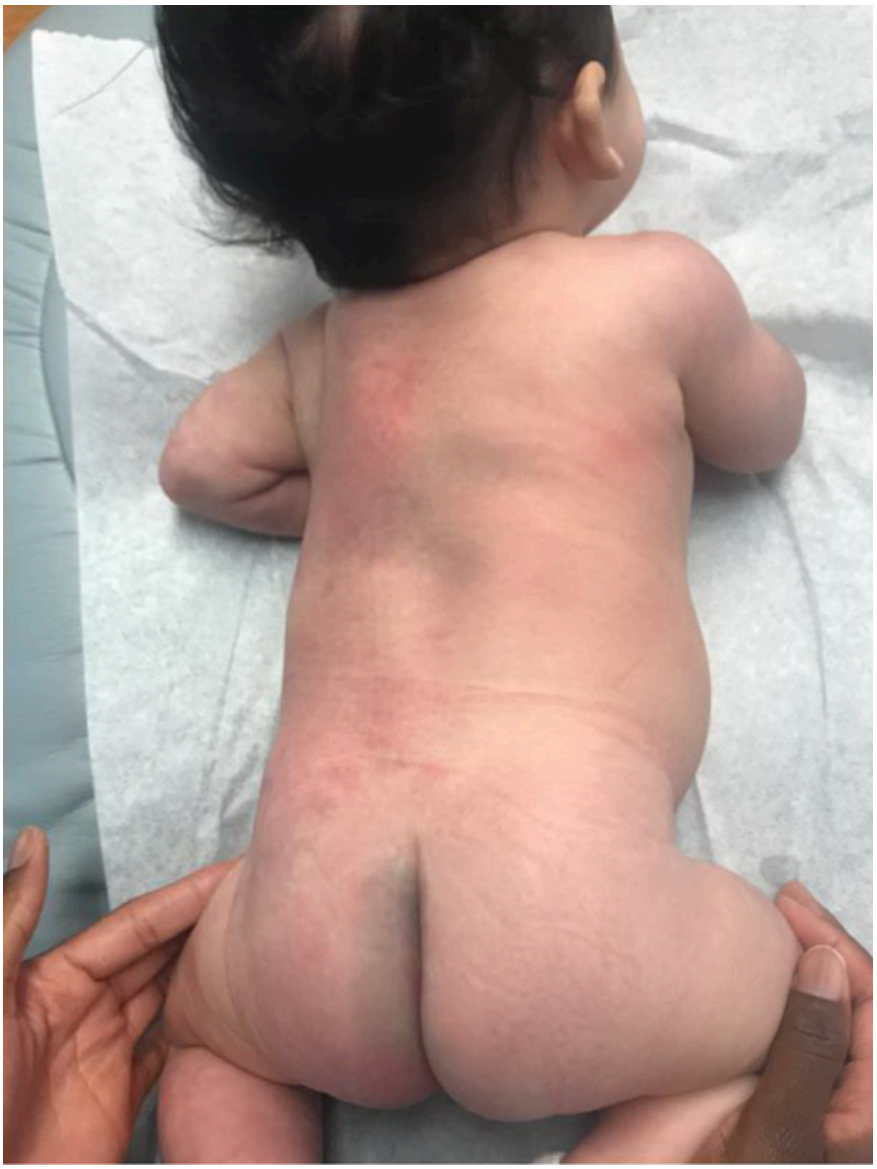

Figure 4. *Dermal melanocytosis on the buttocks and central back.*

The patient was referred to dermatology, where no gross asymmetry was noted, and head circumference was at the 75th percentile, although his parents reported a personal history of large head sizes.

Upon follow-up evaluation at 13 months of age, the patient's left leg was noted to be grossly larger than the right, and additional workup by specialists in orthopedic surgery, genetics, and imaging were pursued. Orthopedic evaluation revealed hemihypertrophy of the left leg. Genetic testing revealed normal DNA microarray results and normal findings of a workup for Beckwith-Wiedemann syndrome. Abdominal ultrasonography findings were normal without hypertrophy or neoplasm.

Based on his clinical presentation of facial capillary malformation, cutis marmorata telangiectatica congenita, and dermal melanocytosis, the boy received a diagnosis of phakomatosis pigmentovascularis (PPV) type V.

Discussion. PPV is a rare congenital syndrome characterized by the association of a pigmentary nevus with a cutaneous vascular malformation. PPV was originally classified in 1985 into 4 types based on the epidermal component that accompanies the vascular malformation.[1] In 2003, a fifth type involving cutis marmorata telangiectatica congenita (CMTC) and dermal melanocytosis (nevus cesius) was first described.[2] In 2005, Happle proposed a new and simplified classification system using descriptive terms, in which PPV type V, or phakomatosis cesiomarmorata, is characterized by the presence of nevus cesius and CMTC with no other additional skin lesions.[3] To date, only 9 cases of PPV type V have been reported in the literature.[4,5]

Pigmented birthmarks such as dermal melanocytosis are a common occurrence in newborns. Dermal melanocytosis is particularly common in skin of color, with an estimated 80% to 90% of newborns in certain black and Asian populations affected, and as few as 3% to 10% of white neonates.[6] However, these lesions are often transient, and it is very likely that dermal melanocytosis is underreported.

Dermal melanocytosis involving unusual or large areas of the body, as well as more permanent lesions, have been associated with other anomalies such as lysosomal storage disease and ocular disorders.[7] In combination with vascular malformations in PPV, dermal melanocytosis is associated with certain ocular, neurologic, skeletal, and neoplastic complications that occur in as many as 50% of reported patients.[8]

With underreported lesions and an increased prevalence of offspring from parents of discordant skin Fitzpatrick phototypes, we speculate that the impact of these pigmentary birthmarks on overall diagnosis and management is relatively small. The prevalence of persistent dermal melanocytosis and the impact of these lesions is only recently being better understood.[9]

In contrast, the systemic consequences of various presentations of capillary malformations and their associated syndromes are better understood. The distribution of facial capillary malformation, particularly if involving V1 trigeminal distribution, may indicate risk of Sturge-Weber syndrome and prompt imaging of the brain.[10,11] Limb hemihypertrophy in the setting of capillary malformation, macrocephaly, and distribution of a capillary malformation on the face are suggestive of overgrowth syndromes such as macrocephaly-capillary malformation or diffuse capillary malformation with overgrowth (DCMO) and should prompt imaging with complete abdominal ultrasonography and surveillance for Wilms tumor.[12] It is important to consider workup for additional overgrowth syndromes, such as Klippel-Trenaunay syndrome, Parkes Weber syndrome, CLOVES (congenital lipomatous overgrowth, vascular malformations, epidermal nevi and scoliosis/skeletal/spinal anomalies) syndrome, and Proteus syndrome as appropriate.

The exact mechanism of pathogenesis of both pigmented and vascular birthmarks is not well understood but is theorized to be a postzygotic mosaic disorder with activating mutations in *GNA11* and *GNAQ*, which have also been implicated in the pathophysiology of Sturge-Weber syndrome, overgrowth syndromes, and uveal melanomas.[9] While some relevant pigmented and vascular birthmarks naturally fade over time, such as banal dermal melanocytosis and capillary malformations in distributions of the eyelids (previously termed *nevus simplex*), the timing of diagnosis may play a role, and the significance of these lesions relevant to overall health is potentially minor.

Outcome of the case. The patient continued to reach normal developmental milestones at 22 months, including gross and fine motor skills and speech milestones. This evaluation also revealed normal development of the hips with full range of motion, and the patient had begun walking at 10 months without any apparent difficulty; thus, no interventions were recommended. His head circumference has remained stable in the 75th percentile.

The patient will be monitored over time for possible DCMO and will continue to undergo serial abdominal ultrasonography to monitor for additional complications including Wilms tumor. The patient also was to receive pulsed-dye laser treatments for lightening of the facial capillary malformation.

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