# Midline Skin Anomalies: A Small Clue to a Larger Diagnosis

A 3-month-old girl, born at term, presented for evaluation of multiple vascular skin lesions on the face. The lower-lip lesion had been noted at 2 weeks of age, with the others developing over weeks on the right temple, left chin, left ear, and submental region. The lesions appeared to be asymptomatic without any bleeding or breakdown, and her parents denied breathing or feeding difficulties. She was otherwise healthy and was reaching normal developmental milestones.

Physical examination revealed a vascular red thin plaque on the right lower lip (**Figure**, A), with speckled vascular papules on the left temporal scalp (**Figure**, B), left neck, and left chin and submental region (**Figure**, C), all felt to be consistent with infantile hemangiomas. There was no hoarseness or stridor. Full skin examination was performed and revealed an atrophic sternal pit and a smaller midline superior abdominal atrophic defect (**Figure**, D). Given her constellation of findings, magnetic resonance (MR) imaging and MR arteriography of the head and neck were performed, as were echocardiography and flexible laryngoscopy. Her MR imaging was unremarkable for structural brain anomalies but revealed a large left parotid infantile hemangioma, whereas MR arteriography revealed a markedly diminutive left cervical internal carotid artery with tortuosity and a severely hypoplastic right A1 segment. The posterior inferior cerebellar artery was also tortuous with a significantly hypoplastic right V4 segment distal to the posterior inferior cerebellar artery origin. Echocardiography revealed a trivially patent foramen ovale with left-to-right shunt but was otherwise normal. Laryngoscopy was unremarkable, as were her audiologic and ophthalmologic examinations. Thyroid function testing was normal. This constellation of clinical and radiographic findings was diagnostic of PHACE (posterior fossa anomalies, hemangioma, arterial lesions, cardiac abnormalities/coarctation of the aorta, eye anomalies)

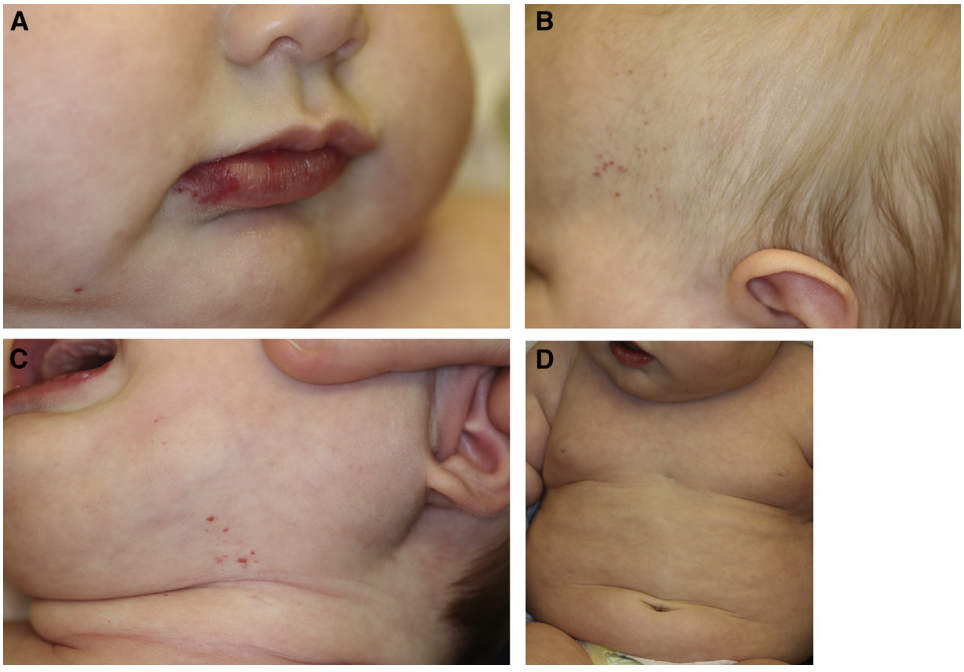


**Figure.** **A,** Vascular thin plaque on the right lower lip, representing infantile hemangioma. Speckled papular infantile hemangiomas were also present, as shown on the **B,** left scalp, and **C,** left chin and submental region. **D,** An atrophic sternal pit and midline superior abdominal atrophic defect also were noted.

A.M. serves on the Editorial Board of *The Journal of Pediatrics*. The other authors declare no conflicts of interest.

J Pediatr 2021;232:299-300.

https://doi.org/10.1016/j.jpeds.2021.02.023

syndrome, and the patient was referred for neurologic consultation.

PHACE syndrome is an acronym that was coined in 1996 to describe this constellation of clinical features,[1] with consensus-derived diagnostic criteria that were updated in 2016.[2] The syndrome is variably referred to as PHACES, to account for the midline chest and abdominal anomalies (including sternal pits and clefts and supraumbilical raphe). Ventral midline skin blanching also has been described.[3] Although the infantile hemangiomas in PHACE syndrome are typically large and segmental (lesions that cover a larger anatomic territory of the face or body), they can be more subtle, as demonstrated in our patient. However, their multifocal nature, in conjunction with the midline developmental defects, heightened our concern for this syndrome, prompting further evaluations that confirmed the diagnosis. Given that the cerebral arteriopathy in PHACE syndrome may predispose patients to arterial ischemic stroke and impact risk stratification of infantile hemangioma treatment with beta blockers, as well as other potential long-term morbidities, prompt recognition and diagnostic confirmation are vital. We present this patient to highlight the importance of meticulous skin examination in this setting, including evaluation for subtle midline developmental anomalies, which may serve as a critical diagnostic finding. ■

**Jinia R. El-Feghaly, MD**
**Matilde Krisha P. Montenegro, MD**
Division of Pediatric Dermatology
Ann & Robert H. Lurie Children's Hospital of Chicago
Department of Pediatrics
Northwestern University Feinberg School of Medicine

**Anthony J. Mancini, MD**
Division of Pediatric Dermatology
Ann & Robert H. Lurie Children's Hospital of Chicago
Departments of Pediatrics and Dermatology
Northwestern University Feinberg School of Medicine
Chicago, Illinois

## References

1. Frieden IJ, Reese V, Cohen D. PHACE syndrome. The association of posterior fossa malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, and eye abnormalities. Arch Dermatol 1996;132:307-11.
2. Garzon MC, Epstein LG, Heyer GL, Frommelt PC, Orbach DB, Baylis AL, et al. PHACE syndrome: consensus-derived diagnosis and care recommendations. J Pediatr 2016;178:24-33.
3. Feigenbaum DF, Sybert VP, Vanderhooft SL, Siegel D, Drolet BA, Frieden IJ, et al. Ventral midline blanching in the setting of segmental infantile hemangiomas: clinical observations and pathogenetic implications. Pediatr Dermatol 2015;32:180-7.

# Isolated Forehead Swelling


A previously healthy 3-year-old girl presented to the dermatology department with a 3-week history of swelling of the right forehead. The lesion was a nonpainful, nonfluctuant, nonpulsating, indurated mass of about 3 × 3 cm, increased in size over the past 2 weeks, and with overlying skin eventually turned purple (**Figure 1**, A). Ultrasound examination revealed a solid hyperechoic subcutaneous lesion with enhanced Doppler signal, intact underlying osseous plate, and ipsilateral submandibular and cervical lymphadenopathy. Abdominal ultrasound and chest radiogr aph were normal. Skull radiograph ruled out osteolytic bone damage. Laboratory tests showed isolated high white blood cell (WBC) count (16 600/mm$^3$: neutrophils 10 800, lymphocytes 3700, monocytes 880, eosinophils 1000) with no elevation of inflammatory markers, and lactate dehydrogenase, ferritin, and neuronal specific enolase within the normal range for age. No peripheral blood smear was obtained on that occasion. In the hypothesis of a skin infection, a course of oral amoxicillin-clavulanate was started.

Ten days later, the patient was brought to the emergency department for high fever and severe back pain: the skin lesion had further increased in size despite the antibiotic therapy. Repeated blood tests showed WBC count increased up to 25 000 cells/mm$^3$ (neutrophils 13 000, lymphocytes 6790, monocytes 3910, eosinophils 1000) with mildly elevated C-reactive protein (41 mg/L). A peripheral blood smear showed the presence of 47% monocytic-like blasts (**Figure 2**, A). Nevertheless, the bone marrow aspirate displayed a predominance of lymphoid blasts (**Figure 2**, B), and the immunophenotypic markers confirmed the diagnosis of common B-cell acute lymphocytic leukemia (ALL). A leukemia cutis with newly onset systemic ALL was diagnosed. Steroid prephase was started, and the mass completely resolved by day 8 (**Figure 1**, B).

Leukemia cutis consists of the infiltration of lymphoid or myeloid blasts into the epidermis, dermis, or subcutaneous tissues. It can manifest as petechiae, purpura, macules, papules, patches, plaques, or nodules, mainly involving the head and the lower extremities. Leukemia cutis is most commonly associated with acute myeloid leukemia, and in this case, it is specifically labeled as myeloid sarcoma,

J Pediatr 2021;232:300-2.

https://doi.org/10.1016/j.jpeds.2021.01.001