

improved sodium 147 mEq/L, blood urea nitrogen 18 mg/dL, creatinine 0.93 mg/dL, and serum osmolality 303 mOsm/Kg. Urine osmolality was 366 mOsm/Kg and serum copeptin 11 pmol/L; both were inappropriate for the serum hyperosmolality, consistent with partial central diabetes insipidus.

Magnetic resonance imaging of the brain showed a dysplastic subcallosal region and focal area of midline fusion anterior to the anterior commissure, consistent with septopreoptic holoprosencephaly (HPE) (Figure). The pituitary gland was of normal size but had an atypical convex superior margin.

HPE, the most common forebrain malformation, results from incomplete division of forebrain into right and left cerebral hemispheres.<sup>1</sup> The common subtypes include alobar, lobar, and semi-lobar HPE. Septopreoptic HPE is mild form of HPE in which nonseparation is limited to the septal (subcallosal) and/or preoptic regions. Patients with septopreoptic HPE often have mild midline craniofacial malformations, including single median maxillary central incisor.<sup>2</sup> Our patient had a history of single median maxillary central incisor that was removed during cleft lip and palate repair. Diagnosis may be delayed in patients with normal or mildly abnormal facies and mild features and may be diagnosed only after neuroimaging studies reveal HPE, as occurred in our patient.<sup>1</sup> Involvement of osmoreceptors in the anterior hypothalamus can cause impaired or absent thirst,<sup>3</sup> and chronic, inadequate fluid intake can result in hypernatremia, as was seen in our patient. Partial diabetes insipidus is due to abnormalities in the hypothalamic regions responsible for release of arginine vasopressin. Our patient had a variable appearance of the posterior pituitary bright spot (Figure), which has been reported to reflect fluctuating vasopressin stores.<sup>4</sup>

The patient tolerated mild hypernatremia and only became symptomatic (although still without increased thirst) when the sodium concentration approached 180 mEq/L. Following discharge from the hospital, he remained stable and symptom free, with serum sodium values between 143 and 148 mEq/L on a fixed daily fluid regimen of 1920 mL. ■

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## Limb Overgrowth with Vascular Anomalies

**A** 17-year-old girl presented with progressive overgrowth in the right lower extremity, varicose veins, and ecchymosis. She had been followed up in another medical center for 8 years with a diagnosis of vascular malformations and Kasabach-Merritt syndrome. Physical examination revealed significantly enlarged right lower extremity with a port-wine stain and varicosities. Laboratory evaluation revealed normal results, except for thrombocytopenia (thrombocyte count, 88 000; normal range, 150 000-400

000). Computed tomography (CT) angiography of the lower extremity was performed to investigate the presence of high-flow vascular components. CT angiography demonstrated overgrowth of the right limb with an arterial blush adjacent to the posterior tibial artery, consistent with a high-flow vascular malformation (Figure, A-C). Moreover, venous phase CT showed hypervascularity in the venous components (Figure, D). These imaging findings suggested the diagnosis of Parkes Weber syndrome; genetic testing revealed the RASA1 gene mutation and confirmed the diagnosis. Angioplasty and embolization were performed on the high-flow vascular component, and sclerotherapy was performed on the low-flow components.

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**Figure.** **A**, Three-dimensional volume-rendering CT angiography image of the limbs shows overgrowth of the right limb (*arrows*) with an arterial blush adjacent to the posterior tibial artery (*arrowhead*), consistent with a high-flow vascular malformation. **B**, Three-dimensional volume-rendering CT angiography image shows arterial blush adjacent to the posterior tibial artery (*arrowhead*). **C**, Coronal CT angiography image of the limb shows arterial blush adjacent to the posterior tibial artery (*arrowhead*). **D**, Venous phase CT of the limb reveals varicose veins in the right limb.

Parkes Weber syndrome is a rare congenital disorder characterized by combined high and low flow vascular malformations with an overgrowth of an entire limb.<sup>1</sup> Patients with Parkes Weber syndrome are usually misdiagnosed as Klippel-Traunanay syndrome due to overlapping clinical and radiologic findings. Klippel-Traunanay syndrome is characterized by low-flow vascular malformations.<sup>1,2</sup> The prognosis of Parkes Weber syndrome is worse because of high-flow and progressive vascular malformations, and therefore the distinction is important.<sup>1</sup> Radiologic findings are helpful in the detection, characterization, and in determining the severity of vascular malformations. ■

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