Klippel-Trénaunay Syndrome and Arnold-Chiari Type I Malformation

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To the Editor:

Klippel-Trénaunay syndrome is a rare disease of unknown etiology that affects 1 in 27,500 newborns.1 It is usually located on the limbs of one side, typically the lower limbs, and is characterized by combined or complex vascular malformation (International Society for the Study of Vascular Anomalies classification) with venous, lymphatic, and capillary disorders associated with hypertrophy of adjacent soft tissue and bone growth. The diagnosis is primarily based on the clinical symptoms, although neuroimaging procedures may be required on occasion. Its course is benign, but progressive, and treatment is usually conservative, consisting of treating the complications of the disease.2,3 It has been described in association with vascular malformations and other cardiovascular, skeletal, gastrointestinal, or neurological congenital abnormalities. Exceptionally, it presents along with other skin processes such as phakomatosis pigmentovascularis.4

Arnold-Chiari malformation is a neurological disease of unknown etiology believed to be due to insufficient development of the posterior cranial fossa, with the resulting expansion of the cerebellum toward the spinal canal. Four variants of the malformation have been described, of which type I is the most common. This type is characterized by caudal displacement of the cerebellum with tonsillar herniation below the foramen magnum and wedge-shaped elongation of the tonsils. Arnold-Chiari malformation is more common in women (1:3) in the fifth decade of life. Diagnosis is based on the typical magnetic resonance findings described above. The most common symptoms are occipital headache, triggered by Valsalva maneuver or neck extension, retroorbital pain, and visual disorders or symptoms simulating Ménière disease with hearing loss, vertigo, and tinnitus.5 Additionally, compression of the brainstem can lead to hydrocephalus or syringomyelia in up to 40% of cases.6

We describe a 54-year-old patient with a history of thalassemia minor who had undergone surgery for L5-S1 disc herniation as well as for a tumor of the right parietal bone in 2000, for which no written reports were provided. Following a dermatological assessment in 2003, she was diagnosed with Klippel-Trénaunay syndrome due to a nevus flammeus vascular malformation on the left leg with well-defined limits, along with varicose veins inside that gave it an erythematous violaceous tone, all accompanied by noticeable elongation of that limb (Figures 1 and 2). She later required neurological assessment for instability and vertigo lasting several months. The neurological examination revealed Romberg’s sign with falling toward the right and unstable gait when walking. Imaging revealed descent of the cerebellar tonsils into the foramen magnum consistent with type I Arnold-Chiari malformation (Figure 3).

In neurological disorders, Klippel-Trénaunay syndrome and Arnold-Chiari malformation are rarely found together. We found only 1 published case in the literature reviewed (MEDLINE1966–2007).7 The low prevalence of both processes makes it somewhat unlikely for such an association to be incidental and, therefore, we believe it is important
to investigate the presence of neurological symptoms suggestive of Arnold-Chiari malformation in any patient with cutaneous lesions related to Klippel-Trénaunay syndrome, using magnetic resonance imaging in all these patients.

References