

LETTERS TO THE EDITOR

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



Figure 1. Vascular malformation in the lower limb.

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



Figure 2. Elongation of the affected limb.

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

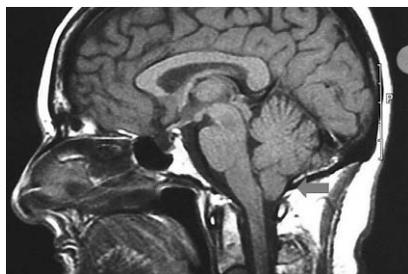


Figure 3. Tonsillar herniation below the foramen magnum.

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.

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Hair Collar Sign Associated with Scalp Aplasia Cutis Congenita

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To the Editor:

The term “hair collar sign” was first introduced into the dermatological lexicon by Commens et al¹ in 1989. The sign consists of a ring of long, dark, thick, and rough hair surrounding a congenital nodule of cystic, blister-like, or atrophic appearance located on the scalp. Histology reveals numerous horizontally oriented hypertrophic hair follicles emerging from the edge of the lesion. Drolet et al² were the first to highlight the importance of the hair collar sign as a marker of spinal dysraphism. They proposed that its formation could be caused by cerebral herniation that would produce, early in embryonic development, abnormal shearing forces during formation of hair follicles, causing them to point outward from the defect. The proximity of the neuroectoderm, which expresses neural cell adhesion molecules, could

also alter normal dermal-epidermal interactions, and as a consequence induce the development of large abnormal follicles.

We present the case of a newborn child from healthy parents. The mother was monitored during her pregnancy, had no adverse events, and had not taken medications. The delivery was vaginal, without the use of instruments. At birth, the child presented a rounded cutaneous defect with a diameter of 1 cm in the left parietal area, near the vertex. The defect was composed of an erythematous, slightly protruding lesion with an edematous appearance and covered by a fine, atrophic, translucent membrane. It was surrounded by abundant thick, dark, rough hairs that were horizontally arranged and oriented toward the periphery of the lesion (Figure 1).

There were no palpable underlying bone abnormalities. The infant also presented a symmetric defect on both hands involving duplication of the thumb, which also presented syndactyly (Figure 2). Ultrasound of the brain and through the fontanelle ruled out abnormalities of the bone or nervous tissue. We finally established



Figure 1. Hypertrophic hair collar surrounding the cutaneous defect.