A Child With Self-Improving Hypotonia: Look at the Skin!

A 9-month-old infant was referred for evaluation of an extensive skin bruise of the hypogastric region that appeared after minor trauma (Figure 1). Abuse-related trauma was suspected; however, findings of the physical examination showed moderate hypotonia with normal reflexes, hyperelastic skin, severe kyphoscoliosis (Figure 2), pectus excavatum, and sternal retractions while breathing.

The history was remarkable for parental consanguinity, severe hypotonia since birth, a right-side clubfoot, and sensorineural hearing loss. During the first months of life, enteral nutrition, due to poor suction capability, was required, and autonomous oral feeding started at the age of 6 months. Muscular tone progressively improved, and at the age of 7 months, the child achieved head control, and at 8 months, he could maintain the sitting position. Previous investigations had ruled out spinal muscular atrophy and Prader–Willi syndrome, whereas creatine-phosphokinase levels and general blood tests were all normal.

Due to the history of easy bruising, self-improving hypotonia, early-onset progressive kyphoscoliosis, and hyperelastic skin, a genetic connective tissue disorder was suspected.

Considering parental inbreeding, a single-nucleotide polymorphism array to detect homozygosity areas was performed. In an analysis of the homozygous areas, the *FKBP14* gene, which encodes the protein family involved in the assembly of the extracellular matrix,[1-3] was sequenced, resulting in a homozygous mutation (c.362dup; p.Glu122Argfs*7) causative of an autosomal-recessive variant of kyphoscoliotic Ehlers–Danlos syndrome.

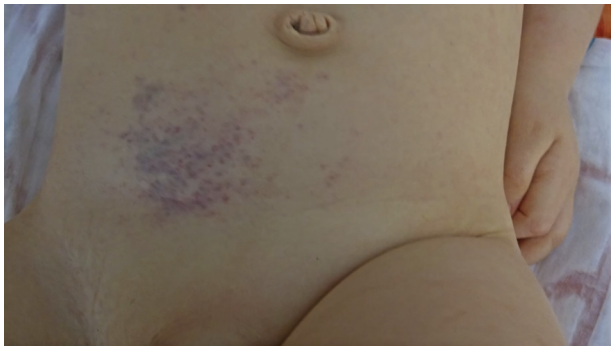

Figure 1. A large bruise of the hypogastric region.

The hallmarks of Ehlers–Danlos syndrome include severe congenital muscle hypotonia at birth, significantly improving during infancy, hyperelastic skin with easy bruising, early-onset progressive kyphoscoliosis, and sensorineural hearing loss.[1-4] In these patients, prompt surgical correction of the kyphoscoliosis is recommended to avoid respiratory failure. In our case, this was successfully performed before the age of 3 years. ■

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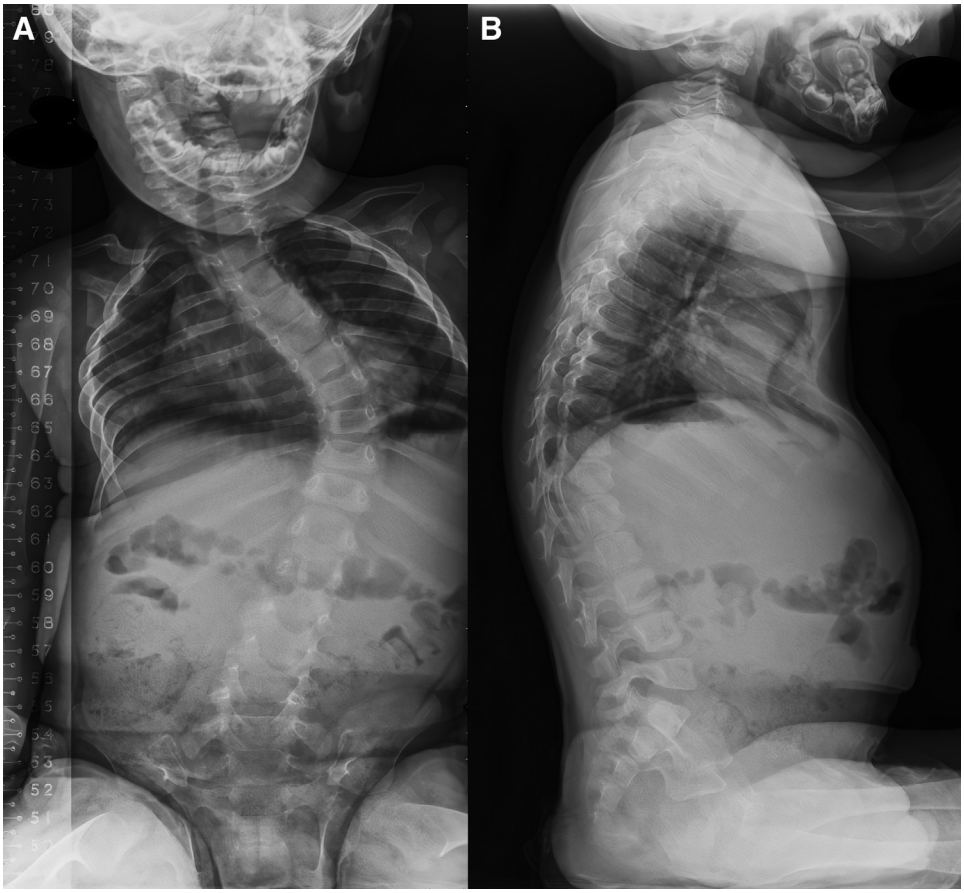

Figure 2. First spinal radiographs performed, at the age of 11 months, showing **A,** severe left dorsal scoliosis and **B,** kyphosis.

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Tick-Tock on the Ward: Essential Palatal Tremor in a Pediatric Patient

A previously healthy 14-year-old girl presented with a 2-week history of tinnitus and continuous movement in the throat. There was no impairment of speech or swallow. Sleep was disturbed due to tinnitus, with a "tick-tock" sound; sensory relieving techniques were not identified. Examination revealed a repetitive rhythmic movement of the uvula and soft palate (levator veli palatini) with an audible click (**Video** and **Figure**). There was inability to voluntarily suppress the movement. Neurologic examination was otherwise normal. Brain magnetic resonance imaging

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