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Visual Diagnosis

Herculean Boy With Facial Myokymia

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This two-year-old boy presented with episodic facial and periorbital twitching along with generalized stiffness since infancy. The twitching episodes were intermittent, of variable duration, and exacerbated by excitement and irritability. Temperature and diurnal variations were absent. On examination, he had short stature, wasting, and microcephaly (<-3 *z* score). His other findings included a Herculean build, long eyelashes, blepharospasm, long philtrum, upturned nose, high arched palate, puckered lips, micrognathia, and triangular face. He also exhibited selective hypertrophy of arm, forearm (Fig 1A), and thigh muscles as well as bilateral knee contractures. He had episodic perioral and periorbital myokymia (Video 1, Fig 1B). A diagnosis of the myogenic variant of Schwartz-Jampel syndrome (SJS) was considered.

Investigations revealed normal serum creatinine kinase levels, thyroid profile, serum sodium, and potassium levels. Skeletal radiographs showed metaphyseal widening of long bones (Fig 2). Needle electromyography revealed normal insertional activity; occasional myokymic discharges and abnormal high frequency decrementing repetitive discharges with pinging sound suggestive of neuromyotonia and polyphasic motor unit action potentials. The next-generation sequencing revealed a heterozygous missense variation in exon 6 of the SCN4A gene (c.952T>T/C) (confirmed by Sanger sequencing) suggestive of sodium channel myotonia. However, his asymptomatic parents were not tested.

The characteristic herculean appearance, myokymia, and metaphyseal widening suggested a physical diagnosis of SJS. However, his genetic testing was suggestive of sodium channel myotonia. His clinical presentation was indicative of "myotonia permanens" with the presence of persistent stiffness, severe myotonia, and muscle hypertrophy. Recently sodium channel myotonia has emerged as an important differential of the SJS phenotype. A Japanese boy with skeletal dysplasia and infantile onset myotonia developed recurrent episodes of paralysis at age seven years, mandating a change in diagnosis from SJS to sodium channelopathy.[1] Muscle sodium channelopathies exhibit various phenotypes, including potassium-aggravated myotonia, congenital myasthenic syndromes, and periodic paralysis.[2,3] The treatment in SJS and sodium channel myotonia is symptomatic with noteworthy response to carbamazepine and antiarrhythmics. Identification of the genotype in a Herculean child with myokymia and skeletal dysplasia is essential because oral potassium or exercise can precipitate acute hypoventilation and life-threatening events in individuals with sodium channel myotonia.

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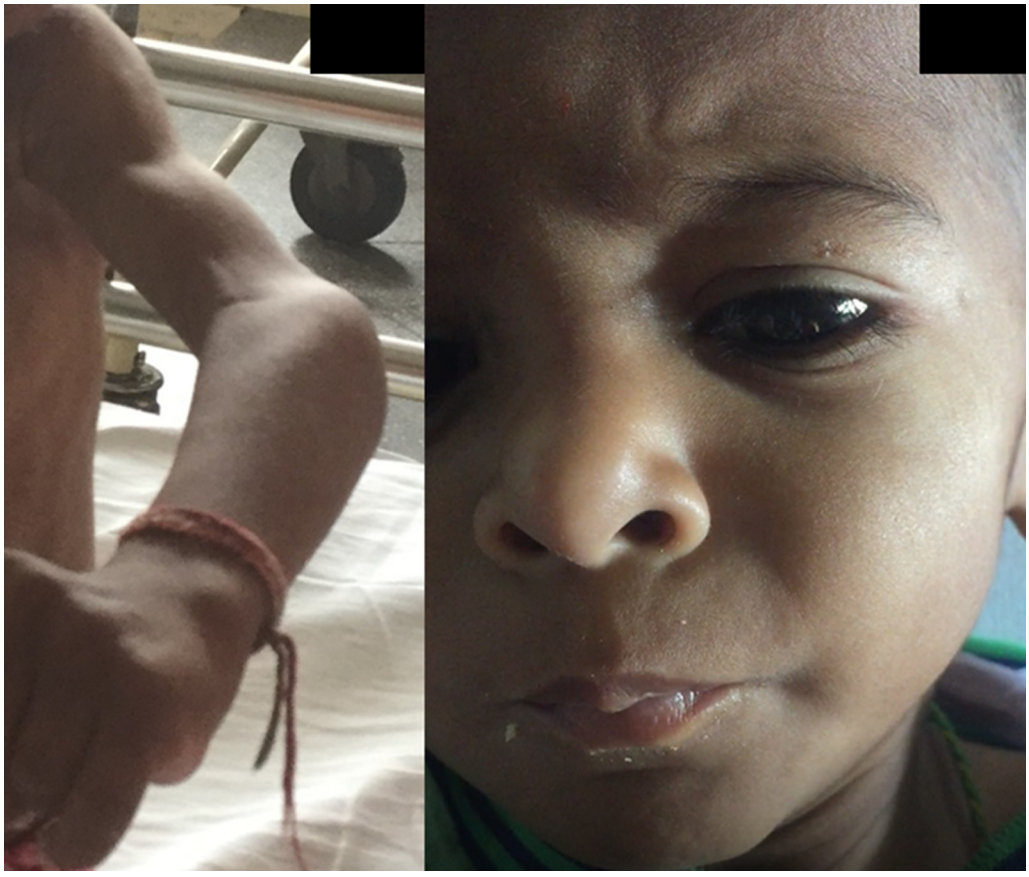

FIGURE 1. Left: Photograph of the left arm illustrates selective hypertrophy of arm and forearm (deltoid, biceps, and brachioradialis muscles). Right: Facial myokymia (the video related to this figure can be found at https://doi.org/10.1016/j.pediatrneurol.2019.02.011).

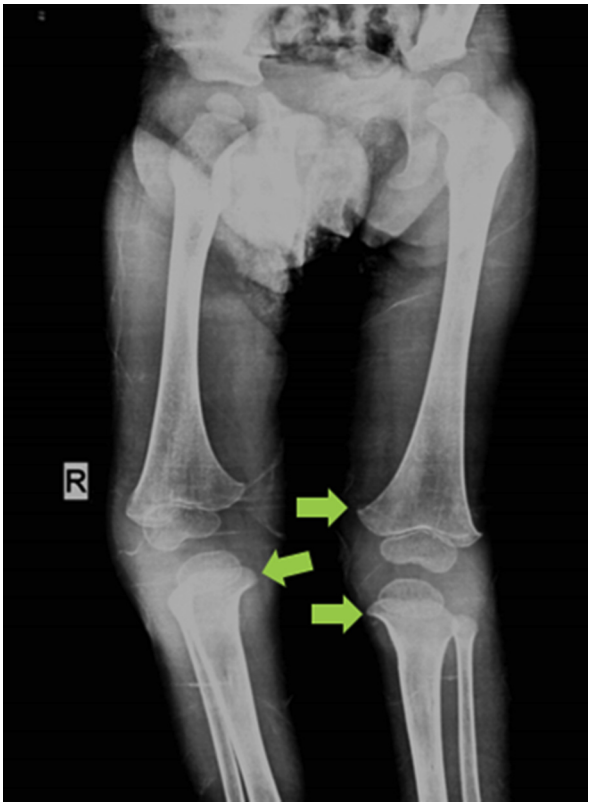

FIGURE 2. Skeletal radiographs showing diffuse osteopenia and metaphyseal widening of long cylindrical bones (arrows).

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