Myotonia in a Child with Muscle Hypertrophy

A 5-year-old child was brought with complaints of loss of appetite and not gaining weight. There was no history of contact with tuberculosis and other systemic illness. Diet history revealed inadequate calorie intake.

On examination, child was alert, active, oriented with normal speech output. His weight (13 kg) was <3rd centile and height (110cm) was at 50th centile. His mid arm circumference was 14.5 cm. Detailed examination of all muscle groups revealed hypertrophy of arm, thigh and leg muscles, with normal muscle power and tone. Upper limb and lower limb reflexes were brisk. Plantar response was flexor. On further examination, percussion myotonia and grip release myotonia of upper limb was present **(*Web Video* 1** and ***Fig.* 1)**. We made a provisional diagnosis of myotonia congenital. He was treated with phenytoin for myotonia, and dietary advice for adequate weight gain.

Myotonia is defined as a disturbance in muscle relaxation after voluntary contraction. Common diseases of children associated with myotonia include Myotonia congenita, Myotonia fluctuans and Neuromyotonia. In these disorders, weakness is not prominent but stiffness may impair muscle function. Abnormalities in the potassium and chloride channels underlie most cases.

FIG. 1 *Elicitation of myotonia in a child.*

Myotonia associated with muscle hypertrophy is characteristic of Myotonia congenital (Becker's type). EMG establishes the diagnosis. Repetitive discharges at rates of 20 to 80 cycles per second are recorded when needle is inserted into the muscle. The waxing and waning of the amplitude and frequency of potentials produce a characteristic sound. Mexilitine and Phenytoin is used for reducing muscle stiffness in myotonia. This case scenario explains the need of eliciting myotonia in children with muscle hypertrophy to differentiate from muscular dystrophies, as children with myotonia with muscle hypertrophy are easily treatable.

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