Myotonia Congenita (Becker’s Variant)


A 21 year old right handed male resident of Bihar presented with 4 years history of gradual onset progressive stiffness in both thighs and calves which improves on continued activity along with hypertrophy of muscles of all the limbs and dimpling on hitting over thigh and hands. He also has history of inability to walk fast or run and delayed opening of left eye after forceful closure of both eyes for past 8 months.

His general examination was normal. On neurological examination he was conscious, oriented with normal higher mental functions and cranial nerves examination. He also had slurred speech. His motor system examination revealed selective hypertrophy of bilateral biceps (Figure 2), latissimus dorsi, forearm muscles (Figure 2), quadriceps (Figure 1), gastrocnemius (Figure 1) and extensor digitorum brevis, tone and power was normal in all the muscles groups. His muscle stretch reflexes were normal and plantars were bilateral flexors. Clinical myotonia and percussion myotonia were present over thighs (Figure 3), thenar eminence (Figure 4) and tongue. No cerebellar, bowel and bladder dysfunctions were noted.

Myotonia congenita is an inherited voltage gated chloride channelopathy, can be either autosomal dominant (Thomsen's disease) or autosomal recessive (Becker's disease). In the recessive variant age of onset is 10-14 years, is more severe, characterised by muscles hypertrophy more prominent in legs and myotonia with warm up phenomenon which appears abruptly after rest. Myotonia responds to sodium channel blockers.

References


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