A case report of neuromyotonia (Isaacs syndrome)

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Clinical presentation
A 58-year-old man presented with numbness and muscle twitching in the lower limbs since 2.5 months and muscle twitching in the upper limbs since 2 days. In early April 2015, the patient noticed numbness and coolness in the toes of both his feet without any obvious cause, and occasional muscle twitching in the left calf. He developed pain in both calves and twitching in the left thigh; and the muscle twitching gradually worsened in May. On June 9, he experienced muscle twitching in the upper extremities. He had a normal diet with increased sweating, chills, poor sleep, and a weight loss of 15 kg in 2 months.

Physical examination: The patient had increased sweating in the facial and neck regions. There was atrophy in the muscles in the extremities and the shoulder girdle muscles. The muscle strength in the upper and lower extremities was weak (grade 4), and the muscle tension was reduced. The biceps jerk, triceps jerk, knee jerk, and ankle jerk reflexes were diminished, the Babinski sign was negative. The patient had pain in the lower extremities; Visible muscle cramps were present in the biceps, deltoid, both gastrocnemius muscles, and both quadriceps (Supplementary video 1).

Laboratory and neuroradiological examination: He had significantly increased serum creatine kinase (CK), 1783 U/L (range 24-200 U/L). The squamous cell carcinoma (SCC) antigen, 8.7 ng/mL (range 0-1.5 ng/mL) was slightly elevated. Nodules and lymph node enlargement were found on lung CT.

Electrophysiological examination: EMG revealed myokymic and neuromyotonic potentials (Supplementary video 2).

Treatment
The patient was diagnosed with Isaacs syndrome and administered oral carbamazepine, 0.2 g twice a day. He
responded well and the muscle cramps almost vanished after 8 week’s medication (Supplementary video 1). He was asked to undergo regular review of tumor markers during follow-up.

Discussion

Isaacs syndrome is a rare disease characterized by hyperexcitability of the peripheral motor nerves. Its main clinical manifestations include muscle cramps, increased sweating, and occasionally muscle weakness [1], with possibly increased concentrations of serum CK. Currently, it is believed that Isaacs syndrome is a result of anti-voltage–gated potassium channel (VGKC) antibody–induced autoimmunity [2,3].

The electrophysiological markers for Isaacs syndrome include myokymic and neuromyotonic potentials. Myokymic potentials usually consist of 2–10 motor unit action potentials and discharge at a frequency of 20–150 Hz. Muscles discharging myokymic potentials typically present worm-like movements on physical examination. Neuromyotonic discharges are similar to myokymic discharges, but present a higher discharge frequency (150–300 Hz) [4].

Our patient underwent many medical procedures, and 2 out of 3 times, myokymic and neuromyotonic potentials were not reported on electrophysiological examination, suggesting that the treating neurologists were unfamiliar with these discharges. This case report includes videos of muscle cramps before and after treatment, videos of EMG, in an effort to provide a reference for clinicians encountering this rare disease in their clinical practice.

References


Video legends (Supplementary files)

Video 1: Muscle twitching and cramps.
Video 2: EMG.