CASE REPORT

"Isaacs Syndrome A Rare Case Report"

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Abstract:
Isaac's syndrome also known as neuromyotonia is a rare disorder characterised by spontaneous, continuous muscle activity of peripheral nerve origin. The clinical findings seen are muscle twitching, cramps, muscle stiffness, muscle hypertrophy, increased sweating. The main oral cavity related complaint is related to decreased mouth opening. Electromyography shows spontaneous, continuous, irregularly occurring doublet, triplet or multiplet single motor unit discharges, firing at a high intraburst frequency along with fasciculations and fibrillation potentials. Elevated antibody levels against voltage gated potassium channels are also seen in 40% cases. There is no known cure for this syndrome however relief from muscle stiffness and spasms are seen with the administration of anticonvulsants like phenytoin and carbamazepine.

Key Words: Isaacs’ syndrome, Continuous muscle fibre activity, decreased oral opening, anticonvulsants.

Case Report
A ten year old female patient came with a chief complaint of limited mouth opening and muscular stiffness since four to five years. On general examination, facial grimacing along with twitching of tongue muscles and constriction of facial muscles was seen.

Figure 1: showing facial grimacing
Figure 2: Constriction of facial muscle

Due to the stiffness of the jaw musculature, intra oral examination was not successful. Due to this an examination of the tongue for stiffness of the tongue musculature could not be done. Behavioural changes namely anxiety, restlessness and sleep disturbance was reported by the patient. An EMG carried out of right bicep muscle (Fig 3) and
right quadricep muscle (Fig 4) at rest showed continuous and spontaneous muscle activity suggestive of Neuromyotonia Continua.

**Fig 3:** EMG of Right Bicep muscle showing continuous muscle activity

**Fig 4:** EMG of Right Quadricep muscle showing continuous muscle activity

**Discussion**

Isaacs syndrome also known as acquired neuromyotonia first described by Issacs in the year 1961 is a rare disorder characterised by hyperexcitability of peripheral motor nerves leading to muscle twitching, cramps, myokymia, pseudomyokymia (slow muscle relaxation after forceful contraction) and mild weakness (1). Excessive sweating and weight loss are also seen associated with this disorder (2) along with behavioral changes like anxiety, restlessness, hallucinations and sleep disturbances have been reported. (3) Continuous spontaneous discharges in doublets, triplets or multiplets with high intraburst frequency now known as “neuromyotonic discharges” is seen in Electromyography. (4)

An autoimmune etiology is postulated because of the association of Isaacs syndrome with thymoma, myasthenia gravis, vitiligo, Hashimoto's thyroiditis and penicillamine treatment also spontaneous remission has been observed which is a consistent finding seen with autoimmunity. In addition to this, neuromyotonia has also been reported in patients with lung cancer, which raises the possibility that tumour antigenic determinants are perhaps capable of triggering an autoimmune response producing antibodies which cross-react with neuronal voltage-gated ion channels. Demonstration of a significant reduction in the number of neuromyotonic discharges recorded by EMG after plasma exchange was also seen as the first direct evidence which showed an autoimmune etiology along with increased nerve terminal excitability postulated that voltage-gated potassium channels (VGKCs) were the primary target for the pathogenic antibodies. (5)

Diagnosis is based on the clinical findings and EMG findings. The most commonly seen clinical findings are myokymia, pseudomyotonia and stiffness of trunk and limbs without severe pain which is more pronounced in the distal rather than proximal muscles. Abnormal muscle activity persists during sleep and dyspnea
may occur if respiratory muscles are involved. Weight loss and excessive sweating are the associated symptoms observed. (1, 2)

Oral findings include stiffness of jaw and tongue leading to difficulty in swallowing turning the voice hoarse along with facial muscle twitching which is seen in about a fourth of the cases. (1, 2, 6) In the patient observed in our case, any attempt at oral examination was extremely difficult given the limitation of mouth opening.

Autoimmune diseases such as chronic inflammatory demyelinating polyneuropathy, myasthenia gravis or the presence of antiacetylcholine receptor antibodies maybe associated with this syndrome (7) along with hematologic malignancies such as thymoma, (8) plasmacytoma, (9) Hodgkin’s lymphoma (10) and bronchogenic carcinoma paraneoplastic syndromes (11). Isaac’s syndrome is often treated with the help of antiepileptic drugs or immunotherapy which often improves the clinical and electrophysiologic findings. (12) Carbamazepine, phenytoin, lamotrigine and sodium valproate can be used in combination or as a single drug whereas Paraneoplastic neuromyotonia often improves after treatment of the underlying cancer. (13) In patients with debilitating symptoms or those refractory to symptomatic therapy, immunomodulatory treatment should be tried. (14, 15) Good trials of long-term oral immunosuppression are not available. However, in selected patients, prednisolone, with or without azathioprine or methotrexate, has proved to be useful. (16)

References


