Case Report

Satoyoshi syndrome

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Satoyoshi syndrome (Komuragaeri disease) is a rare disorder of presumed autoimmune etiology, characterized by painful muscle spasms, alopecia, diarrhea, endocrinopathy with amenorrhea and secondary skeletal abnormalities. Most of the previous reports are of the Japanese people. We report the first case from India.

Key Words: Satoyoshi syndrome, Komuragaeri disease.

Introduction

Satoyoshi syndrome is a rare syndrome first described by Satoyoshi and Yamada\(^1\) in 1967. So far, less than 40 cases have been reported worldwide.\(^2-8\) It is considered to be a sporadic disease of presumed autoimmune etiology. We report a typical clinical case of Satoyoshi syndrome in a 20-year-old woman, because of its rarity and lack of earlier descriptions from India.

Case Report

A 20-year-old woman was referred for botulinum toxin treatment for jaw dystonia. She was born of a non-consanguineous union and had normal birth and development till 10 years of age. Her initial symptom was failure to gain height or weight in spite of adequate food intake. She had recurrent episodes of watery and frothy diarrhoea after high carbohydrate meals and sweet food items. She then noticed patchy hair loss in the eyebrows and eyelashes, and later progressive baldness. She did not attain menarche or develop secondary sexual characteristics.

At 15 years of age, she developed painful spasms in the calf muscles of both legs. Later, it involved the arms, fingers, neck and the jaw muscles. The muscle spasms were painful and the muscles became board-like during these attacks. The spasms lasted for 1-2 minutes and restricted the use of the limbs. The jaw spasms occurred while talking and eating and led to frequent jaw closure and inability to talk or eat until it abated. She also developed bowing of legs and pain in the knee joints. This was treated by corrective bilateral orthotic surgeries. Two years later, she developed involuntary, sudden, brief shock-like jerks of her upper limbs 5-6 times per day. She received various treatments including parenteral and oral calcium, vitamin D3, cyclical hormonal therapy for amenorrhoea, minoxidil for hair loss, clonazepam, tetrabenazine, baclofen, and diazepam for muscle spasms, all without any improvement.

On examination, we found a short-statured lady, 142 cm in height and weighing 38 kg, who had alopecia totalis and no secondary sexual characteristics. She had fixed bony deformities at both knees. Her masseters, biceps and brachioradialis muscles were hypertrophied. Frequent painful spasms of the masseter, deltoid, biceps, index finger and thumb lasting 1-2 minutes were observed. She also had spontaneous multifocal myoclonic jerks. Her neurological examination was otherwise normal.

Laboratory examination showed hypochromic, microcytic anemia. A bone marrow examination revealed absent iron stores. Serum calcium, phosphorus and creatine phosphokinase (CPK) were normal. Her fasting blood sugar was 68 mg%. A glucose tolerance test was attempted but could not be completed, as she was intolerant to oral glucose. Serum lactate (pre and post-exercise) was normal. Anti-nuclear antibody, antiphospholipid antibody, C-reactive protein and rheumatoid factor were negative. Serum cortisol, follicle stimulating hormone, leutinising hormone, growth hormone, thyroid hormones and parathormone were normal. Ultrasonography abdomen revealed a hypoplastic uterus. Her bone biopsy, upper gastrointestinal endoscopy and biopsies, CT abdomen and CT brain were normal. Chromosomal analysis showed a 46XX karyotype. A skeletal survey revealed thinning of long bones, osteotomised genu varus deformity, multiple cysts and crack fractures.

Nerve conduction study was normal. Electromyography showed no spontaneous discharge at rest, normal motor unit potential and normal interference pattern. During the spasms, continuous motor unit potentials of 4-10 mV amplitude at a frequency of 40-50/sec were seen (Figure 1). Spasms could be induced by single and repetitive nerve stimulation.

She was started on a combination of tablet phenytoin 200 mg daily and prednisolone 40 mg per day. Her muscle spasms including jaw spasms and myoclonic jerks stopped within 3 days of treatment. At the last follow up, two years after treatment, she had new hair growth over the eyebrows and scalp and regular menstrual cycles and she remained free of spasms.

Discussion

Satoyoshi syndrome was first reported in 1967 by Eijiro Satoyoshi and Kaneo Yamada, in Tokyo.\(^1\) In the next 10 years,
Satoyoshi collected an additional 15 cases with autopsy studies in 2. The Japanese also call this disorder Komuragaeri disease (Komura=calf, gaeri=turnover or spasm). The majority of the reported cases were in the Asian population though non-Asian cases were reported from Britain, Russia, Argentina and one in a European American.

Satoyoshi syndrome is a sporadic disease with a mean age of onset of 10 years (ranges from 6 to 15 years). It is commoner in females. The usual initial symptoms are muscle spasms in the legs and alopecia. The spasms are painful and progressive and their frequency varies from 1 or 2 to 100 per day, each lasting a few minutes. It can be sufficiently severe to produce abnormal posturing of the affected limbs, particularly the thumbs. With progression the illness involves the pectoral girdle and trunk muscles and finally the masseters and temporalis muscles. The spasms usually spare the facial muscles. Severe spasms can interfere with respiration and speech. During an attack-free period, non-stimulus-sensitive myoclonus can occur in the arms, legs and neck. Diarrhea occurs in the first 2-3 years with intolerance to carbohydrate and high glucose diets. Endocrinopathy manifests as amenorrhea and hypoplasia of the uterus. Affected children fail to attain height after 10-12 years of age. They develop bony deformities like genu varus/valgus, lumbar lordosis and pes planus. Skeletal survey may show slipping of multiple epiphysis, cystic lesions, acet-o-osteolysis, and bone fragmentation at tendinous insertions, fatigue fractures and early osteoarthrosis. The disease is progressive and in Satoyoshi’s series, 4 patients died, 2 due to malnourishment and the others due to respiratory distress.

The disease is thought to have an auto immune basis because of the improvement with steroids, its association with other autoimmune diseases, deposition of immune complexes in the muscles and ANA positivity in some cases. Intravenous calcium gluconate, dantrolene sodium, quinine, procainamide and phenytoin have been used to treat the muscle spasms. Botulinum toxin is also useful in treating severe spasms. Frequent pulse therapy with intravenous immune globulin has also been found to be effective and it may be a safer alternative to long-term steroids in young patients.

We report this case to draw attention to this rare disease with multiple system involvement, which may hamper its early detection. The disease may be mistaken for masticatory dystonia, malabsorption syndrome, systemic vasculitis or an orthopedic disorder unless the index of suspicion is high. The clinical manifestations in our case did not differ from reports from other parts of the world.

References


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