Case Report

Satoyoshi Syndrome: Difficult to find or treat?

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ABSTRACT

Satoyoshi Syndrome, also known as Komuragaeri disease, is a rare multisystemic autoimmune disorder characterised by alopecia, intermittent painful muscle spasms, diarrhoea, endocrinopathies, skeletal deformities, and positivity for antinuclear antibodies (ANA). The etiology of this condition remains unclear. This case report discusses Satoyoshi Syndrome in a 10-year-old girl, emphasising the diagnostic challenge, clinical course, treatments, and outcomes.

Key words: Satoyoshi syndrome, a multisystemic, autoimmune disorder, autoantibodies, alopecia, and muscle spasm

Satoyoshi syndrome was first described in 1967 by Eijiro Satoyoshi and Kaneo Yamada, who reported two cases with painful, intermittent muscle spasms of a slowly progressing nature, beginning in early life. It is commonly observed in the Japanese population, where its colloquial name is Komuragaeri disease *(komura* implying calf and *gaeri* implying spasm) [1]. Thereafter, fifteen more cases were reported with the same disease [2]. Most cases described were sporadic. It is a rare multisystem disease usually presenting with progressive painful muscle spasms, diarrhoea, endocrinopathy, alopecia, and skeletal abnormalities. An autoimmune basis is likely, supported by associations with other autoimmune conditions, the presence of autoantibodies, and successful treatment of symptoms with immunosuppressants [3].

Approximately 80 cases have been reported worldwide, of which about 33 (40%) are of Asian origin (Japan, India, Thailand, China) [2]. Rudnicka et al. [4] suggested diagnostic criteria that included diffuse hair loss or alopecia (an obligatory criterion) with intermittent painful muscle spasms, diarrhoea, and positive antinuclear antibody (ANA). Antinuclear antibodies are present in 60% of patients. The condition occurs more commonly in females, with a mean age of onset omit of around ten years (range 6 to 15 years). No established genetic pattern has yet been described [4]. The characteristic painful intermittent muscle spasms are progressive, frequently severe enough to cause abnormal posturing of the limbs, and lasting several minutes. It may progress to involve the limb-girdle muscles, temporalis, and masseters and rarely interfere with speech and respiration. The diarrhoea may lead to carbohydrate malabsorption. The endocrinopathy usually manifests as amenorrhea or as a hypoplastic uterus [5].

If treatment is not initiated in time, it may result in severe disability or death. Since the description of the syndrome, multiple therapeutic strategies have been implemented. The mainstay of treatment of Satoyoshi syndrome is the administration of corticosteroids. It has been the main treatment that has allowed an improvement in the prognosis of this disease. Others include muscle relaxants and antiepileptics to relieve muscular spasms that can be very troublesome and cause significant disability. Corticosteroids or other immunosuppressive therapies, such as ciclosporin, mycophenolate mofetil, azathioprine, methotrexate, tacrolimus, cyclobenzaprine and high doses of IVIG, have been found to improve muscular spasms as well as diarrhoea and alopecia [6]. We are presenting an interesting case report including clinical features, management and outcome of Satoyoshi syndrome in an early adolescent girl.

CASE REPORT

A 10-year-old girl, born to non-consanguineous parents from West Bengal, India, presented with progressive hair loss over 3 years and recurrent painful muscle spasms in the thigh and neck for 7 months. The hair loss started with eyebrows, eyelashes, and other body parts in 2019, followed by scalp hair loss since March 2021. The painful, recurrent muscle spasms also developed around the knees, along with abdominal spasms involving the rectus abdominis, simulating a visible, ill-defined

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swelling over the abdomen. The muscle spasms affected daily movements and led to limping, and she also experienced weight loss. Perinatal history and development were normal, and there was no history suggestive of neuroregression. Her school performance was average, although the severe muscle cramps caused frequent absenteeism.

She weighed 26 kg (at 4th percentile), her height was 127 cm (below 3rd percentile), and her head circumference was 48 cm (below 3rd percentile). No secondary sexual characteristics had appeared. There was generalised wasting, pallor and a few thin, sparse hairs over the scalp region (Figure 1a). Muscle tone was normal; grade 5 power in all muscles, and no muscle tenderness (except for spasm episodes) and wasting were noted. Tendon jerks were normal bilaterally in both upper and lower limbs, and there was no evidence of any neurodeficit. Other systemic examinations were within normal limits. No significant skeletal abnormalities were observed. Laboratory evaluation revealed microcytic hypochromic anaemia, normal blood count, and liver and kidney function tests. Serum calcium, phosphate, and alkaline phosphatase were normal, but Creatine phosphokinase (CPK) was marginally high (246 U/L). Endocrine evaluation was within normal limits, including thyroid function tests, parathormone, and vitamin D.

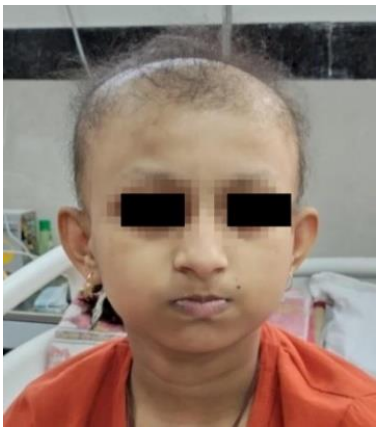

Figure 1a: Only a few thin, sparse hairs over the scalp region

Anti-TPO (Anti-Thyroid Peroxidase Antibodies) levels were high (3.87 IU/ml). Blood sugar and ANA levels were normal. Urine screening for abnormal metabolites and tandem mass spectroscopy (TMS) for Inborn errors of metabolism were negative. Chest X-ray and ultrasound abdomen were normal. Skeletal survey revealed multiple defects: (a) X-ray spine (Figure 1b) showed spina bifida of L5 & S1, and (b) X-ray bilateral knee & lower legs show metaphyseal sclerosis involving bilateral distal femur (Right>Left) and fracture involving medial cortex of fibula on left side with callus formation (Greenstick fracture) (Figure 1c). She was treated with pulse methylprednisolone (30mg/Kg/day) for five consecutive days. We initially started her on oral Prednisolone omit one @ @ 2 mg/kg/day and followed by tablet deflazacort @ 0.9 mg/kg/day. She is also on oral Baclofen, Diazepam, Calcium, and Vitamin D supplements.

For alopecia, she received topical 2% Minoxidil and betamethasone lotion. These medications are low-cost and comparatively have fewer side effects. There has been a significant improvement in her hair growth, and no behavioural abnormalities are currently observed. She was on regular follow-up and was spasm-free. However, a few days ago, she came with a recurrence of painful spasm after a symptom-free interval of 2 years. History revealed poor compliance with medications for the past few months. She is now on 2mg/kg/day oral Prednisolone, following which her symptoms showed marked improvement. She has reasonable hair growth over the scalp. There is no steroid side effect so far.

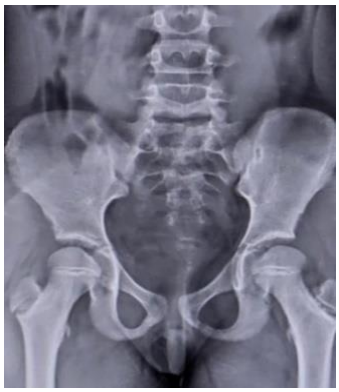

Figure 1b: X-ray whole spine shows failure of midline fusion of the spinous process of L5 & S1, suggestive of spina bifida

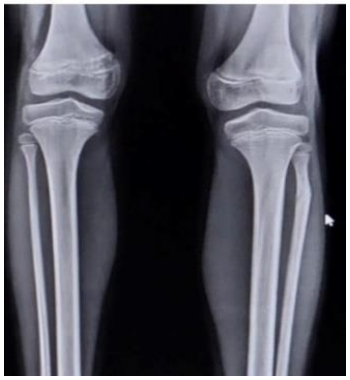

Figure 1c: X-ray bilateral (B/L) knee & lower legs show metaphyseal sclerosis involving B/L distal femur (Right>Left) and fracture involving the medial cortex of the fibula on the left side with callus formation (Greenstick fracture)

DISCUSSION

Satoyoshi syndrome is a rare, sporadic disease [1]. Although a few consider the disease to have a neurogenic origin, most authors believe Satoyoshi syndrome to have an autoimmune pathogenesis, based on ANA positivity in the majority of cases, coexisting autoimmune diseases, and responsiveness to corticosteroids and immunosuppressants. Severe alopecia (alopecia totalis or universalis) is more commonly seen in

ANA-positive patients. In contrast, ANA-negative patients present more commonly with partial hair loss [4, 7], as our patient had few sparse hairs on the scalp. In a systematic review, ANA positivity was reported in 58.3% of 77 cases [2]. Incidental skeletal abnormalities, possibly secondary to painful muscle spasms, are reportedly more common in ANA-negative than in ANA-positive patients [5, 7].

Other common associations of Satoyoshi syndrome include diarrhoea, malabsorption, growth retardation, and amenorrhea. Elevated Creatine phosphokinase (CPK) levels have been reported in 23% of patients, while anaemia and other metabolic abnormalities are reported occasionally [8, 9]. The anti-TPO antibody positivity in this child may be due to autoimmune thyroiditis, which has been reported previously [3]. Corticosteroids remain the mainstay of treatment, with adjuvants including cyclosporine, phenytoin, diazepam, dantrolene sodium, and plasmapheresis also reported to be effective in the reduction of diarrhoea and neuromuscular symptoms [9, 10].

A review article indicates that corticosteroid treatment significantly improves prognosis and reduces mortality compared to earlier reports. However, the optimal dose, duration, and timing of adding other immunosuppressants are still unclear, as these agents are mostly used alongside corticosteroids to minimise side effects. Anticonvulsants and muscle relaxants have shown little benefit, with only dantrolene partially being effective for muscle symptoms. Management also includes supportive therapies, but due to limited case-based evidence and lack of long-term follow-up, standardised treatment guidelines are yet to be established [6]. However, only about 50% of ANA-negative patients responded to this line of treatment. Refractory spasms may be treated with botulinum toxin [11]. In patients with severe side effects of long-term glucocorticoid use, a safer alternative is frequent pulse therapy with intravenous immune globulin (IVIG) [12].

CONCLUSION

Satoyoshi Syndrome, though rare, should be thoroughly investigated when clinical suspicion arises. The mainstay of treatment for Satoyoshi Syndrome is the administration of corticosteroids. The use of corticosteroids and immunosuppressants significantly improves prognosis. Early recognition and prompt management are key to improving patients' quality of life and long-term outcomes.

As Satoyoshi Syndrome is a complex and multisystemic disease, patient management must be individualised according to clinical manifestations, and a multidisciplinary team involving paediatricians, neurologists, dermatologists, and endocrinologists is essential for optimal care. Given the rarity of the condition, further research is needed to elucidate the underlying immunopathology and to develop standardised diagnostic and treatment protocols. Collaborative case reporting and long-term follow-up studies are needed to establish more effective strategies. Ultimately, understanding Satoyoshi syndrome not only offers the potential to improve outcomes for affected individuals but also sheds light on broader mechanisms of autoimmunity and neuromuscular dysfunction.

Declaration of patient consent

The authors certify that they have obtained all appropriate consent and assent for images and other clinical information to be reported in the journal. The patient and parents understand that identity will not be disclosed, but anonymity cannot be guaranteed.

Authors' Contributions

AM extracted the data and drafted the manuscript. AD, RB drafted the manuscript. NB, SS, RS, NM revised it critically for important intellectual content. All authors approved the final version. NM will act as guarantor for this manuscript.

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