with these features.

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Satoyoshi Syndrome

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Satoyoshi syndrome was first described by Satoyoshi and Yamada in 1967. It is common in Japanese population where it’s colloquial name is komura-gaeri disease (komura implying calf and gaeri implying spasm) [1]. Our patient presented with the classical features of alopecia totalis, painful muscle spasms and skeletal anomalies but did not show any evidence of endocrinopathy.

Key words : Alopecia, India, Muscle spasm, Skeletal deformity.

CASE REPORT

This 11 year old girl, presented with progressively increasing alopecia for last 5 years and painful recurrent muscle spasms for last 3 months. These intense, painful muscle spasms often occurred in the thigh, neck, around the knees, and occasionally jaw spasms, and abdominal spasms simulating a visible ill-defined swelling over
abdomen. Loss of hair over scalp, eyebrows (Fig. 1) and general body surface was alarming to the parents. In addition, there was weight loss and deformity of knees and lower limb resulting in movement restriction and limping. Perinatal history and development were normal and there was no history suggestive of neuroregression. School performance was average, although the severe muscle cramps caused frequent absenteeism.

At 11 years, she weighed 18 kg (below 3rd percentile), height was 131 cm (at the 5th percentile), and head circumference was normal. No secondary sexual characteristics had appeared. There was total alopecia, generalized wasting, pallor, bowing of lower limbs with genu valgum and pes planus. There was normal muscle tone and grade 5 power in all muscles and no muscle tenderness (except for episodes of spasm). Jerks were normal bilaterally in both upper and lower limbs and there was no evidence of any neurodeficit. Other systemic examination also did not reveal any abnormality.

Laboratory evaluation revealed microcytic hypochromic anemia and a normal blood count, liver and kidney function tests. Bone marrow examination showed decreased iron stores. Chest X-ray and USG were normal. Serum calcium, phosphate, alkaline phosphate, CPK were normal. Endocrinological evaluation including thyroid function tests, parathormone, growth hormone, follicle stimulating hormone, leutenizing hormone were within normal limits. Blood sugar and ANA levels were normal. Nerve conduction studies and electromyography done during the spasm free periods were normal with no spontaneous discharge at rest, normal motor unit potential and normal interference pattern.

Skeletal survey revealed multiple defects: (i) narrowing of ends of clavicle and terminal phalanges suggestive of acroosteolysis (ii) irregular sclerotic distal femoral metaphyses (Fig. 2), (iii) left sided genu varus, right sided genu valgum; (iv) and delayed bone age.

The child was treated with oral phenytoin (50 mg twice daily), prednisolone (20 mg twice daily), Vitamin D and calcium supplements. On follow up after 3 weeks, her spasms had significantly improved but alopecia did not resolve. She is now spasm free but on follow up and under consideration for methotrexate therapy.

**DISCUSSION**

This multisystem disease occurs more commonly in females, with mean age of onset around 10 years (range 6 to 15 years). Etiology is unknown and there is no established genetic pattern yet described [2]. It is speculated to be a sporadic disease of autoimmune origin [3, 4]. The autoimmune basis of the disease is because of the improvement with steroids, its association with other autoimmune diseases, deposition of immune complexes in the muscles, and in few cases, a positive anti-nuclear antibody [3, 5]. Studies have also demonstrated antibodies against brain and gastrointestinal tissue [6].

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 and also the temporalis and masseters and rarely may interfere with speech and respiration. The diarrhea may lead to carbohydrate malabsorption. The endocrinopathy usually manifests as amenorrhea or as hypoplastic uterus [3].

**FIG. 1** Face showing complete loss of hair from scalp and eyebrows.

**FIG. 2** Irregular sclerotic distal femoral metaphyses.
The unique feature of Satayoshi’s syndrome are the myriad skeletal abnormalities presumed to be due to recurrent vigorous muscle spasms causing repeated injuries to the growth plates, epiphyses, and tendon attachments in the growing skeleton [7]. Severe muscle spasms may respond to intravenous calcium gluconate, dantrolene sodium, quinine, procainamide and phenytoin [8]. Refractory spasms may be treated with botulinum toxin [9]. In those patients with severe side effects to long term glucocorticoids, a safer alternative is frequent pulse therapy with intravenous immune globulin [10].

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Wiedemann-Rautenstauch Syndrome

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Wiedemann-Rautenstauch Syndrome is a rare autosomal recessive neonatal progeroid syndrome with only few published case reports. We describe a neonate showing clinical features of WR syndrome with peeling of skin, and presented with weak cry and breathing difficulty since birth.

Key words: Neonate, Progeria, Wiedemann-Rautenstauch syndrome.

Wiedemann-Rautenstauch Syndrome (WR) syndrome is a known neonatal progeroid syndrome comprising of generalized lipoatrophy except for fat pads in the supraboruttock areas, hypotrichosis of the scalp hair, eyebrows and eyelashes, relative macrocephaly and macroglosia [1]. Till date, total 34 cases have been reported and none from India. [2-9].

CASE REPORT

This newborn infant, delivered in a district hospital, was admitted with complaints of weak cry and breathing difficulty since birth. She was the first daughter of healthy non-consanguineous 23-year-old mother and 27-year-old father. Delivery was normal at 36 weeks of gestation and birthweight was 1.5 kg, length 43 cm and occipito-frontal...