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].

Contributors: DM: data compilation and manuscript drafting; AG: guidance for manuscript writing and design; MM: guidance for manuscript writing and design.
Funding : None.
Competing interests: None stated.

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Wiedemann-Rautenstauch Syndrome

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Received: January 22, 2010;
Initial review: March 25, 2010;
Accepted: June 28, 2010.

Wiedemann-Rautenstauch (WR) 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-Rantenstrauch syndrome.*

Wiedemann-Rautenstauch (WR) syndrome is a known neonatal progeroid syndrome comprising of generalized lipoatrophy except for fat pads in the suprabuttock 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

head circumference was 34 cm. There was no history of birth of similar children in family and in close relatives. No significant antenatal history was present and baby died on third day of an undetermined cause. Physical examination at the time of admission showed apparent growth retarded baby with macrocephaly with frontal and biparietal bossing, craniofacial disproportion, almost total alopecia, large fontanels and wide sutures, prominent scalp veins, hypoplasia of facial bones, small nose, upward slanting palpebral fissures, hyperteleorism, ocular proptosis, sparse eyebrows and eyelashes, low set and small ears with normal configuration, down turned angle of mouth, long filtrum, high arched palate, and a sharp and pointed chin (***Fig.*1**).

The neck was short with redundant skinfolds, nipples wide spaced and there was no cardiac murmur and air entry was bilaterally equal and normal. Abdomen was slightly distended. Liver and spleen were palpable. The external genitalia were of a normal female. There was generalized deficient subcutaneous fat, with the exception of excessive fat on the buttocks. The skin was thin, shiny, erythematous and there was peeling of skin. Fingers and toenails were normal. She was hypertonic. A complete blood count showed Hb 15.6 g/dL, TLC 9800, DLC N-65%, L-28%, blood sugar, calcium were normal, X–ray showed bilateral infiltration, and USG cranium was normal. Karyotyping was not sent because parents refused for it. Above clinical findings confirmed a clinical diagnosis of typical WR progeroid syndrome.

DISCUSSION

The clinical features of our case are similar to those described earlier [2-9]. Patients with this syndrome can be recognized at birth because of distinct clinical features that include short stature, failure to thrive, progeroid appearance, apparent macrocephaly with frontal and parietal bossing, wide fontanels and sutures, prominent scalp veins, hypoplasia of facial bones, sparse scalp hair, eyebrows and eyelashes, and generalized lipoatrophy. Most patients showed neonatal teeth, which were lost early [6]. These patients also have endocrine abnormalities such as hypertriglyceridemia, hyper-cholesterolemia and hyperinsulinemia but not required for diagnostic purpose [3]. Majority of these patients die during the first few days or months after birth [6]. At present, there is no treatment.

Contributors: JP and GP: patient history taking and management, and review of iterature. SN: review of literature and manuscript writing. PG supervised the management and drafted the manuscript. JP shall act as a guarantor.
Funding: None.
Competing interests: None stated.

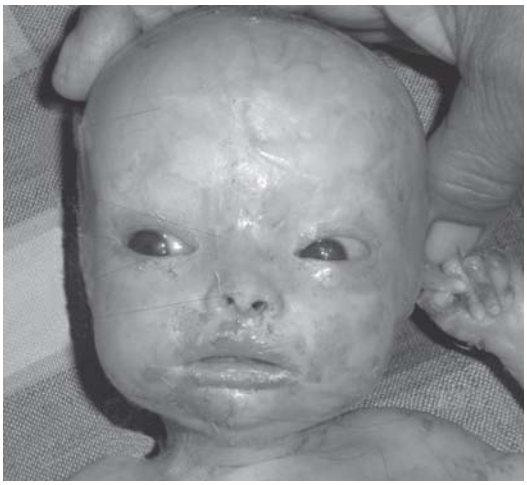

FIG. 1 *Macrocephaly with craniofacial disproportion, total alopecia, hypoplasia of facial bones, upward slanting palpebral fissures, hypertelorism, sparse eyebrows and eyelashes, and downturned angle of mouth.*

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