CASE REPORTS

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 (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-Rautenstauch syndrome.

Wiedemann-Rautenstauch (WR) syndrome is a known neonatal progeroid syndrome comprising of generalized lipatrophy 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 boss-
ing, craniofacial disproportion, almost total alopecia, large
fontanels and wide sutures, prominent scalp veins, hypop-
plasia of facial bones, small nose, upward slanting palpe-
bral fissures, hypertelorism, ocular proptosis, sparse eye-
brows 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
scapal 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 hypertriglyceridermia, 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.

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