Unusual Presentation of Seckel's Syndrome

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Seckel's syndrome, a rare form of short stature was first described by Mann(1) in 1959 and later studied in detail by Seckel in 1960(2). Most of the patients in this syndrome have characteristic features along with mental deficiency. In this communication we present a case with characteristic features of this syndrome with normal intelligence.

Case Report

A 5-year-old girl presented to us with the history of growth failure. There was no history suggestive of chronic infections, respiratory, cardiovascular, renal or central nervous system disorders. The dietary intake was normal for age. She was full term infant with a birth weight of 1500 g, delivered to a 30 yr old, second gravida mother. There was no evidence of intrauterine infections, or exposure to drugs and radiation antenatally. There was no consanguinity and the only other sibling was normal. The child had attained the various social, motor, language and adaptive milestones in time. A social smile was present at approximately 2 months of age; she started sitting at around 8 months and standing at one year of age. She could dress and undress herself at the age of 3 years. She was able to relate a story at about 4 years and is now able to draw a triangle. Mentally the child matched well with peers of her age, but was shy by nature.

On physical examination, height, head circumference, and weight were 87 cm, 38.2 cm and 7.6 kg respectively against an expected of 107 cm, 50 cm and 18kg. The upper to lower segment ratio was 1:1 against an expected of 1.4:1. The child had a characteristic hypoplastic facies, synostosis of cranial sutures, prominent nose, missing ear lobule, teeth enamel hypoplasia, inability to fully supinate left forearm and clinodactyly of fifth finger (*Fig. 1*).

Investigations revealed hemoglobin of 8 g/dl with microcytic hypochromic anemia, alongwith normal blood sugar, urea and calcium levels. Skeletal survey revealed bone age of 3 years alongwith a deformed head of radius in the left arm (*Fig. 2*). The *X*-ray skull showed fusion of all sutures alongwith prominent convolutional pattern (*Fig. 3*). The intelligence quotient (IQ), assessed on the Stanford Binnet Scale, was 98 (normal).

Discussion

Seckel's syndorme is a rare cause of marked growth retardation in children with probable autosomal recessive inheritance(3).

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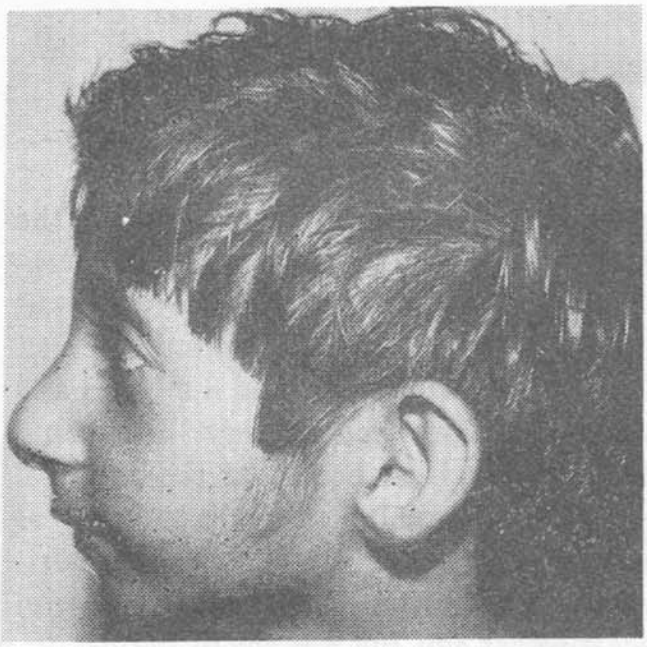

Fig. 1. Characteristic facies of seckels syndrome with absent ear lobule.

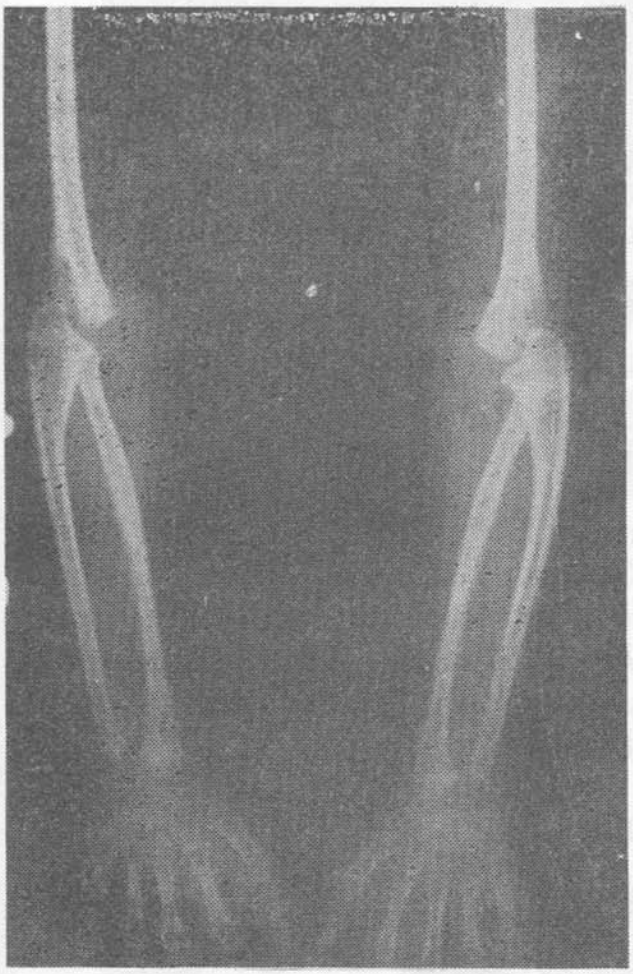

Fig. 2. Hypoplastic head of radius (Left).

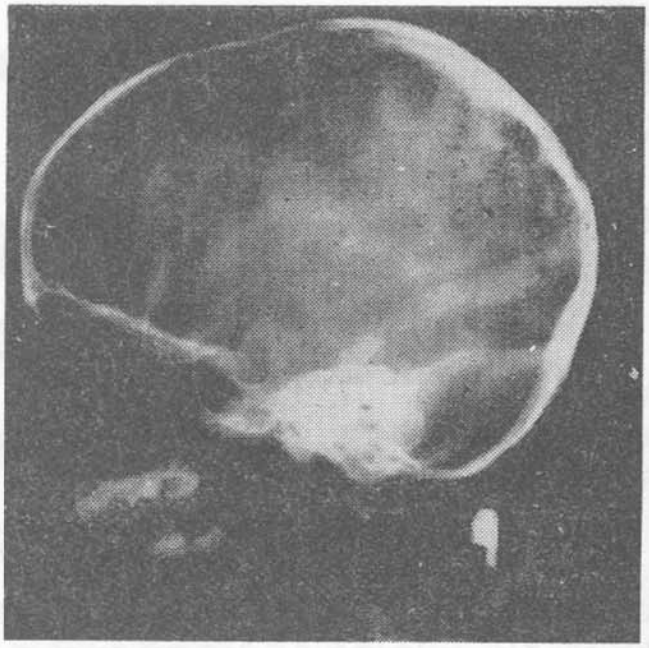

Fig. 3. Silver beaten appearance (Skull).

Majewski and Goecke studied 60 cases reported in the world literature and outlined the various clinical features(4). Few cases have been reported from India also(5-7). The features include growth retardation, microcephaly with premature synostosis, moderate to severe mental deficiency, prominent nose, low set or malformed ears especially a missing ear lobule. Upper extremities exhibit clinodactyly of fifth finger, simian crease, absence of few phalangeal epiphysis and hypoplasia of proximal radius. Lower extremities may show dislocation of hip and hypoplastic proximal fibula(8). Occasional abnormalities include anodontia, enamel hypoplasia, sparse hair, club feet and hypoplastic external genitalia(3). Renal hypoplasia, another rare feature was also reported(7). Our case showed many of these anomalies with the height age of 2 years and bone age of 3 years.

Natural history of disease shows that gestational timing may be prolonged with birth weight of 450 to 1900 g, birth length of 33-42 cm. The final height attained is about 90 to 105 cm. Patients show moderate to severe mental deficiency though early motor progress may be normal. Though our patient showed physical characteristics similar to those of Seckel's syndrome described previously, she had a normal intelligence for this age, despite having microcephaly. This was an unusual finding in this case. Majewski and McKusick *et al.*, reported a few cases with normal intelligence of minimal mental retardation(4,9).

By natural history, we can prognosticate final height or age in this patient. However, autopsy studies in these patients have shown the cerebrum to be small with simple, primitive convolutional pattern resembling that of chimpanzees but all these cases presented with mental deficiency right from the beginning(9). Therefore, the future mental status of this patient can not be predicted accurately.

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Antenatal Diagnosis of Ebstein's Anomaly

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Major cardiac anomalies can be detected in the antenatal period by targetted imaging of the fetal heart. This unusual case reported here illustrates a fetus with Ebstein's anomaly born out of consanguinous marriage. There were

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