Seckel Syndrome in a 9 Year Old Child

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ABSTRACT

Seckel Syndrome first defined by Seckel in 1959, is a rare (incidence 1:10000) genetically heterogeneous, autosomal recessive disorder presenting at birth. This syndrome is characterised by a proportionate dwarfism of prenatal onset, severe microcephaly with a bird headed appearance (beaked nose, receding forehead, prominent eyes and micrognathia) and mental retardation in addition to the characteristics craniofacial dysmorphism and skeletal defects, abnormalities have been described in the cardiovascular hematopoietic, endocrine, gastrointestinal and central nervous system. Usually such patients have poor psychomotor development.

Keywords: Seckel syndrome, dwarfism, microcephaly.

1. INTRODUCTION :

Seckel Syndrome reported by Mann and Russel in 1959, this condition was extensively studied by Seckel in 1960. More precise criteria for diagnosis have been recently been set forth [1, 2].

ABNORMALITIES: The abnormalities regarding growth are- Prenatal onset of marked growth deficiency. Average birth weight at term is 1543g (1000 to 2005g) Mean postnatal growth is 1543 deficiency is -7.1 SD +/- 2.08. One adult was 104 cm. Delayed bone age. Genitalia showingCryptorchidism. In Craniofacies abnormalities are: Microcephaly with secondary premature synostosis. In one half of cases, head circumference is more retarded than height, while for the remainder it is as retarded as height. Receding forehead. Prominent nose, micrognathia, low- set malformed ears with lack of lobule. Relatively large eyes with down slanting palpabral fissures. Occasionally facial asymmetry, strabismus, partial anodontia, enamel hypoplasia, sparse hair & cleft palate may be there. Mental deficiency was noticed, nearly one half with IQ less than 50 and occasional seizures. Skeletal abnormalities in upper extremitiesare- Clinodactyly of fifth finger, simian crease, absence of pharyngeal epiphysis, hypoplasia of proximal radius with dislocation of the radial head. Lower extremities shows- Dislocation of Hip, hypoplasia of proximal fibula, a gap between first and second toes, Inability to completely extend knees, with occasional scoliosis, talipes, pes planus, hypoplastic external genitalia. Hypoplastic anemia and chromosome breakage may be seen occasionally. Abnormalities in thorax showing only 11 pairs of ribs.

2. CASE REPORT :

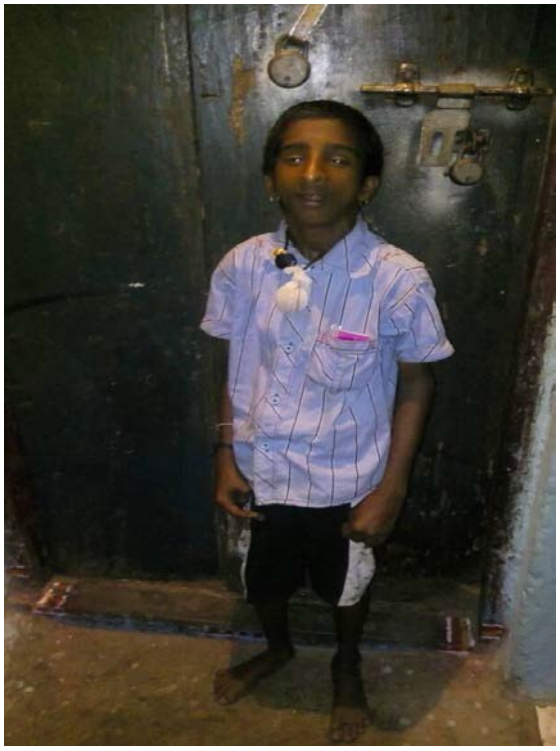
9 year old male child presented with complaints of failure to thrive and global developmental delay since infancy along with short stature. Antenatal ultrasound showed sever IUGR baby. Birth history was uneventful & birth weight not known. Born out of 2nd degree consanguineous marriage with no similar history in the family. There was gross growth retardation, global developmental delay & mental retardation. On examination- Microcephaly, bird headed like appearance, prominent nose, receding forehead, lobeless ear, high arched palate, antimongoloid slant (prominent eye), empty scrotum (undescended testis). Anthropometry- Weight: 6.2 kg, Height: 83cm, Head Circumference- 39cm, US/LS: 0.6. All being <3rd percentile as IAP growth chart. On systemic examination- no abnormality detected. A diagnosis of Seckel Syndrome was

made using diagnostic criteria. Bone age corresponding to 2-3 years, rudimentary testis on the abdominal scan, empty sella reported on CT scan.

3. DISCUSSION :

Seckel Syndrome is a rare AR inherited trait with the malformation. The male to female sex ratio 9:11. This syndrome is a heterogeneous form of primordial dwarfism. The synonyms of this syndrome include seckel dwarfism, bird head dwarfism, nanocephalic dwarfism and microcephalic primordial dwarfism [3]. Rudolph Virchow introduced the term “bird headed dwarf” in the context of proportionate dwarfism with low birth weight, mental retardation, a pointed nose, and micrognathia.
DIAGNOSTIC CRITERIA: Prenatal onset of marked growth deficiency, microcephaly with secondary premature synostosis [3]. Receding forehead, prominent nose micrognathia [4] low set ears malformed ears with the lack of lobule down slanting palpebral fissures [5]. Clinodactyly of fifth finger dislocation of the hip gap between the first and second toe. Male- cryptorchidism.
NATURAL HISTORY: Seckel Syndrome have moderate to severe mental deficiency. Early motor progress maybe near normal. Gestational timing may be prolonged. Although moderate to severe mental deficiency occurs, early motor progress may be near normal. The cerebrum is small, with a simple primitive convolutional pattern resembling that of a chimpanzee. Though they tend to be friendly and present, these patients are often hyperkinetic and easily distracted. Poor joint development and support may be evident by dislocations of hip, elbow or both. And by the later development of scoliosis, kyphosis or both. Survival to an age of 75 has been recorded.
ETIOLOGY: Autosomal recessive. Chromosome 3 and 18 involved.

Fig. 1: Seckel Syndrome. A 9 year old with a height of 83cms, weight of 6.2 kgs, head circumference of 39cms. US/LS:0.6. Note the bird head like appearance, prominent nose, receding forhead, prominent eyes and gross growth retardation and microcephaly.



4. CONCLUSION :

The majority of the cases of severe IUGR and microcephaly and prominent facial feature are detectable by serial USG scans. Parents may be informed about importance of USG during pregnancy in the context of proportionate dwarfism with low birth weight mental retardation a pointed nose and micrognathia.

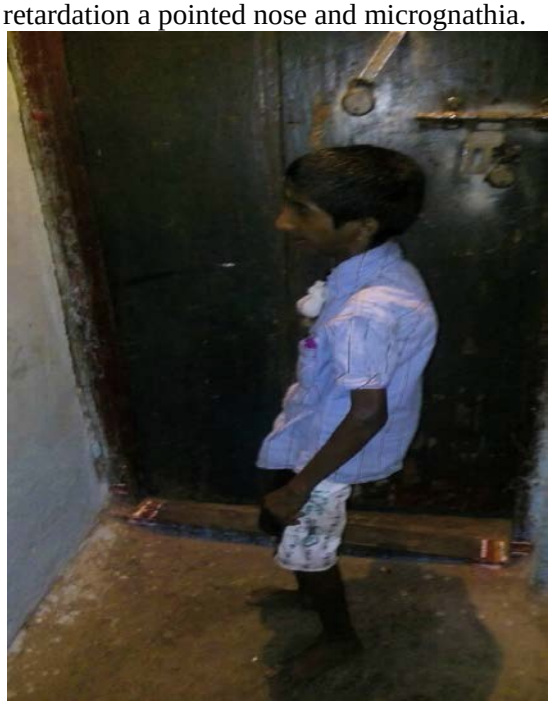


Fig 2: Note the bird head like appearance, prominent nose.

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