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SECKELS SYNDROME: A RARE CASE REPORT

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ABSTRACT

Seckel syndrome, first defined by Seckel in 1960, is a rare, genetically heterogeneous autosomal recessive disorder presenting at birth with incidence of 1:10,000. This syndrome is characterized by a proportionate dwarfism of prenatal onset, severe microcephaly with a "bird-headed" like appearance (beaked nose, receding forehead, prominent eyes, and micrognathia) and mental retardation. In addition to the characteristic craniofacial dysmorphism and skeletal defects, abnormalities have been described in the cardiovascular, hematopoietic, endocrine, gastrointestinal, and central nervous systems. Usually such patients have poor psychomotor development. We hereby present a rare case of Seckel syndrome.

KEYWORDS Craniofacial dysmorphism; Mental retardation; Seckel syndrome

INTRODUCTION:

Seckel syndrome, first defined by Seckel in 1960, a rare autosomal recessive condition without any sex predilection, with a reported incidence of 1:10,000 live born children.[1] Rudolf Virchow introduced the term "bird-headed dwarf" in the context of proportionate dwarfism with low birth weight, mental retardation, a pointed nose, and micrognathia.[2] This syndrome is also considered to be a variant of Harper syndrome.

CASE REPORT:

A 14-year-old male patient born of third degree consanguineous married couple, presented with complaints of delayed developmental milestones. Mother was 21yr old when she conceived and her antenatal period was not booked and no ultrasound scans were taken. Birth history revealed that the child was born of uneventful full-term normal delivery with birth weight of 2 kg. Developmental history revealed grossly delayed developmental milestones, severe growth and mental retardation. Similar history noted in his younger sibling (8yr old). On general examination, both the siblings had short stature with weight, height and head circumference being <3rd centile. On physical examination, both the siblings presented with receding forehead, prominent eyes, pointed nose, and micrognathia, the characteristic features of "bird-headed dwarf". Systemic examination showed hypertonia of all limbs with exaggerated deep tendon reflexes. A diagnosis of Seckels syndrome was made based on these clinical findings.

Fig 1: Siblings with Seckel syndrome

Fig 2: Dysmorphic features in both siblings

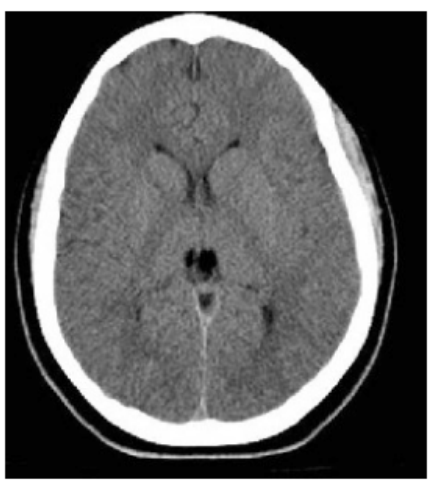

Fig 3: CT scan showed agenesis of corpus callosum which is a feature of Seckels syndrome.

DISCUSSION:

Seckel syndrome is a rare constellation of malformations, presumably inherited as an autosomal recessive inherited trait. The male to female sex ratio is 9:11. This syndrome is a heterogeneous form of primordial dwarfism.[3] The synonyms of this syndrome include Seckel dwarfism, bird-headed dwarfism, nanocephalic dwarfism, and microcephalic primordial dwarfism. Seckel syndrome encompasses a number of facial and brain abnormalities possibly associated with prenatal and postnatal growth restriction.[4]

The syndrome is characterized by intrauterine growth retardation (average birth weight 1540 g) and severe proportionately short stature with severe microcephaly[5]. In the present case, developmental history revealed delayed developmental milestones and severe growth retardation. The syndrome is characterized by mental retardation with an IQ less than 50.[5] These patients are often hyperkinetic and easily distracted. In the present case, patient was mentally retarded and mental-age of the patient was less when compared to the chronological age. The etiopathogenesis of this syndrome remains unclear. The mode of inheritance in Seckel syndrome is thought to be autosomal recessive. Advances in molecular genetics have shown someaberration in a few chromosomes. It can be due toincreased chromosomal instability or chromosomal breakage. Chromosomal aberrations causing Seckel syndrome have been reported in earlier studies in genes 2q33.3-34, 18p11.31-q11.2,and 3q22.1-q24.[6]

Clinical facial features of this syndrome include"bird-headed profile" with receding forehead, largeeyes, beak-like protrusion of the nose, narrow face,receding lower jaw, and micrognathia. In the presentcase, the patient presented with characteristic features of a "bird-headed profile."Other occasional features include high-archedpalate, cleft palate, malocclusion, enamel hypoplasia, premature closure of cranial sutures secondary to diminished brain growth, antimongoloid slant of palpebral fissures, dysplastic ears, clinodactyly of the fifth fingers, cryptorchidism, clitoromegaly, hirsutism, agenesis of corpus callosum, pachygyria, retarded bone age, frequent hip dysplasia, dislocationof the head of radius, low-set ears, and 11 pairs of ribs. In addition, abnormalities have been found incardiovascular, hematopoietic, and endocrine, as well as nervous system.

The primary diagnostic features are severe intrauterinegrowth retardation, microcephaly, characteristic"bird-headed profile," and mental retardation.[3] Inmost cases, diagnosis depends upon recognition of clinical findings. In the present case, a diagnosis of Seckel syndrome was made based on history, delayeddevelopment, and clinical features (bird-headedprofile).

The craniofacial features of Seckel syndrome allow its differentiation from other syndromes of growth deficiency with microcephaly, suchas Dubowitz syndrome, fetal alcohol syndrome,trisomy18 syndrome, Cockayne syndrome, progeria, Hallermann-Streiff syndrome (HSS), DeLange syndrome, and Fanconi syndrome.[3] SS comprises of a combination of growth retardation, microcephaly, micrognathia, and a beak-like nose and is termed as bird-headed dwarfism.

Children affected with Seckel syndrome do have anormal lifespan although they often have profoundmental and physical deficits. Survival of thesepatients till the age of 75 years has been recorded.[3] Recurrence of the disease can be suspected by theobservation of intrauterine growth retardation withmicrocephaly in the second trimester of pregnancywhen a first child was born with Seckel syndrome. Early molecular antenatal diagnosis can be performedfor a couple who had a first child with Seckel syndrome if the familial mutations have been identified.

CONCLUSION :

The importance of serial ultrasound scans during pregnancy is revealed once more by the present case report. Because our patients were not followed by serial ultrasound scans, severe intrauterine growth retardation (IUGR), microcephaly, and prominent facial features could not be diagnosed at prenatal period. Parents may be informed that the majority of cases of severe IUGR and microcephaly are detectable by serial ultrasound scans from 16 to 20 weeks of pregnancy.

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