Kuş Kafalı Cücelik (Seckel Sendromu) - Nadir Bir Olgunun Sunumu

[Bird-Headed Dwarfism (Seckel Syndrome) - A Rare Case Report]

ÖZET
Seckel Sendromu, tipik kuş yüzü görünümü, büyüme geriliği, mikrosefali, zekâ geriliği, değişken kromozom instabiliteleri ve hematolojik problemler görülen nadir otozomal resezif bir bozukluktur. Biz anemi ile beraber seyreden Seckel Sendromuna benzer klinik bulguları olan 16 yaşında bir kız hastayı rapor ettiğimiz.

SUMMARY
Seckel syndrome is a rare autosomal recessive disorder with a typical bird face appearance, growth retardation, microcephaly, mental retardation, variable chromosomal instability, and hematological disorders. We report a case of 16 years old girl with clinical features resembling seckel syndrome along with anaemia.

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INTRODUCTION
Seckel syndrome, first defined by Seckel in 1960, is characterized by microcephaly, proportionate dwarfism of prenatal onset, and a typical “bird-headed” profile (beaked nose, receding forehead, prominent eyes and micrognathia). Various other facial and skeletal abnormalities have been documented by subsequent authors, including low-set ears with hypoplastic lobules, premature closure of cranial sutures, fifth finger clinodactyly, dislocation of radial heads and, eleven pairs of ribs (1).

CASE REPORT
An 18 years old female patient complained of forwardly placed upper front teeth. No history of consanguineous marriage. She had growth retardation with IQ slightly below normal. On extraoral examination depicted severe mandibular deficiency with midface hypoplasia, bulbous nose with overhanging columella, loss of hair in the frontal region with receding forehead, microcephaly, low set ears, along with palpbable upslant of both eyes giving a “Bird headed dwarfism” (figure 1, 2 and 3). She had club foot of left leg (figure 4). On intraoral examination there was Class 2 malocclusion, partial ankyloglossia along with high arched palate (figure 5) and speech problems was present. An array of investigations was carried out which depicted severe anemia with hemoglobin level below 8 gm/dl.
Figure 1. Upslanting Palprbral fissure

Figure 2. Micrognathia, beaked nose, low set ears

Figure 3. Micrognathia
DISCUSSION

Seckel devoted a monograph to a group of patients for whom he used the designation 'bird headed dwarf'. In that monograph he called attention to a specific type of dwarfism with mental retardation, low birth weight, small head, large eyes, a beaklike nose, narrow face, receding mandible, and dental anomalies (2).

The incidence of this syndrome is less than 1 in 10,000. Krishna et al. in 1994 and Shanske et al. in 1997 characterized this disorder as autosomal recessive, resulting from consanguineous marriages. A total of 60 cases have been reported since 1960, out of which only 19 have been described of having classical Seckel syndrome by Sugio et al in 1993. Most recently, the locus of this syndrome has been mapped to chromosome 3q22.1-q24 by Goodship et al (3).

Due to inconsistency in diagnosis, less than one-third of the reported cases appear to fulfil Seckel’s original criteria (4).

The mode of inheritance in Seckel syndrome is thought to be autosomal recessive. The rarity of the syndrome, lack of a definite diagnostic test, frequent phenotypic variability as well as the recent finding of involvement of different chromosomes have resulted in confusion and debate over exact definition of this condition (1).

It is supposed to be caused by defects of genes on chromosome 3 and 18. One form of Seckel syndrome can be caused by mutation in the gene encoding the ataxia telangiectasia and Rad3 related protein (ATR)
which maps to chromosome 3q22.1-q24. This gene is central in the cell's DNA damage response and repair mechanism (5).

Craniofacial feature includes severe microcephaly, receding forehead, relatively large eyes, and micrognathia, lending prominence to the midface and curved nose. Other craniofacial features may include facial asymmetry, down-slanting palpebral fissures, highly arched or cleft palate, enamel hypoplasia, crowded teeth, and class II malocclusion. The significance of dental alterations in Seckel's bird-headed dwarfism resides in the defect, hypoplastic enamel, being limited to the primary dentition; in most instances the second primary molar tooth is not affected (6).

Limb anomalies include clinodactyly of fifth finger, abnormal finger flexion creases, dislocation of radial head, and hip dysplasia (6).

In most of the cases, diagnosis depends on recognition of clinical finding. In some cases, increased chromosomal breakage has been reported, but it was not confirmed in all the cases of Seckel syndrome and cannot be used as a tool for the diagnosis. X ray features include retarded bone age, frequent hip dysplasia and dislocation of the head of the radius (7).

Seckel syndrome is rare anomaly even though treatment of such cases is still in search but a cosmetic surgery may play a pivotal role in repay the normal appearance along with psychological benefit to the patient. Clear pathogenesis with understanding of chromosomal instability is required which may figurative to prevent this syndrome.

REFERENCES