CASE REPORT

SECKEL SYNDROME: REPORT OF A RARE CASE

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ABSTRACT

Seckel syndrome is a rare (incidence 1:10,000), genetically heterogeneous autosomal recessive disorder manifesting at birth. It is named after Seckel who first described it in 1960. Mental retardation and proportionate dwarfism of prenatal onset are the most notable features of this syndrome. It is also marked by a receding forehead, prominent eyes, micrognathia, and a 'bird-headed' appearance. Amongst dental manifestations hypoplastic enamel is most commonly found in the primary dentition in this syndrome; the second primary molar is usually not affected. Other dental manifestations include retrognathia, hypodontia, crowding, cleft palates, and Class II malocclusions. This is a case report of a pediatric Seckel syndrome patient describing the clinical features and management.

KEY WORDS

Seckel, Cryptorchidism, Bird-headed dwarfism.

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INTRODUCTION

Seckel syndrome is an extremely rare congenital autosomal disorder. It is also known as bird-headed dwarfism, Harper's syndrome, Virchow-Seckel dwarfism, and Bird-headed dwarf of Seckel[1].It is named after Helmut Paul George Seckel, an American physician.[2] Seckel syndrome is also referred to as Harper's syndrome, named after RitaG. Harper.[3] Rudolf Virchow first introduced the term "bird-headed dwarf". Seckel syndrome is a rare, congenital heterogenous autosomal disorder that presents at birth in one in 10,000 live-born children.[4,5]

There is severe microcephaly and intrauterine growth retardation associated with this syndrome."bird-headed" appearance (beaked nose, receding forehead, prominent eyes, and micrognathia), and mental retardation are also seen.[6] There were also a number of other facial and skeletal abnormalities noted, including hypoplastic ear lobules, premature closure of cranial sutures, clinodactyly of the fifth finger, dislocation of the radial heads, and 11 pairs of ribs.[7] Dental anomalies include retrognathia, hypodontia, hypoplasia of the enamel, crowding, cleft palates, and Class II malocclusions[8] The case report highlights the clinical characteristics and management of a pediatric patient with Seckel disease.

CASE REPORT

A 10-year-old male patient reported to the department of Pedodontics and preventive dentistry with the chief complaint of pain in the upper and lower back tooth. An examination of the patient's birth history revealed that he was born to non-consanguineous parents at full-term cesarean delivery. The weight of the patient at birth was 1.5 kg. Developmental milestones were delayed and growth retardation was observed in the child. Past medical history revealed patient underwent orchidopexy of the right testis 2 years ago. In light of the patient's history, developmental status, and clinical findings, Seckel syndrome was diagnosed.

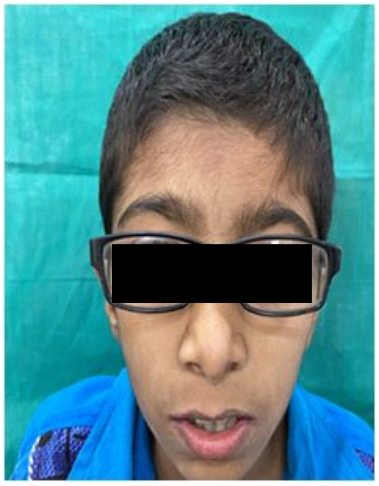

Figure – 1: Frontal view

Figure – 2 : lateral view

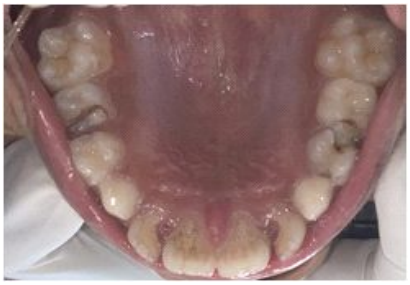

Figure – 3 : upper occlusal view

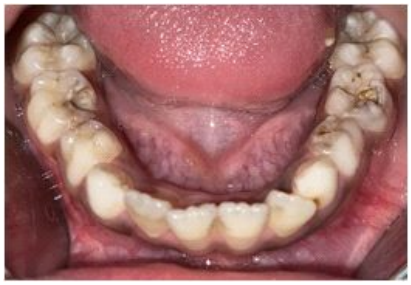

Figure – 4 : lower occlusal view

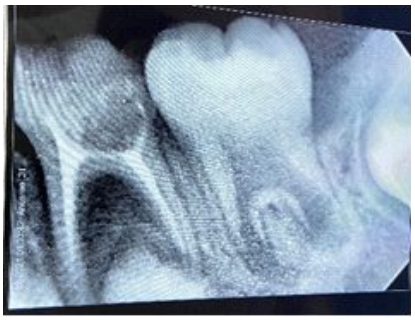

Figure -5 : IOPAR of 75 after endodontic treatment

Figure -6 : short stature

EXAMINATION

The patient is currently ten years old, has short stature, and sports a height of 118.5cm and a weight of 18kg. The patient's head circumference is 40cm.A general examination suggested mental impairment, an ataxic walk, and aberrant speech.

Upon extraoral examination, a patient was found to have microcephaly, low-set ears with small earlobes, a large nose, prominent eyes, with strabismus, short height (118.5 cm), and a fairly small mandible, with a head circumference in the range of 40 cm. [Fig-1. Fig-2, Fig-6]

Intraoral examination revealed a V-shapedhigh-arched palate, mixed dentition with 24 teeth, and an increased overjet pattern was observed in class 1 dentoskeletal pattern. [Fig-3, Fig-4]

Dental treatment included oral prophylaxis, restoration was done on 12 and 22, and pulpectomy followed by stainless steel crown was given on 75. [Fig.5]

DISCUSSION

Seckel syndrome is a rare collection of birth defects that is likely passed down through the autosomal recessive gene. This syndromeis a diverse type of primordial dwarfism. The terms "seckel dwarfism," "bird-headed dwarfism," "nanocephalic dwarfism," and "microcephalic primordial dwarfism" are all synonyms for this syndrome. Linkage studies may enable to locate the gene, which will enable the cloning and uncovering of the underlying pathogenesis, responsible for the symptoms of Seckel syndrome. The etiopathogenesis of this syndrome is still a subject of investigation.

Seckel syndrome is hypothesized to have an autosomal recessive mode of inheritance. Chromosome breakage or increased chromosomal instability may be to blame. The genes 2q33.3–34, 18p11.31–q11.2, and 3q22.1–q24 have been implicated in earlier investigations as having chromosomal abnormalities that cause Seckel syndrome.[9] Intrauterine growth retardation (average birth weight: 1.5 kg), severe proportional short height, and severe microcephaly are the hallmarks of the syndrome. The developmental history in the present instance showed missed developmental milestones and significant growth retardation. The patient was born weighing 1.5 kg, and at the time of the examination, he weighed 18kg. He was 118.5 cm tall, and his head circumference was 40 cm, which is significantly smaller than that of a typical boy his age.

This syndrome's clinical facial characteristics include a "bird-header profile" with a receded forehead, prominent eyes, a protruding beak on the nose, a narrow face, a retracted lower jaw, and micrognathia. In this instance, the patient displayed the tell-tale signs of this profile. Other features include agenesis of the corpus callosum, pachygyria, delayed bone age, frequent hip dysplasia, dislocation of the head of the radius, low-set ears, eleven pairs of ribs, premature closure of cranial sutures secondary to diminished brain growth, dysplastic ears, strabismus, hirsutism, pachygyria, clinodactyly of the fifth fingers, cryptorchidism, clitoridomegaly[10]. Intraoral findings commonly seen are high-arched palate, cleft palate, malocclusion, and enamel hypoplasia.

There have also been reports of more severe defects such as newborn cholestasis, tetralogy of Fallot, coarctation of the aorta, atrial septal defect, cardiomegaly, and cardiac arrhythmias that occasionally result in death. Also noticed are renal conditions such as renal tubular leakage, localized medullary hypoplasia, renal cysts, or renal hypoplasia[11].

Diseases like Dubowitz syndrome, fetal alcohol syndrome, Trisomy[18], De Lange syndrome, and Bloom syndrome are the primary differential diagnoses for this syndrome. The majority of these diseases exhibit traits like microcephaly, facial asymmetry, micrognathia, and disparity in the midface. However, other characteristics of Seckel syndrome, which are practically visible in our case report, include delayed cranial sutures, cryptorchidism large ears with low-set earlobes, a large nose, and a comparatively short jaw.

CONCLUSION

Seckel syndrome is a relatively rare condition caused by gene abnormalities that cause chromosome instability and a variety of haematological, hormonal, and physical disorders. To avoid a poor prognosis and any related consequences like death brought on by cardiac insufficiency, arrhythmias, or pituitary insufficiency, various signs of the illness should be examined and diagnosed as early as feasible. To prevent these kinds of illnesses, genetic counseling is typically advised.

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