CASE REPORT

Seckel Dwarfism—A Rare Autosomal Recessive Inherited Syndrome: A Case Report

Neeti Tatiya[1], Rituraj Kesri[2], Ankita Ukey[3]

Abstract

Seckel syndrome, also commonly called Seckel dwarfism, is a rare congenital disorder and always associated with severe growth retardation *in utero*. This retarded growth lingers on and causes serious developmental deformities ensuing to short stature, microcephaly, mental retardation, and a beak-like nose. This case report intends to present an interesting case of a 14-year-old female patient with various clinical manifestations, typical radiographic features, and characteristic dental manifestations correlated with the literature. A detailed understanding of the present case would assist pediatric dentists in correct and prompt diagnosis, precise treatment, and the prevention of severe consequences caused by Seckel syndrome.

Keywords: Autosomal recessive disorders, Bird nose dwarfism, Case report, Seckel syndrome.

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Introduction

Genetic material chromosomes are present in every human cell; there are 23 pairs of chromosomes embraced from both parents that carry our genetic information as genes. If these genes undergo any harmful change (mutation), they affect the normal physiologic development and functions of the body. Autosomal recessive genetic disorders are seen in individuals inheriting two genes for the same trait.[1] Seckel syndrome is such a rare autosomal recessive disorder; the disease is a rare entity as very limited reported cases of the disease have been seen in literature. The syndrome is characterized by low birth weight, microcephaly, typical facial appearance, receding forehead and chin, a large and prominent beaked nose and large or bulging eyes, proportionate dwarfism, moderate to severe mental retardation, antimongoloid slant of the eyes, delayed mental development, microcephaly, dental abnormalities, and clinodactyly of the fifth finger, 11 pair of ribs, receding hair and redundant wrinkled skin on the palms, dislocation of the radial head, absent earlobes, secondary premature synostosis, retarded bone age.[2–5] This disease does not show any sex predilection. The sex ratio of males to females is 9:11, with an incidence of 1:10,000 children.[3,4] The syndrome was first defined by Helmut PG Sickle.[6] The other name for the syndrome was used by Rudolf Virchow; he described it as "bird-headed dwarf" in the context of proportionate dwarfism with low birth weight, mental retardation, a pointed nose, and micrognathia. There are instances of recurrence of the syndrome in succeeding siblings of up to 25%.[7] This case report intends to discuss various clinical features and management in a pediatric patient with Seckel syndrome.

Case Description

A 13-year-old child reported to the Department of Pedodontics and Preventive Dentistry with a complaint of tooth decay and pain in the lower right back tooth region. On physical examination, she was 100 cm in height, weighing 15 kg, and had a head circumference of 36 cm (Fig. 1). Parental history revealed parents were unaffected, and the child was born to consanguineous marriage, born preterm at 30 weeks gestation with a weight of 930 gm at birth. History revealed that during the gestation period, ultrasonography was not performed to detect fetal defects. Postbirth, all the developmental milestones were delayed. A history of child's diet revealed that the child was not on regular solid food. The oral hygiene was restricted to cleaning his mouth once a day in the morning.

On extraoral examination, the patient showed a microcephalic head, receding forehead, sparse hair, prominent eyes, midface hypoplasia, deficient mandible, low set ears, retarded bone age, frontal bossing, hirsutism, dysplastic ears, dilated veins in the temporal region, and clinodactyly (Fig. 2). The response and behavior of child suggested mild to moderate mental retardation. Frankl's behavior rating scale was definitely positive with incomprehensible speech. Intraoral examination revealed generalized microdontia, lower incisors missing, and multiple over-retained teeth (74,75,85) with moderately inflamed gingiva and mobility (Figs 3 and 4). Dental caries were seen in 24, 25, 16, 74, 75, and 85; all these teeth were symptomatic and suggestive of pulp involvement. All teeth exhibited increased mobility.

On investigating in full-mouth intraoral radiographs, conical roots of all teeth were seen with involvement of pulp in the over-

[1]Department of Pedodontics and Preventive Dentistry, Saveetha Dental College, Chennai, Tamil Nadu, India

[2]Consultant Pedodontist, Raipur, Chhattisgarh, India

[3]Department of Pedodontics and Preventive Dentistry, Maitri Dental College and Research Centre, Durg, Chhattisgarh, India

Corresponding Author: Neeti Tatiya, Department of Pedodontics and Preventive Dentistry, Saveetha Dental College, Chennai, Tamil Nadu, India, Phone: +91 7987959356, e-mail: drneetitatiya@gmail.com

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Fig. 1: A 14-year-old girl with exhibiting short stature and delayed developmental milestones

retained teeth (Fig. 5). A handwrist radiograph was also taken, which showed 1-year-old skeletal maturation (Fig. 6). The lateral cephalogram revealed beaked prominent soft tissue shadow of nose and pneumatization of the mastoid sinus (Fig. 7). Child was investigated for complete blood picture and reported anemic with 8.1 gm% of hemoglobin. The treatment procedure charted was the extraction of 85 mobile grossly decayed teeth under local anesthesia.

Discussion

It was in 1960 that Seckel, after studying nanocephalic dwarfs reported in the literature over more than two centuries, came up with a syndrome that was characterized by proportionate dwarfism with mental retardation, low birth weight, small head,

Fig. 2: Lateral view showing beaked nose

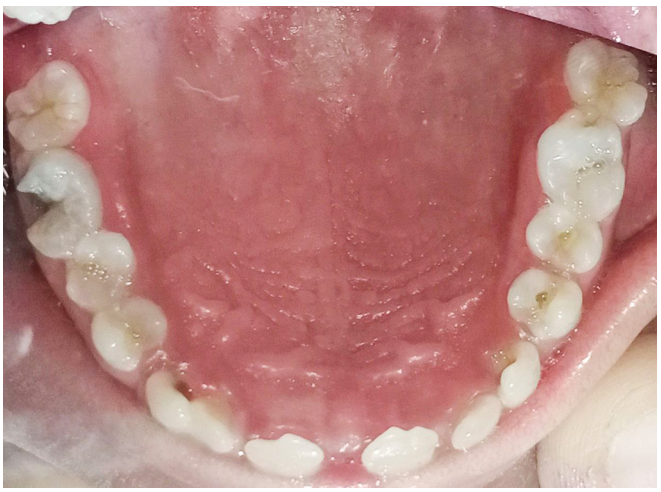

Fig. 3: Intraoral view—maxilla

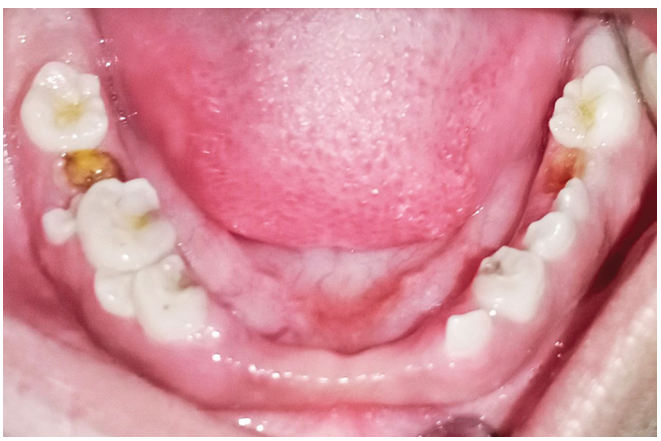

Fig. 4: Intraoral view—mandible

large eyes, beak-like nose, receding mandible, narrow face, and dental abnormalities.[8] Seckel syndrome is diagnosed on the basis of clinical features seen in the present case; these can be compared with the features reported by Majewski and Goecke,[9] which were supported by similar findings given by Thompson and Pembrey, which are explained and tabulated (Table 1).[10] To confirm the radiological findings and to see for any bone

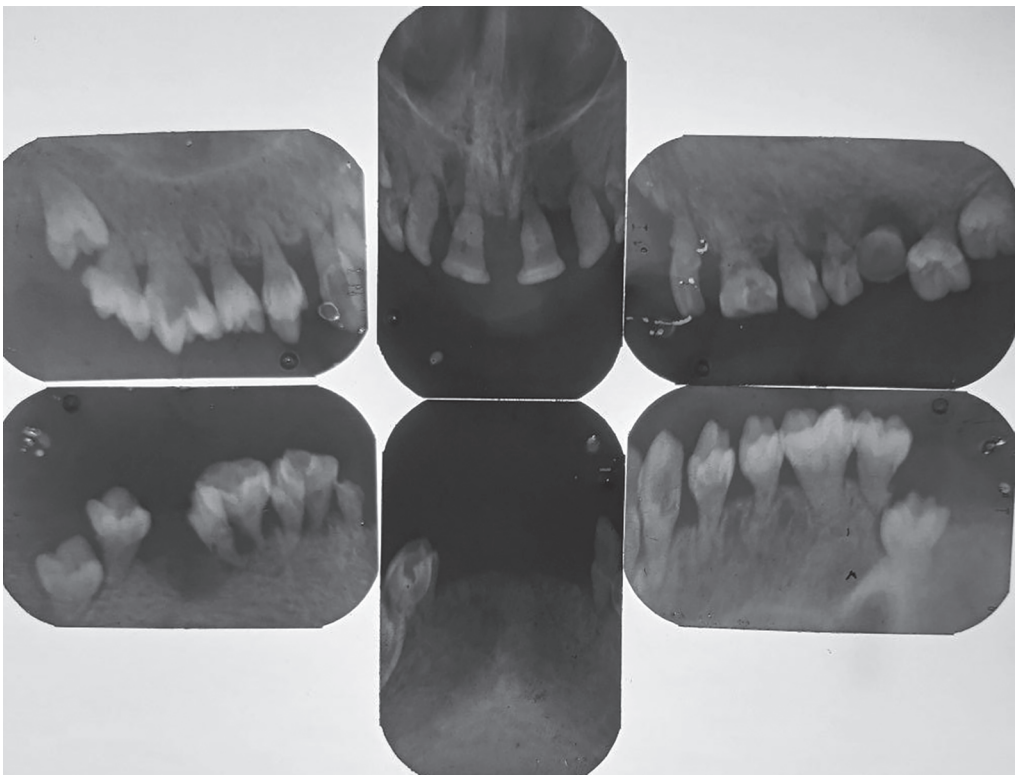

Fig. 5: Full-mouth IOPA

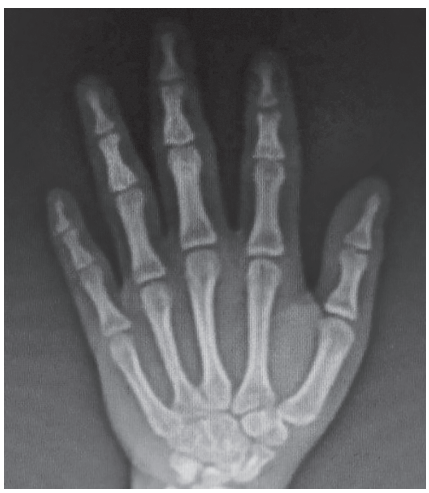

Fig. 6: Handwrist radiograph showed 1-year-old skeletal maturation

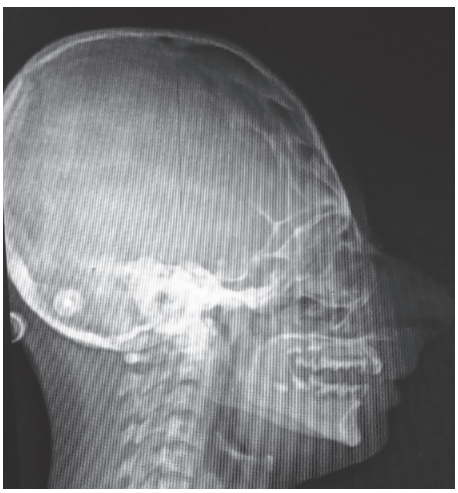

Fig. 7: Lateral cephalogram revealed beaked prominent soft tissue shadow of nose and pneumatization of mastoid sinus

deformities, a radiographic examination of the teeth and perioral structure was seen in intraoral periapical radiograph (IOPA), lateral cephalogram, and handwrist radiograph, which revealed signs of growth retardation, microcephaly, micrognathia, and a beak-like nose which are characteristic features and in accordance with the findings observed by Sisodia et al.[11] Although there is no definite etiological reason for the development of this syndrome, apart from various factors, consanguineous marriage has been considered a major factor for genetic autosomal recessive diseases like Seckel syndrome. A positive familial history in the present case is seen, as the parents are cousins in close relation, which has been supported in the patient history reported by Rao et al.,[12] which clearly showed the inheritance of the autosomal recessive syndrome from a maternal great aunt. Genetic engineering with all its advancements has led to discoveries which have been helpful in diagnosis of various genetic disorders, gene responsible for Seckel syndrome (SCKL1) was mapped on chromosome 3q22.1-q24 in two families and later identified ataxia–telangiectasia and Rad3-related protein in the year 2000.[13] Another locus was mapped to chromosome 18p11.31-q11.2 (SCKL2) in 2001 and to chromosome

Table 1: Clinical features of Seckel syndrome

Features	*Thompson and Pembre, 1985*	*Majewski and Goecke,1982*	*This patient*
Intrauterine growth retardation with proportionate trunk-to-leg length	3/3	17/17	Present
Short stature (height—3 SD)	3/3	16/17	Present
Microcephaly and mental retardation	3/3	17/17	Present
Facial feature	3/3	17/17	Present
Micrognathia and large nose	3/3	17/17	Present
Receding forehead	3/3	NR	Present
Small palpebral fissures	3/3	NR	Present
Telecanthus	2/3	10/16	Absent
Bulging eyes	None	7/11	Present
Antimongoloid slant	NR	NR	Present
Cataract	1/3	3/17	Present
High-arched palate	1/3	3/17	Absent
Clefting	3/3	NR	Present
Small ears	3/3	10/12	Absent
Lobeless ears	3/3	NR	Absent
Crowded and maloccluded teeth			
Skeleton	3/3	8/8 clinodactyly V	Present
Hand abnormalities	3/3	Gap between first and second	Present
Feet abnormalities	3/3	toes, hallux valgus	Absent
Dislocated head of radii	3/3	3/6	Absent
Hips—fixed flexion	None	5/9	Absent
Hips—congenital dislocation	3/3	None	Absent
Knees—fixed flexion		NR	
Cryptorchidism	None	3/4	Absent
Clitoromegaly	None	3/7	Absent

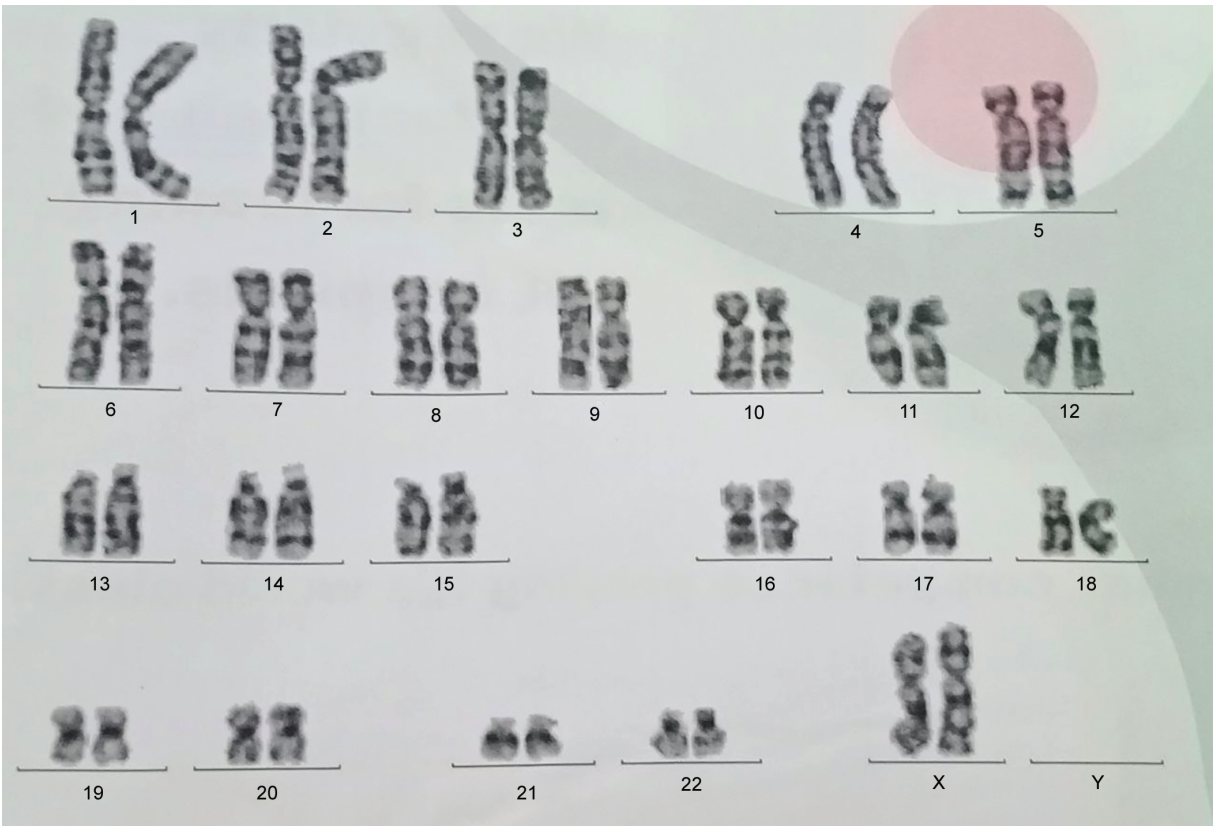

Fig. 8: The karyotyping showed chromosomal analysis (GTG-banding) revealing a normal female karyotype

14q23 (SCKL 3) in 2003.[14] The karyotyping done in the present case showed chromosomal analysis (GTG-banding) revealing a normal female karyotype but also concluded that single gene abnormalities cannot be ruled out by karyotyping test alone and clinical correlations have to be considered while reaching to a conclusive diagnosis (Fig. 8). Thus, correct evaluation of the dental features and a thorough past medical and dental history relating the normal developmental landmarks and other systemic clinical features are mandatory to differentiate in determining the correct diagnosis of Seckel syndrome as it closely resembles Cockayne syndrome, progeria, Hallermann–Streiff syndrome, and Dyggve–Melchior–Clausen syndrome.[15–18]

Conclusion

Seckel syndrome is an extremely rare disorder that shows severe retarded developmental milestones, along with various dental anomalies. The patient and the caregivers must be educated about the potential problems that they would encounter if proper care and oral hygiene methods were not followed, as seen in the present case. It is important for both the parents and the dentist to undergo regular follow-ups and provide anticipatory preventive measures and interventions in order to avoid invasive and complicated treatment protocols.

Orcid

Neeti Tatiya https://orcid.org/0000-0002-0099-8064

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