IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 22, Issue 3 Ser.14 (March. 2023), PP 10-15
www.iosrjournals.org

A Rare Case Report of Seckel Syndrome: Dentofacial Management

Dr Shruti Sudarsanan[1], Dr Anupam Kumar[2], Dr Arya S Kumar[3], Dr Umm Kulthum[4]

[1](Department of Pediatric and Preventive Dentistry, Government Dental College Kottayam, India)
[2](Department of Pediatric and Preventive Dentistry, Government Dental College Kozhikode,India)
[3](Department of Pediatric and Preventive Dentistry, Government Dental College Kottayam, India)
[4](Department of Pediatric and Preventive Dentistry, Government Dental College Kottayam, India)

Abstract:
Seckel syndrome, autosomal recessive in its inheritance, is a rare genetic disorder presenting at birth. Growth retardation, a very small head (microcephaly), intellectual disability and distinctive physical traits like unusually large eyes, a beak-like nose, a narrow face, and a receding lower jaw are all characteristics of Seckel syndrome. Due to the multiple systemic involvements in Seckel syndrome, a single approach is not enough. The treatment is mainly supportive and based on the presentation of symptoms. Thus, a multidisciplinary team is needed for oral and maxillofacial management.We present a case report describing the dentofacial management executed on a 9-year-old female Seckel syndrome patient who came in with the complaint of a cystic lesion.

Key Word*: Seckel Syndrome; Bird-Headed Dwarfism; Craniofacial Anomalies; Dentofacial Features; Genetic*

Date of Submission: 10-03-2023 Date of Acceptance: 22-03-2023

I. Introduction

Seckel syndrome is a rarely seen hereditary condition that impairs a child's development both during pregnancy and after birth[1]. Low birth weight, small height, and achondroplasia are its defining characteristics. The Seckel syndrome affects roughly 1 in 10,000 live-born children[2,3]. Other names for the Seckel syndrome include bird-headed dwarfism, Bird-headed dwarf of Seckel, Harper's syndrome, and Virchow-Seckel dwarfism[4]. The condition bears Helmut Paul George Seckel's name, an American physician[5]. Rita G. Harper is honoured with the term Harper's syndrome, which is frequently used for describing Seckel syndrome[6].

Seckel first identified the condition in 1960 based on two individuals he had seen in Chicago and 13 instances of nanocephalic dwarfs that had been documented in the literature for more than 200 years[7]. Seckel's mentioned patient's skeleton is currently on display in the Royal College of Surgeons in London. The phrase "bird-headed dwarf" was first used by Rudolf Virchow to describe proportional dwarfism in conjunction with other features like low birth weight, micrognathia, a marked prominence of the nose, and mental disability[8].

Both men and women are equally affected by Seckel syndrome. Seckel syndrome is inherited as an autosomal recessive pattern. Genes on chromosomes 3 and 18 have abnormalities or mutations that lead to this condition. When it comes to recessive conditions, parents who were married to each other in a consanguineous union have a greater probability of having offspring who will either be a carrier or have the condition.This recessive genetic condition develops when each parent passes on two defective genes to the child. The individual becomes a carrier if they receive one functional gene and one non-working gene. Carriers are usually asymptomatic.Depending on the involved gene, Seckel syndrome may be divided into 10 distinct types. Some of these are ATR (SCKL 1), RBBP8 (SCKL 2), CEP 152 (SCKL5), CEP 63 (SCKL 6), NINI (SCKL 7) and ATRIP (SCKL 8). These genes are involved in maintaining genomic stability[9].

This syndrome is characterized by intrauterine growth retardation leading to low birth weight and postnatal dwarfism due to postnatal growth retardation, with severe microcephaly, narrow face, “bird-headed” appearance (beak-like protrusion of nose, receding forehead, prominent eyes, and micrognathia), and intellectual disability (In some cases, the intelligent quotient (IQ) can be significantly less than 50)[10]. Various other facial and skeletal abnormalities noted are large and abnormal eyes, low-set ears with hypoplastic ear lobules, premature closure of cranial sutures, fifth finger clinodactyly (permanent deviation or deflection of one or more

fingers), dislocation of radial heads (pelvis and elbows), and 11 pairs of ribs in some cases[11]. Other features like blood disorders like pancytopenia, anaemia, and acute myeloid leukaemia (less than 25% of the Seckel syndrome patients may exhibit blood cancer), presence of large clitoris in females or Cryptorchidism or undescended testis in males, presence of simian crease, which is a solitary and deep line present on the palm that forms during the development of an embryo might be seen. Defective hypoplastic enamel, tooth crowding, late exfoliation of primary teeth, and delayed eruption of permanent teeth are the major dental changes associated with this illness. Another possibility is a high-arched palate.

II. Case Presentation

A 9-year-old female patient presented to the Department of Pediatric and Preventive Dentistry with a complaint of swelling in the lower left back teeth region for a few weeks. The swelling was initially small but gradually increased in size.

The patient had peculiar dental and craniofacial characteristics, which prompted a more thorough investigation of the case. The patient was identified as a known case of Seckel Syndrome. She was born to consanguineously wed parents. The patient was born preterm and had a low birth weight of about 1.9 kg. From birth onwards, her head was small in shape and size and it became more noticeable as she grew. There was no history of the same problems, other congenital anomalies, or old paternal age in the family. General examination revealed short stature, microcephaly, abnormal speech, and intellectual disability.

Extraoral examination revealed that she has abnormal facies with low set dysplastic ears, cachexia, bird like features of face that include receding forehead, beak like nose, convex nasal bridge, micrognathia, large eyes and narrow face (Figure 1, 2, 3). Other features like clinodactyly of fifth finger of hand and hyper keratinised skin were also present (Figure 4).

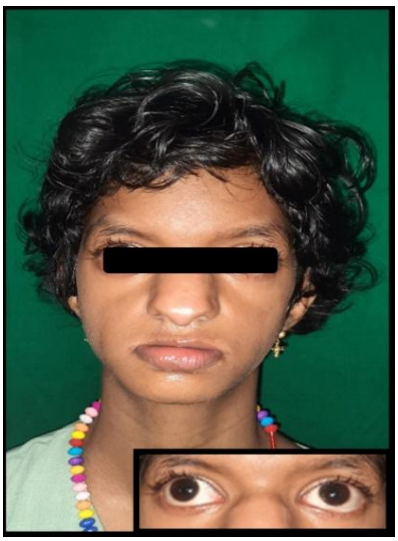

FIGURE 1: Extraoral features of the patient: front view

The image in the inset shows large eyes

FIGURE 2: Profile view of the patient

Arrow shows low set dysplastic ears

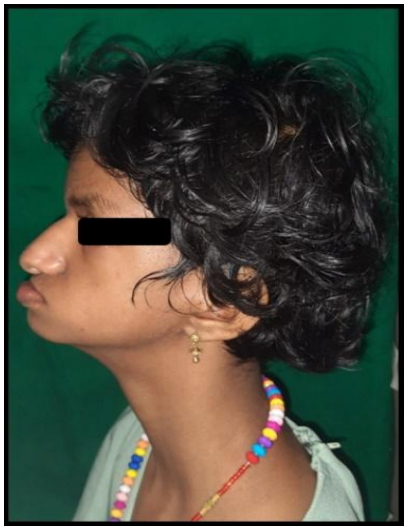

FIGURE 3: Left profile view of the patient

Bird like features: Receding forehead, convex nasal bridge, micrognathia

FIGURE 4: Clinodactyly of fifth finger of hand

Clinically when the patient opened the mouth a swelling could be appreciated on the left side of face(Figure 5). On intraoral examination a swelling of size 1x2 cm was seen obliterating the lower left vestibule near the first and second primary molar region. These molars were grossly decayed (Figure 6).

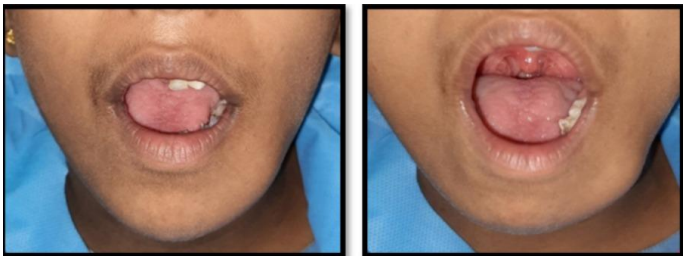

Figure 5: Extra oral swelling found on opening mouth

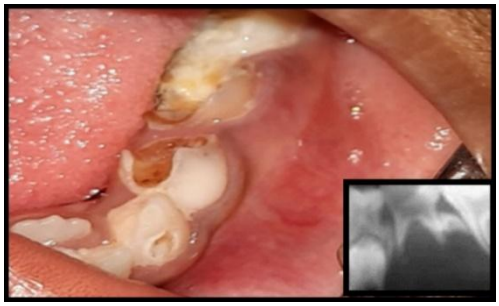

FIGURE 6: Intraoral view of left side of lower arch showing carious teeth and vestibular obliteration due to swelling. The image in the inset shows radiolucency surrounding the primary molars

On intraoral examinationdefective hypoplastic enamel, delayed exfoliation of primary teeth as well as eruption of permanent teeth were evident (Figure 7, 8).

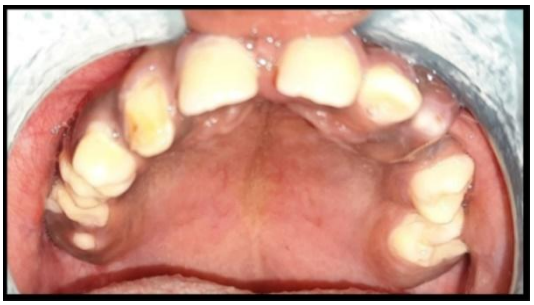

FIGURE 7: Maxillary occlusal view

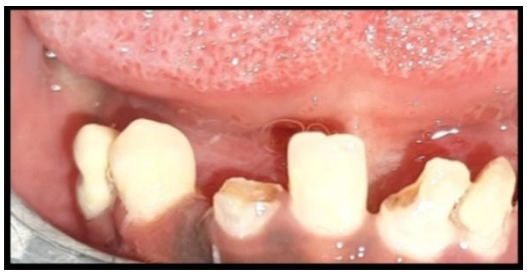

FIGURE 8: Mandibular teeth showing defective hypoplastic enamel anddelayed exfoliation of primary teeth

The patient was not cooperative for taking orthopantomogram, hence intra oral periapical radiograph of lower left first and second molar region was taken. The radiograph depicted a cystic radiolucency surrounding the roots of molars.

For the child cyst enucleation under local anesthesia was carried out. The lower left primary molars were extracted (Figure 9). The cystic contents were sent for histologic examination and was diagnosed as Radicular cyst.

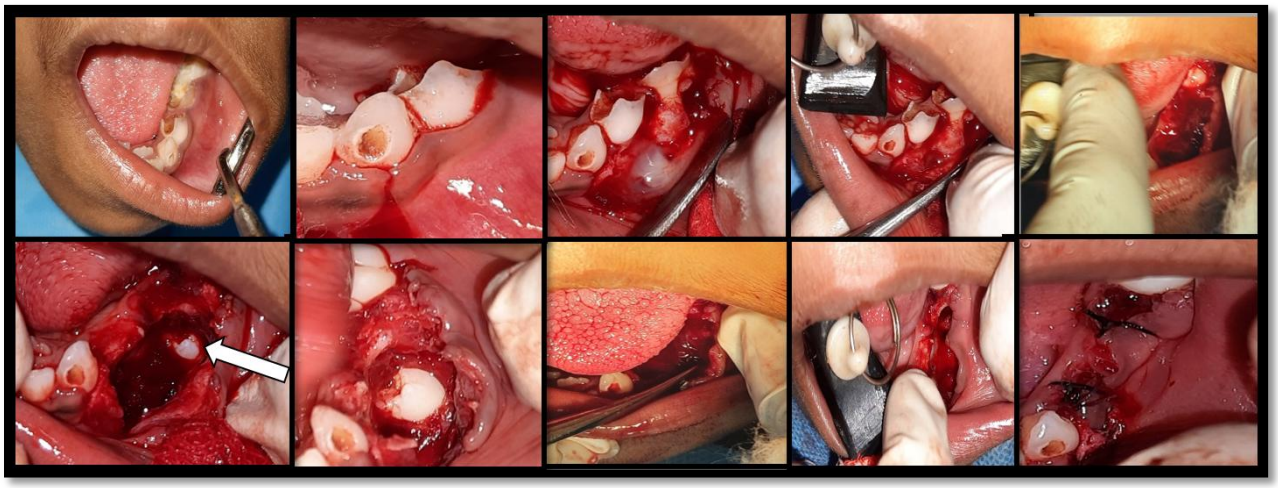

FIGURE 9: Enucleation of Radicular Cyst and suturing the site

In the postoperative period a full mouth supra gingival scaling was carried out. As the grossly cavitated teeth were extracted, space maintainer was advised. Topical fluoride application, advice on maintaining good dental hygiene at home, and nutrition counselling were all performed. Patient is now in maintenance phase with special emphasis on preventive and interceptive dental management of developing malocclusion.

III. Discussion

The prevalence of genetic diseases worries dentists in terms of effectively identifying and treating these patients. Ultrasound scanning in the 2nd trimester, genetic family history, brain imaging of fetus and molecular genetic testing can help diagnose Seckel syndrome. However, in the majority of instances, a complete clinical assessment and specialised testing, including the molecular genetic test, are used to confirm the diagnosis at birth or during the first few months of infancy. A correct diagnosis should be made before treatment begins immediately after delivery. Since it affects various systems, a multidisciplinary (cerebral, dental, ophthalmic, and orthopedic) approach is essential. Both surgical and supportive treatment are needed. Surgical digit correction and cosmetic face remodeling can be done to enhance both function and appearance. To fully rehabilitate the child in society, early hearing optimization with potential hearing aids, psychological counselling, speech therapy, and genetic counselling are necessary. In patients with Seckel syndrome, severe skeletal malocclusion can be corrected by orthodontic treatment and orthognathic surgery. Oral hygiene maintenance is difficult in these patients because of intellectual disability, hand deformities and malocclusion. The latest electric toothbrushes and fluoride mouthwashes might make the process simpler. Dental problems can be avoided with professional care, such as routine dental check-ups, prophylactic dental hygiene, fluoride treatments, and tooth sealants.

IV. Conclusion

Seckel syndrome is a relatively uncommon condition caused by gene abnormalities that cause chromosomal instability and a variety of hematological, hormonal, and physical problems.To avoid a poor prognosis and any related consequences like mortality brought on by cardiac insufficiency, arrhythmias, or pituitary insufficiency, various manifestations of the syndrome should be examined and recognized as early as feasible. Genetic counselling is mostly recommended to prevent this type of disorders[12]. Due to the multiple systemic involvements in Seckel syndrome, a single approach is not enough. The treatment is mainly supportive and based on the presentation of symptoms. Thus, a multidisciplinary team is needed for oral and maxillofacial management.

References

[1]. Seckel Syndrome | Causes | Symptoms | Diagnosis | Treatment [Internet]. 2022 [cited 2023 Feb 17]. Available from: https://www.icliniq.com/articles/genetic-disorders/seckel-syndrome
[2]. D'Angelo,V., Ceddia, A., Zelante, L. *et al.* Multiple intracranial aneurysms in a patient with Seckel syndrome. Child's Nerv Syst 14, 82–84 (1998) https://doi.org/10.1007/s003810050181
[3]. Wynbrandt J, Ludman MD. The Encyclopedia of Genetic Disorders and Birth Defects. Infobase Publishing; 2010. 705 p.

[4]. Harsha Vardhan BG, Muthu MS, Saraswathi K, Koteeswaran D. Bird-Headed Dwarf of Seckel. Journal of Indian Society of Pedodontics and Preventive Dentistry. 2007 Jan;25(Supplement):S8. PMID: 17921644
[5]. Børglum AD, Balslev T, Haagerup A, Birkebaek N, Binderup H, Kruse TA, et al. A new locus for Seckel syndrome on chromosome 18p11.31-q11.2. Eur J Hum Genet. 2001 Oct;9(10):753–7.
[6]. Harper RG, Orti E, Baker RK. Bird-beaded dwarfs (Seckel's syndrome). A familial pattern of developmental, dental, skeletal, genital, and central nervous system anomalies. J Pediatr. 1967 May;70(5):799–804.
[7]. Goss CM. Bird-headed dwarfs — Studies in developmental anthropology including human proportions. By Helmut P. G. Seckel.Charles C Thomas, Publisher, Springfield. 1960. The Anatomical Record. 1961;139(1):92–92.
[8]. Virchow R. Zwergenkind. Zeitschrift fur Ethnologic. Berlin 1882; 14: 215
[9]. Seckel Syndrome - Causes, Symptoms, Diagnosis, Treatment, Health Tips [Internet]. Medindia. [cited 2023 Feb 17]. Available from: https://www.medindia.net/patients/patientinfo/seckel-syndrome.htm
[10]. Ramalingam K, Kaliyamurthy SD, Govindarajan M, Swathi S. Seckel syndrome: A report of a case. Journal of Indian Society of Pedodontics and Preventive Dentistry. 2012 Sep;30(3):258.
[11]. Sudarshan R, Annigeri RG, Vijayabala GS. Bird-Headed Dwarfism (Seckel Syndrome) – A Rare Case Report -. TAF Preventive Medicine Bulletin. 2013;12(2):209–12.
[12]. F. Chentli, S. Belahcene, S. Azzoug. Seckel's-Like Syndrome with Primordial Dwarfism, Marked Mental Retardation, and Severe Heart Malformation. Ibnosina Journal of Medicine and Biomedical Sciences2013; 05(06): 339-344. DOI: 10.4103/1947-489X.210567

Dr Shruti Sudarsanan, et. al. "A Rare Case Report of Seckel Syndrome: Dentofacial Management." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 22(3), 2023, pp. 10-15.