eISSN 2255-0569

CASE REPORT

Seckel syndrome associated dental anomaly; case report

Anomalía dental asociada al síndrome de Seckel; reporte de un caso

Soraya Bakhshani[1], Seyed Hossein Hosseini Zarch[2], Mahrokh Imani Moghadam[2], Atsa Pakfetrat[3]

1. Department of Oral and Maxillofacial Radiology, Faculty of Dentistry, Zahedan University of Medical Sciences, Zahedan, Iran.
2. Department of Oral and Maxillofacial Radiology, Mashhad University of Medical Sciences, Mashhad, Iran.
3. Department of Oral Medicine, Mashhad University of Medical Sciences, Mashhad, Iran

Corresponding author
Soraya Bakhshani
Department of Oral and Maxillofacial Radiology, Faculty of Dentistry, Zahedan University of Medical Sciences, Zahedan, Iran
E-mail: bakhshani69@gmail.com

Received: 7 - VI - 2021
Accepted: 22 - VI - 2021

doi: 10.3306/AJHS.2021.36.03.136

Abstract

Seckel syndrome is a rare autosomal recessive with severe growth retardation, bird-headed profile, and microcephaly. It is characterized by short stature, skeletal defects, mental retardation, and characteristic facial features such as microcephaly, micrognathia, and a bird-head appearance. Dental findings include hypodontia, enamel hypoplasia, crowding, and Class II malocclusion. Craniofacial features may include facial asymmetry, down-slanting palpebral fissures, lobeless ears, and dental abnormalities, including enamel hypoplasia, hypodontia, and microdontia in the patients with SS may be at high risk of developing acute myeloid leukemia. The purpose of the paper is to report a case of the seckel syndrome with a dental anomaly.

Keywords: Seckel syndrome, dental, anomaly, case report.

Resumen

El síndrome de Seckel (SS) es una rara enfermedad autosómica recesiva que cursa con un grave retraso del crecimiento, un perfil de cabeza de pájaro y microcefalia. Se caracteriza por baja estatura, defectos esqueléticos, retraso mental y rasgos faciales característicos como microcefalia, micrognatia y apariencia de cabeza de pájaro. Los hallazgos dentales incluyen hipodoncia, hipoplasia del esmalte, apiñamiento y maloclusión de clase II. Características craneofaciales pueden incluir asimetría facial, fisuras palpebrales inclinadas hacia abajo, orejas sin lóbulos y anormalidades dentales, incluyendo hipoplasia del esmalte, la hipodoncia y la microdoncia. Los pacientes con SS pueden tener un alto riesgo de desarrollar leucemia mieloide aguda. El propósito de este trabajo es informar de un caso de síndrome de Seckel con una anomalía dental.

Palabras clave: Síndrome de Seckel, dental, anomalía, reporte de caso.

Introduction

Seckel syndrome is an autosomal recessive condition without any sex predilection, with an incidence reported of 1:10,000 live-born children[1,2].

The syndrome is characterized by severe intrauterine growth retardation, severe short stature, severe microcephaly, bird-headed profile, receding chin and forehead, sizeable beaked nose, mental retardation, and other congenital anomalies. 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[3].

Polyarthritis nodosa affects multiple organ systems, most commonly kidneys, gastrointestinal tract, nervous system, muscles, and soft tissue[4].

In one-half of cases, head circumference is more retarded than height, while it is as retarded as height for the remainder. Other systemic manifestations associated with SS include Fanconi anemia, leukemia, chronic nephritis, dysgenesis of the cerebral cortex, corpus callosum, and a vast spectrum of skeletal defects[7].

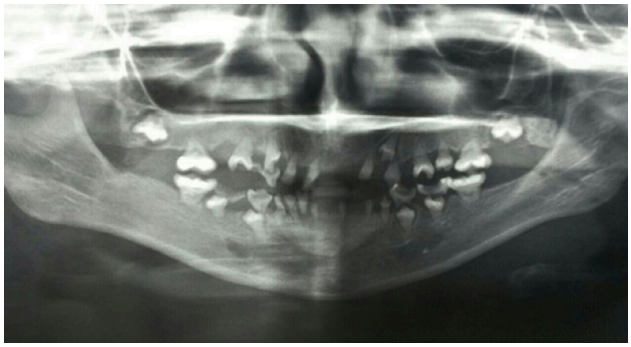
• Small brain with small gyri. The cerebral hemispheres' volume can be reduced to one-third of routine, particularly in the cerebral cortex[8]. Dental findings include hypodontia, enamel hypoplasia, crowding, and Class II malocclusion[9].

This paper's purpose was to report the case of a female patient with Seckel syndrome and describe her dental manifestations.

Case report

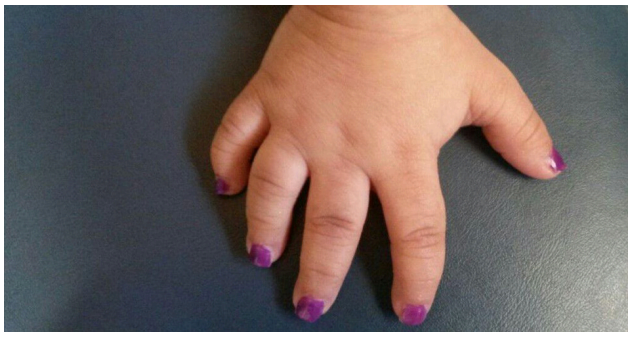
A nine-year-old female child reported to Oral Medicine and Radiology department with a chief complaint of noneruptive teeth and micro dent (**Figure 1**). History revealed that a child with severe growth retardation.

Figure 1: Panoramic view of noneruptive teeth and micro dent were presented in a child.



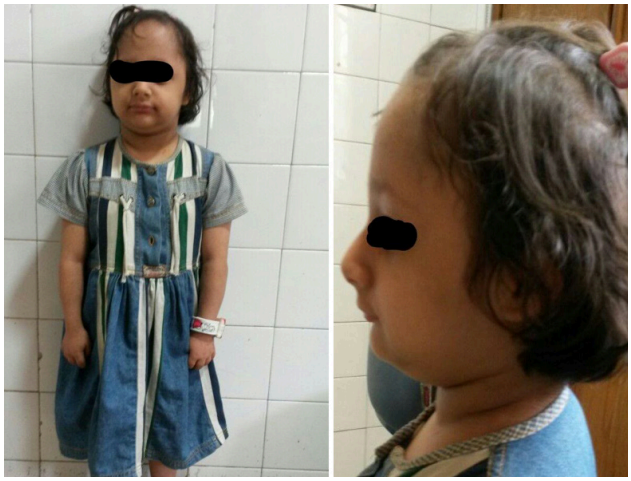
The patient had proportional short stature, weight, height, and head circumference below the third percentile. On physical examination, she had a short finger (**Figure 2**).

Figure 2: A girl case with a short finger.



The patient's general appearance was characterized by a small, prominent forehead, slightly beaked nose, midface hypoplasia, hypertelorism, high hair lin whit fin hair, and eyebrow (**Figure 3**).

Figure 3: The general appearance of a case patient.



Investigation of the orthopantomogram and panoramic radiographs (**Figure 1**) revealed that maxillary permanent left central incisors and right second premolar and mandibular permanent second molar were congenitally missing. The teeth appeared to have enamel hyperplasia whit short roots, partially obliterated pulp. The permanent first molar had a bulbous crown.

On laboratory examinations, results of complete blood revealed low-level hemoglobin (10.9 g/dl, normal range:12-15 g/dl) and MCV(65/9, normal range 77-95), while the levels RDWC (18/3, normal range 11-14) and platelet count (588 1000/*µl*, normal range 150-450 1000/*µl*), D.H.E.A So4 (311 micg/dl, normal range 2/8-85/2). The range of 25OH VitD was insufficient.

Discussion

Seckel syndrome (Online Mendelian Inheritance in Man database Number 210600) is a rare form of primordial dwarfism, which was initially described in 1960.[10]

The syndrome as severe intrauterine growth retardation, severe short stature, severe microcephaly, bird-headed profile, receding chin and forehead, sizeable beaked nose, mental retardation, and other congenital anomalies. 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[3].

Presented herein is a child with classic Seckel phenotype, who had both an open and a closed-lip schizencephaly, resulting in global developmental delay and bilateral sensorineural deafnes[11].

Synostosis of cranial sutures occurs in approximately 50%. Other craniofacial features may include facial asymmetry, down-slanting palpebral fissures, lobeless ears 12 Dental abnormalities include enamel hypoplasia, hypodontia, microdontia, taurodontic root morphology. Several cases have demonstrated a high-arched palate, and cleft palate has also been reported[10].

This patient had enamel hypoplasia, hypodontia, and microdontia, but Cleft palate was not observed in the present cases. Regen reported the patient with Seckel syndrome type II and described her oriental manifestations. She presented interesting dental findings, including gingival hyperplasia, recession and ulceration, significant crowding, and early exfoliation of the primary dentition with the permanent dentition's accelerated eruption.[9]

The dentition has been described sporadically in SCKL, focusing on enamel hypoplasia malocclusion microdontia, dentin dysplasia, and taurodontism[13].

De Coster reported a 14-year-old boy is presented with brain hypoplasia, pachygyria, hydrocephaly, enamel hypoplasia and root dysplasia in the temporary dentition, and oligodontia, severe microdontia, and delayed eruption of the permanent dentition[13].

About 170 cases of Seckel syndrome have been reported worldwide, and there is considerable heterogeneity in their clinical characteristic[11].

Hematopoietic disorders have been reported in approximately 15% of patients with SS[14].

The authors suggested that the patients with SS may be at high risk of developing acute myeloid leukemia. However, the risk of chronic myeloid leukemia was noted following the aspiration biopsies of the bone marrow of both patients, and the relevant specialists planned routine controls of the patients[10].

Kilic et al. reported a case whit low-level vitD and osteomalacia[15]. Our patient had 25OH VitD insufficient. Vitamin D plays an essential role in skeletal development, bone health maintenance, and neuromuscular functioning[16]. Clinical features include growth failure, hypotonia, rachitic bones, and enamel hypoplasia[17].

The craniofacial features of Seckel syndrome allow differentiation from other syndromes of growth deficiency with microcephaly, such as *Dubowitz syndrome, fetal alcohol syndrome, trisomy 18, de Lange syndrome, Bloom syndrome, Nijmegen breakage syndrome*, and Fanconi syndrome[12] also SS should be differentiated from closely resembling Cockayne syndrome, progeria, *Hallermann-Streiff* syndrome (HSS) Dyggve-Melchior-Clausen (DMC) syndrome[3].

- Fanconi anemia is clinically defined by pancytopenia in the first decade of life and complicated by leukemia, pigmentary changes in the skin, cardiac, kidney, and limb (radius aplasia) malformations. Affected adults are also at high risk for non-hematologic malignancies. Mild microcephaly (often without intellectual disability) is observed in 10%-25% of individuals[8].

- Cockayne's syndrome is a rare, autosomal recessive disorder characterized clinically by cachectic dwarfism, cutaneous photosensitivity, loss of adipose tissue, mental retardation, skeletal and neurological abnormalities, and pigmentary degeneration of the retina[18].

While progeria is a rare premature aging syndrome characterized by retarded physical development, abnormal facies, skeletal abnormalities, and early onset of scleroderma[19].

Conclusion

The knowledge of symptoms and risk factors of Seckel syndrome is essential For early intervention and dental problems prevention.

Acknowledgments

The authors would like to thank the Department of Oral and Maxillofacial Radiology, Faculty of Dentistry, Mashhad University of Medical Sciences, Mashhad, Iran, for the clinical supports.

References

1. D'Angelo VA, Ceddia AM, Zelante L, Florio patient with Seckel syndrome. Child Nerv Syst 1998; 14: 82-84.

2. Carfagnini F, Tani G, Ambrosetto P. MR findings in Seckel's syndrome: report of a case. Pediatr Radiol 1999; 29: 849-850

3. Sisodia R, Sundar Raj R.K, Goel V. Seckel syndrome: A rare case report. J Indian Soc Pedod Prev Dent 2014;32:160-3

4. Oran I, Memis A, Parildar M, Yunten N. Multiple intracranial aneurysms in polyarteritis nodosa: MRI and angiography. Neuroradiol 1999;41:436-439

5. Bakkaloglu SA, Ekim M, Tumer N, Tulunay. O, Ozer T. Severe renal impairment in the case of classic polyarteritis nodosa. Pediatr Nephrol 2001;16:148-150

6. Brogan PA, Davies R, Gordon I, Dillon MJ Renal angiography in children with polyarteritis nodosa. Pediatr Nephrol 2002;17:277-283.

7. Shanske A, Caride DG, Menasse-Palmer L, Bogdanow A, Marion RW. Central nervous system anomalies in Seckel Syndrome: Report of a new family and review of the literature. Am J Med Genet 1997;70:155-8

8. Verloes A, Drunat S, Pharm D, Gressens P, Passemard S. Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders. GeneReviews October 31, 2013.

9. Regen A1, Nelson LP, Woo SB. Dental manifestations associated with Seckel syndrome type II: a case report. Pediatr Dent 2010;32(5):445-50.

10. Kirzioglu Z, Ozay Erturk M.S, Erdogan Y. Craniofacial morphology and dental findings of seckel syndrome: case reports of two siblings. J Int Dent Med Res 2011;4(3):139-144.

11. Ramasamy C, Satheesh S, Selvaraj R . Seckel Syndrome with Severe Sinus Bradycardia. Indian J Pediatr 2015;82(3):292-293.

12. Seymen F, Tuna B, Kayserili H. Seckel syndrome: report of a case. J Clin Pediatr Dent 2002; 26(3): 305-310

13. De Coster P.J, Verbeeck R.M.H, Holthaus V, MartensL.C, Vral A. Seckel syndrome associated with oligodontia, microdontia, enamel hypoplasia, delayed eruption, and dentin dysmineralization: a new variant. J Oral Pathol Med 2006;35:639-41

14. Chanan-Khan A, Holkova B, Perle MA, Reich E, Wu CD, Inghirami G, Takeshita K. T-cell clonality and myelodysplasia without chromosomal fragility in a patient with features of Seckel syndrome. Haematologica. 2003;88(5):ECR14

15. Kilic A, Külcü Çakmak S, Tuncali T, Koz O, Ozhamamci E, Yasun O, Artuz F. Seckel syndrome with cutaneous pigmentary changes: two siblings and a review of the literature. Postepy Dermatologii i Alergologii 2015;6:470-474.

16. Bordelon P, Ghetu M.V, Langan R, MD. Recognition and Management of Vitamin D Deficiecy. Am Family Phys 2009;80(8):841-846

17. Forestier I.B ,Berdal A, Vinckier F, De ravel T, Pierre Fryns J, Verloes A. The genetic basis of inherited anomalies of the teeth. Part 2: Syndromes with significant dental involvement. Eur J Med Gen 2008;51:383-408.

18. Gaddam D, Thakur MS, Krothapalli N, Kaniti S. Dental Management of a 14-Year-Old with Cockayne Syndrome under General Anesthesia. Case Report Den 2014;2014:1-3.

19. Sowmiya R, Prabhavathy D, Jayakumar S. Progeria in siblings: A rare case report. Indian J Dermatol 2011;56:581-2.