Seckel syndrome: report of a case

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An interesting case of a seven years old boy with a combination of clinical, genetic, radiological, pathologic and dental findings is presented in view of Seckel syndrome literature. General appearance of the patient was characterized by small forehead, posteriorly slanted ears, slightly beaked nose, midfacial hypoplasia very stunted stature with microcephaly. He had borderline mental retardation with normal motor development.

Class II dentoskeletal pattern with mild overjet and open bite, congenitally missing permanent teeth, microdontia, enamel hypoplasia, taurodontism and dentinal dysplasia was observed according to the clinical and radiographic examination. In conclusion, Seckel syndrome is not encountered routinely in dental clinics, this case illustrates the importance of dental care in such a rare condition.
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INTRODUCTION

Seckel syndrome (SS), is a rare ((1:10000) constellation of malformations, presumably inherited as an autosomal recessive trait. Seckel Syndrome is a heterogeneous form of primordial dwarfism. Its ethiopatogenesis remains unclear.[1,2]

The primary characteristic features of SS include: intrauterine and postnatal growth deficiency, severe microcephaly, orofacial dysmorphism with characteristic "Bird-headed" appearance, prominent beaked nose, large eyes, dysplastic ears, micrognathia and variable mental retardation, which becomes prominent after several months.[1,3-9]

Seckel syndrome is different accompanying multiple anomalies including: dwarfism, club foot and multiple skeletal malformations. In addition to the characteristic craniofacial dysmorphism and skeletal defects, abnormalities have been described in the cardiovascular, hematopoietic, endocrine and central nervous systems.[4] Mental deficiency is present in half of the cases with levels of IQ below 50.[8]

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Moderate mental deficiency has been observed in some instances. Head circumference is the most retarded parameter.[7] The cerebellum is small with a simple primitive convolutional pattern resembling that of a chimpanzee. Though they tend to be friendly and pleasant, these patients are often hyperkinetic and easily distracted. Poor joint development may be evident by dislocations of the hip, elbows or both and by later development of scoliosis and kyphosis. Survival till the age of 75 years has been recorded.[8]

The craniofacial features of Seckel syndrome allows it differentiation from other syndromes of growth deficiency with microcephaly such as Dubowitz Syndrome, fetal alcohol syndrome, trisomy 18 syndrome, de Lange syndrome, Bloom syndrome and Fanconi syndrome.[4,5]

Occasional abnormalities are facial asymmetry, strabismus, sparse hair, scoliosis, hypoplasic external genitalia, hypoplastic anemia, chromosome breakage, highly arched or cleft palate, receding chin, partial anadontia, enamel hypoplasia, crowded teeth and class II malocclusion, limb anomalies include: clinodactyly of the fifth finger, abnormal finger flexion creases and hip dysplasia. Patients may also have radial dislocation and intracranial aneurysmus.[3,4,8,10-11]

In addition to delayed osseous maturation, the phalanges exhibit ivory and cone-shaped epiphyses in the proximal phalanges. The radiological findings also include relatively small carpals and marked disharmonic bone maturation between carpals and phalanges, between individual carpals and from side to side; relatively normal or increased cortical thickness of the metacarpals. Synostosis of cranial sutures occurs in approximately 50% of the cases.[7,12,13]

The male to female sex ratio is 9:11 and both sexes are equally severely affected. All parents have been

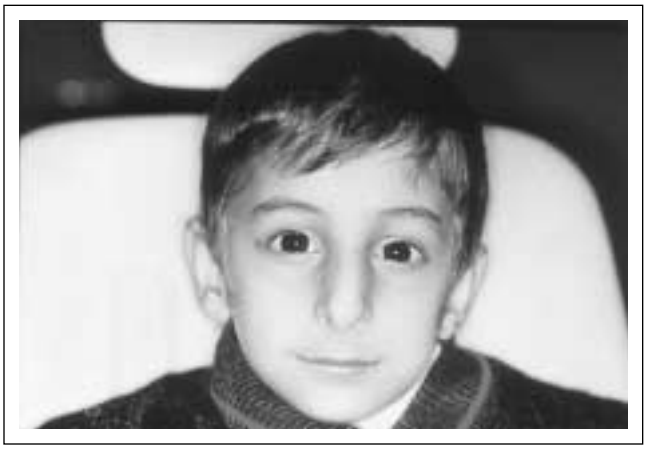

Figure 1. Frontal view of the patient with Seckel syndrome.

Figure 2. Lateral view of the patient with Seckel syndrome.

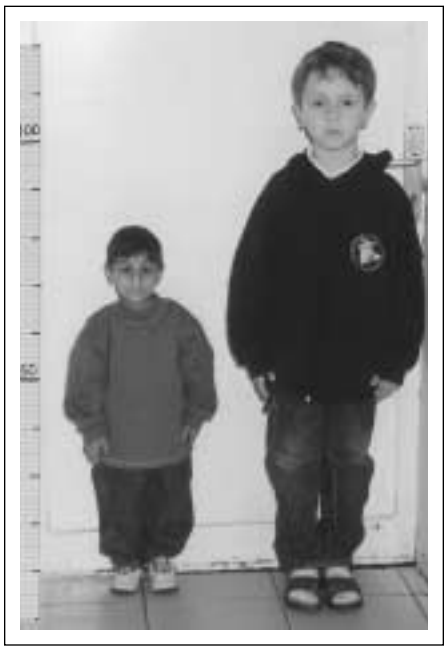

Figure 3. The patient compared to normal child of the same age.

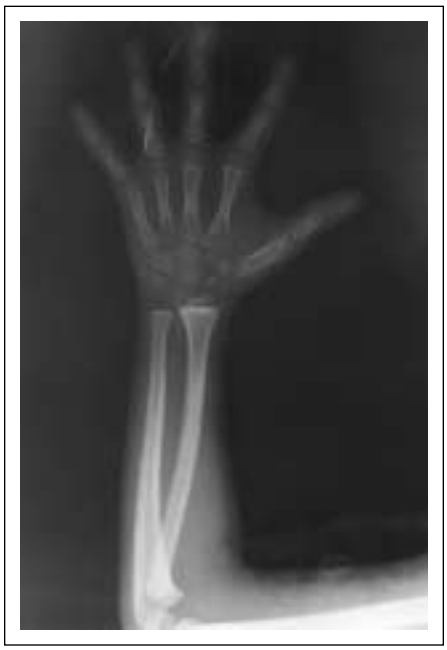

Figure 4. Hand-wrist radiograph of the patient showing five years six month old skeletal maturation.

normal and consanguinity has been reported in many cases.[7]

The aim of the paper is to describe the dental care provided for a seven year-old child with Seckel syndrome.

CASE

Seven years old boy with the diagnosis of Seckel syndrome was referred to Department of Pedodontics, Faculty of Dentistry for the dental management of his dental care.

He was born to non-consanguineous parents and has a healthy 17 years old sister.

He was prematurely born at 7.5 months with a birth weight of 800gm and a height of 35cm head circumference of 25cm. He has been evaluated at 7 months of age at the Pediatric Department, Medical Genetics Division for growth and development delay, microcephaly and facial anomalies and was diagnosed as Seckel syndrome.

Physical examination showed a typical "bird-like" facial appearance, very stunted stature with microcephaly. His dysmorphic features were small forehead, posteriorly slanted ears, slightly beaked nose and midfacial hypoplasia (Figure 1,2).

At 7 years 8 months of age, his weight was 7kg, height was 78cm. and head circumference was 40.5cm., all very below the 3rd centiles (Figure 3). He has a characteristic stance with flexion at the hips and pronation of the forearms, and has borderline mental retardation and with normal motor development.

His bone age was 5 year 6 months when his chronological age was 7 year 10 month old (Figure 4). Intraoral clinical examination revealed microdontia in primary teeth however very mild microdontia in permanent incisors. Enamel hypoplasia and diastemas were observed between the permanent central incisors (Figure 5).

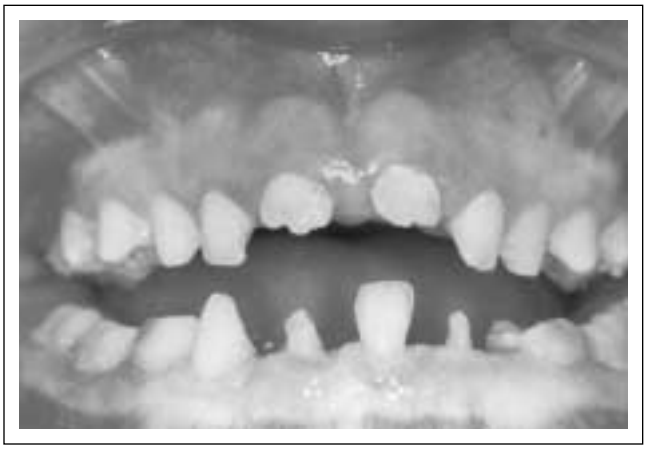

Figure 5. Intraoral view showing the microdontia with enamel hypoplasia and diastemas between the teeth.

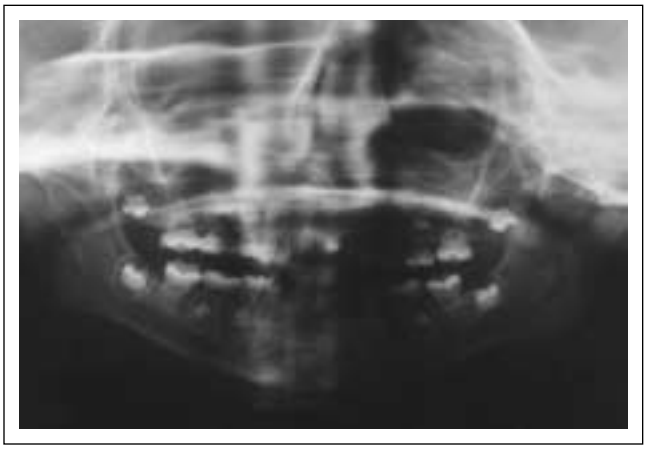

Figure 6. Panoramic radiograph showing congenitally missing maxillary permanent lateral incisors and mandibular permanent central incisors.

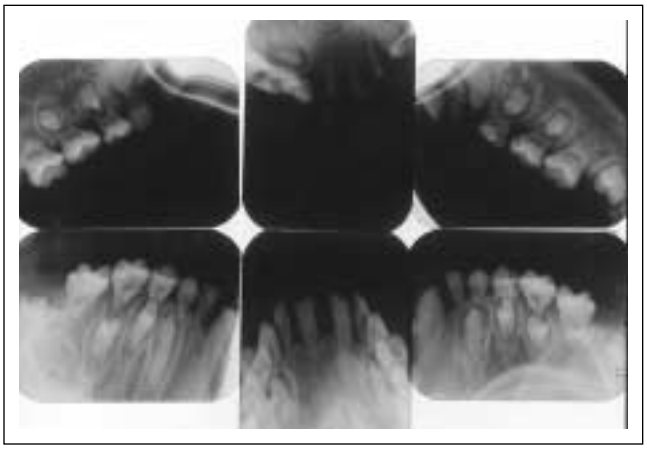

Figure 7. Periapical radiographs showing dentinal dysplasia and taurodontism.

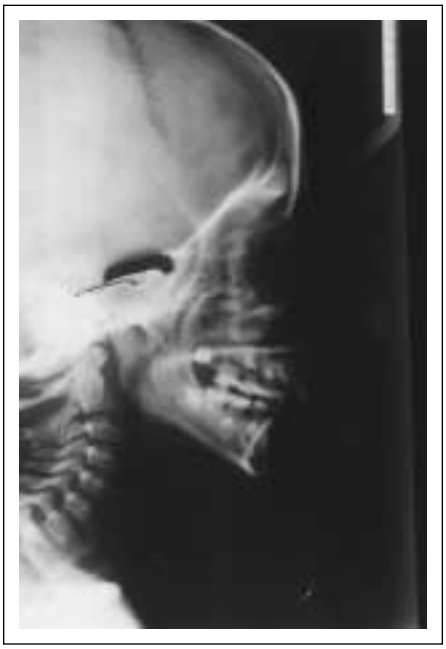

Figure 8. Lateral cephalogram showing that the suture between the frontal and parietal fontanels.

Although there was no clefting of lip and palate, the palate was highly arched and narrow, which necessitated speech therapy. No abnormal bony undercuts or periodontal disease was observed.

Investigation of the orthopantomogram (Figure 6) and periapical radiographs (Figure 7) revealed that maxillary permanent lateral incisors and mandibular permanent central incisors were congenitally missing. Dentinal dysplasia and taurodontism were observed according to radiographic investigation.

Rickett's cephalometric analysis revealed mesofacial (facial) pattern which shows Class II malocclusion due to upper and lower molar, mild overjet and skeletal Class II pattern due to a long anterior cranial base as horizontally and open bite as vertically (Figures 8 to 10).

Dental treatment of the patient consisted of filling the carious mandibular left deciduous molars and fissure sealants were placed on the right and left maxillary and mandibular first permanent molars. The physiologically exfoliated first primary molar tooth was used for histopathological investigation. In decalcified sections dentine including horizontal and longitudinal dentine trabecules can be observed. In the pulp tissue lymphocytes and plasma cells could be observed (Figure 11A-B). The patient was consulted for routine dental care, including oral hygiene instructions, dietary advice and professional fluoride application.

DISCUSSION

Seckel syndrome is a form of primordial dwarfism with a set of primary diagnostic criteria, however, there are also several secondary characteristics that are less well defined. There appear to be several sub-sets of this type of dwarfism, and the literature reports show some controversy in the application of this diagnosis.[2]

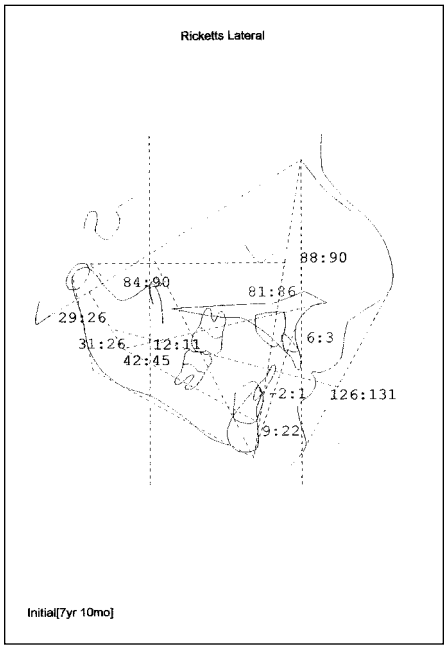

Figure 9. Tracing and landmarks of Rickett's cephalometric analysis

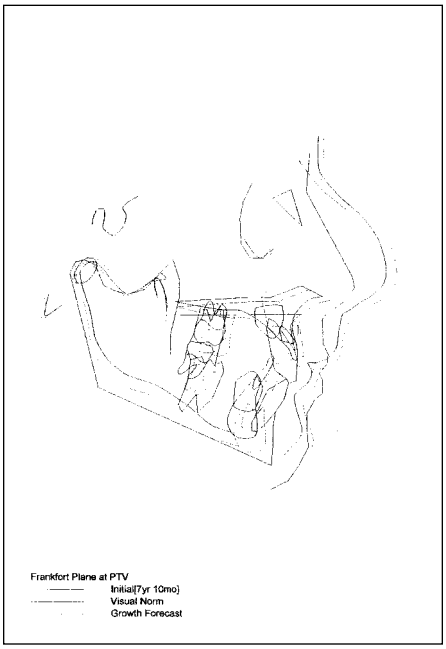

Figure 10. Cephalometric analysis showing the initial, visual norms and growth forecast.

According to the radiological abnormalities, three different forms of the syndrome have been described. This condition was given two names: "bird-headed dwarfism" and "nanocephalic dwarfism". In addition to dwarfism of "low birth weight" type, the features are small head and characteristic facial appearance. An interesting case of a seven years old boy with a combination of clinical, genetic, radiological, pathologic and dental findings is presented in view of Seckel syndrome literature.[1]

The syndrome is assumed to have an autosomal-recessive pattern of inheritance. The primary diagnostic features are severe intrauterine growth retardation, microcephaly, characteristic "bird-like" facies and mental retardation.[2]

Nihill *et al.* reported a 21 years old case with possible diagnosis of Seckel syndrome, who revealed micrognathia, microdontia, severe bony undercuts who sought treatment at a faculty clinic. Treatment included extractions, periodontal therapy, retention of premolars with sealants and over dentures with resilient soft liners.[14]

Although, Seckel Syndrome is seldomly encountered, it shares similarities with many other disorders. There have been several reports of associated hematological abnormalities and chromosomal breakage, findings suggestive of Fanconi anemia.[3]

Abou-Zahr *et al.,*[3] compared two growth profiles of lymphoblastoid cells, and reported that cross link sensitivity comparable to Fanconi anemia is not a uniform finding in patients with Seckel syndrome.

Hayani *et al.*,[15] reported a female patient with Seckel syndrome, who developed acute myeloid leukemia at the age of 26 years. The authors suggested that patients with Seckel syndrome may be at high risk of developing acute myeloid leukemia.

Butler *et al.*,[10] reported two patients with Seckel syndrome. One patient was a 12 years old female, who had pancytopenia, which is occasionally seen in patients with Seckel syndrome and is also feature of Fanconi anemia, a well-recognised autosomal recessive dwarfism syndrome with increased chromosomal breakage. They suggested that there maybe a subgroup of Seckel syndrome patients, which had chromosome instability and hematological problems.

Low birth weight accompanied by delay in weight and stature, microcephaly with the typical bird head and considerable mental retardation are sufficient for the diagnosis.[11] The patient presented here, shows all the manifestations of this syndrome.

Sorof *et al.*,[16] reported a 19 years old male with Seckel syndrome, who presented with malignant hypertension associated with hypertensive nephrosclerosis, dilated cardiomyopathy and a ruptured cerebral artery aneurysm. They speculated that the Seckel syndrome cases may particularly be prone to hypertensive end-organ injury due to extreme short stature.

Howanietz *et al.*,[17] reported a girl with Seckel syndrome, who had a congenital heart failure. Endocrine surveys revealed decreased growth hormone stimulation.

Some authors questioned if the described syndrome was caused by the use of antiepileptic drug exposure during pregnancy or epilepsy of the mother, or if it was causal.

In near future, linkage studies may hopefully allow the localization and of the gene, which will enable the cloning and identify the underlying pathogenesis, which is responsible for the features of the Seckel syndrome.[1,5]

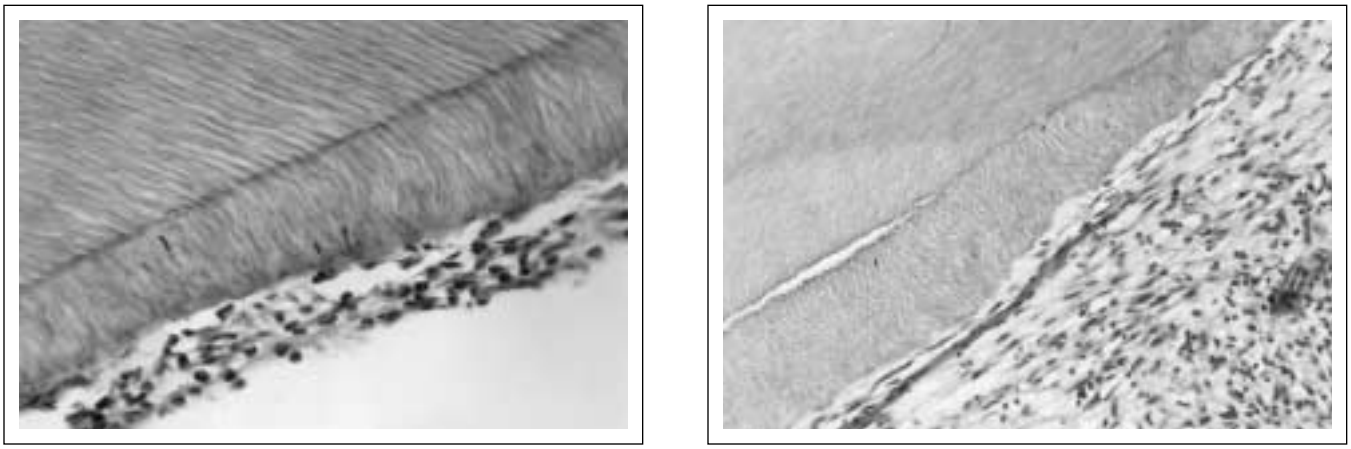

Figure 11(A-B). Histopathological photographs of the first primary molar tooth.

Children affected with Seckel syndrome do have a normal life-span although they often have profound mental and physical deficits. Future pregnancies of Seckel syndrome can be followed by serial ultrasonographies in order to evaluate severe IUGR, microcephaly and prominent facial features.[2,18]

Early intervention, including preventive dental care in this patient would have avoided rampant caries and unnecessary pain. Preventive dental advice, including oral hygiene instructions, diet counselling and the use of fluoride has been given to the patient and his parents. It is essential that the patient be followed up on a regular basis to reduce the risk of subsequent dental disease.

Although Seckel syndrome is not encountered routinely in the dental clinics, this case illustrates the importance of dental care in such a rare condition.

ACKNOWLEDGEMENT

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