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Emmanuel Mazinis[1] and Vasilios Thomaidis[2]

[1]Aristotle University of Thessaloniki
[2]Democritus University of Thrace School of Health Sciences

January 09, 2025

Short Root Anomaly. A rare case of distortion in dental anatomy

Introduction

The short root anomaly (SRA) was first described in 1972 as a developmental anomaly of the dental roots, characterized by a root-to-crown ratio of 1:1 or less (Lind, 1972). The condition may also be associated with a range of other dental anomalies, including hypodontia, microdontia, obliterated pulp chambers, dentin dysplasia type I, taurodontism, dens invaginatus and talon cusp (Puranik et al., 2015). SRA primarily affects the permanent maxillary central incisors and involves other teeth bilaterally, followed by maxillary premolars, maxillary lateral incisors, and mandibular premolars, with the lowest prevalence in canines and molars but rarely affects all permanent teeth (Lind, 1972; Apajalahti et al., 2002). The detection of SRA is often incidental and the prevalence of SRA in any teeth type is estimated to be between 0.6 and 2.4% (Apajalahti et al., 2002; Jakobson, 1973), with a higher incidence observed in females, with a ratio of approximately 1:2.6 to 1:2.7 (Apajalahti et al, 2002; Jakobson, 1973). Despite the short roots, the affected teeth have normal crown morphology (Edwards, 1990, Apajalahti et al., 2002; Puranik et al., 2015) and exhibit normal pulp chambers, root canals, and closed apices, supported by normal periodontal tissues (Lind, 1972; Apajalahti et al., 2002; Puranik et al., 2015). The term 'hereditary idiopathic root malformation' has been proposed for cases not associated with any specific syndrome (Puranik et al., 2015).

Certain syndromes and metabolic disorders also present reduced root length. These include Stevens-Johnson syndrome (De Man, 1979; Gaultier et al., 2009; Yongho et al., 2017), Turner syndrome (Pentinpuro et al., 2013), Fraser syndrome (Kunz et al., 2020), Hallermann-Streiff syndrome (Robotta and Schafer, 2011), and Schimke immuno-osseous dysplasia (Gendronneau et al., 2014). Furthermore, metabolic disorders such as pseudohypoparathyroidism (Gallacher et al., 2018; Hejlesen et al., 2019), hypophosphatasia (Wei et al., 2010), and vitamin D-dependent rickets type I (Zambrano et al., 2003) also present with shortened roots. There is a significant association between maternal or passive smoking during pregnancy and SRA. Children whose mothers smoked were found to be 4.95 times more likely to have SRA (Sagawa, 2021).

Transverse hemimelia is a congenital limb deficiency characterised by the absence of a limb segment, typically involving structures distal to a certain level while the proximal part remains intact. This condition is frequently categorised according to the level of limb absence, with the classification typically based on whether the absence extends to below the elbow (forearm) or below the knee (leg). It is a constituent of a more expansive category of limb deficiencies, designated as terminal transverse limb defects. With regard to the genetic aspects of hemimelia, it typically occurs as a sporadic event and is not generally considered to be of genetic origin (Lenz, 1980). The etiology of hemimelia has been attributed to genetic factors as part of syndromes (Holt-Oram, TAR) as well as to exogenous environmental factors, such as drug intake (thalidomide) (Bermejo-Sánchez et al., 2011). In the majority of cases, the precise aetiology remains undetermined, with no discernible genetic or environmental factors (Bermejo-Sánchez et al., 2011). Furthermore, there is no evidence for familial recurrence (Froster-Iskenius and Baird, 1990). The available data on the prevalence of limb deficiency are limited, with the majority of published articles on this congenital defect comprising single case reports or limited series (Bermejo-Sánchez et al., 2011).

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The purpose of this study is to present a rare case in which both SRA, affecting all permanent teeth, and hemimelia coexist, and to examine the potential factors associated with these developmental disorders.

Case History

A 16-year-old female presented with the chief complaint that, following the delayed exfoliation of the both primary teeth number 45 and 55; the permanent second premolars had not erupted. Her medical history was free. The patient was referred to an orthodontist for further evaluation and diagnosis. The orthodontist advised that a panoramic radiograph be obtained. The panoramic radiograph (Figure1) revealed the following:

Generalized Short Root Anomaly (SRA) in the permanent dentition. In the majority of cases, the root/crown ratio was approximately 1:1 and apices were slightly rounded. Despite the short roots, all teeth exhibited normal pulp chambers and root canals, and the periodontal tissues appeared normal.

A congenital absence of both upper second premolars (hypodontia).

The roots of the third molars were developed, despite the teeth's young age. The shape and root/crown ratio did not clearly indicate SRA, as the shape of the apices was normal.

The second mandibular and maxillary molars and the upper third molars exhibited taurodontism. The type of taurodontism is considered to be mesotaurodontism (Type III) (Shifman and Chanannel, 1978).

All primary teeth had been extracted in a normal manner, and there was no previous orthodontic examination or treatment. The clinical examination revealed that all teeth were of normal shape, color and size. No teeth exhibited mobility and all responded normally to vitality tests. Patient demonstrated excellent oral hygiene, with no evidence of soft tissue inflammation. Carious lesions were identified and scheduled for restoration.

The most important clinical observation was that the patient was born with incomplete formation of the left upper limb with a deficiency in 2/3 of the forearm (transverse hemimelia). A comprehensive medical history was obtained from the parents to ascertain any potential contributing factors.

Methods

For the investigation of possible causes related to vitamin and trace element deficiencies, it was proposed that blood tests be conducted to measure levels of essential vitamins and minerals, including vitamin D, calcium, and phosphate. The blood tests yielded results within the normal range. The family's radiological evaluation revealed that no other family member (father, mother, and elder brother) exhibited the same clinical condition. A systematic re-evaluation with intraoral radiographs every six months was recommended. Furthermore, a Cone Beam Computed Tomography (CBCT) was requested to determine the position of both mandibular third molars in relation to the inferior alveolar nerve. Regarding the hypodontia, it was recommended to preserve the space of the primary teeth and to consider implant placement once the patient reaches adulthood.

Conclusions

A rare distortion that involves the incomplete development of the length of all roots (SRA) and concurrently, hypodontia and taurodontism is presented in a female patient with the associated finding of transverse hemimelia. Although there is no documented evidence of a direct link between SRA and hemimelia, the shared genetic, environmental, and metabolic factors indicate the possibility of a common underlying mechanism.

Discussion

A potential biological foundation for the emergence of short roots can be elucidated through an examination of the regulatory function of the Nfic gene (Park, 2007; Yu, 2019). The aberrant expression of specific factors, including Nfic, Osx, Hh, BMP, TGF-β, Smad, Wnt, β-catenin, DKK1, and other recently identified proteins, can contribute to the development of SRA. The absence of the Nfic gene, which is critical for odontoblast differentiation, has been demonstrated to result in the development of short, abnormal roots in rats through

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immunohistochemical and in situ hybridization studies. It has also been proposed that Nfic knockout mice display abnormalities in bone formation and maintenance, which may indicate a more extensive involvement in skeletal development (Lee et al., 2013). The precise regulation of Wnt/β-catenin signaling is of great importance during root morphogenesis. Further research is required to elucidate the specific mechanisms underlying these effects (Yu, 2019). Given that hemimelia is characterized by skeletal abnormalities, it is plausible that mutations or deletions in genes such as Nfic may contribute to the development of limb deficiencies.

Bone morphogenetic proteins (BMPs) constitute a group of growth factors that are critically important in the regulation of bone and cartilage development. They are members of the transforming growth factor-beta (TGF-β) superfamily and play crucial roles in several developmental processes, including the formation of teeth and limbs. BMPs, which interact with the Nfic gene in dental development, are also essential for limb development (Chen et al., 2012). The involvement of BMPs in both SRA and hemimelia suggests the existence of a potential link through common developmental pathways. Mutations in genes that regulate BMP signaling are critical for the proper development of dental roots (Li et al., 2017).

It is well documented that prenatal exposure to specific drugs is a prevalent risk factor for the development of both SRA and hemimelia. Thalidomide, a known teratogen, has been identified as a potential cause of limb defects such as hemimelia and dental anomalies including SRA (Bermejo-Sánchez et al., 2011; Puranik et al., 2015. Metabolic disorders such as hypophosphatasia, which affects bone mineralization, can also impact dental root development, leading to SRA (Wei et al., 2010). A deficiency of vitamin D and associated conditions such as vitamin D-dependent rickets type I have been linked to SRA due to their role in bone and dental development (Zambrano et al., 2003). Pregnancy-related deficiencies in essential vitamins and minerals, particularly in the context of poor maternal nutrition, can contribute to the development of congenital limb deficiencies (Bermejo-Sánchez et al., 2011). Retinoic acid (RA), a metabolite of vitamin A, plays a critical role in embryonic development. RA levels must be meticulously regulated during limb bud development, as both elevated and diminished levels are linked to developmental abnormalities (Bermejo-Sánchez et al., 2011). Pregnant women with high or low levels of RA have been found to have an increased risk of developing developmental abnormalities. RA regulates the Hox genes involved in limb and dental development, influencing the formation and differentiation of chondroblasts and osteoblasts, as well as regulating chondrocyte maturation and bone formation (Bermejo-Sánchez et al., 2011). It can thus be surmised that irregularities in RA levels may be a contributory factor in the development of both SRA and hemimelia.

Despite the presence of SRA throughout the dentition, the third molars were fully formed, exhibiting no indications of the corresponding appearance of rounded apices, which would be expected given their young age. The formation of the roots of permanent teeth is typically completed four to five years before the formation of the third molars (Wilson et al., 2021). It is not possible to determine whether SRA is present in third molars based on the root/crown ratio alone, as these teeth vary significantly in size and shape. It is plausible that the factor influencing the root length of all other teeth did not affect the third molars.

Hypodontia and taurodontism are common accompanying clinical findings in cases of SRA (Puranik et al., 2015). Hypodontia is a dental anomaly, with a frequency of 6.9% across the general population, with the most affected teeth being the premolars, accounting for up to 2.2% of cases (Polder et al., 2004). Taurodontism is a condition where the ratio of the pulp chamber length to the total length of the pulp is reduced, resulting in teeth having a characteristic appearance. An enlarged pulp chamber, an apical position of the pulpal floor, and a lack of constriction at the level of the cementoenamel junction are the main features, affecting molar teeth and resulting in the roots being closer together or even fused (Shifman and Chanannel, 1978).

SRA primarily raises concerns pertaining to aesthetics and restoration. However, with meticulous treatment, the prognosis of the dentition is typically favorable in cases of isolated dental groups. The occurrence of generalized SRA is considered rare (Apajalahti et al., 2002). The potential for possible deterioration of the clinical condition concerning tooth support has not yet been studied longitudinally in cases of SRA. Patients with SRA are at risk of developing periodontitis or increased tooth mobility due to poor root support. Under protective measures, the clinical picture of SRA remains stable (Valdares-Neto et al., 2013; Trimeridou

et al., 2023). SRA presents a significant therapeutic challenge across all areas of dentistry, particularly in endodontic, periodontal, orthodontic, and prosthetic restoration.

Congenital limb malformations have substantial social and psychological implications for both the patient and the family environment. This was evident throughout the history-taking process, particularly with regard to the pregnancy period, during which the parents were reluctant to provide detailed information about ultrasound examinations and prenatal screening and expressed negativity towards supplementary examinations such as CBCT or intraoral radiographs. Additionally, the parents were opposed to taking any photographs, either intraoral or of the missing limb.

As this is the first documented case presenting with these concurrent conditions, further research is necessary to determine any potential direct associations or shared underlying pathophysiological mechanisms between them.

Key Clinical Message

Short root anomaly (SRA) is characterized by the root-to-crown ratio and can affect all groups of teeth in a patient., while its presence in all permanent teeth is very rare. This condition is frequently associated with various dental anomalies, including hypodontia and taurodontism. Usually, the shape and size of the crown remain within normal limits.

In the case presented, the co-occurrence of transverse hemimelia suggests a potential shared developmental etiology between the limb defect and the dental anomaly, implying that both conditions may arise from similar disruptions in embryonic tissue differentiation or genetic factors.

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Author Contributions:

Author 1 (Emmanuel Mazinis): Co-author

Author 2 (Vasilios Thomaidis): Co-author

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