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Short Root Anomaly and Transverse Hemimelia: A Rare Case Presentation

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ABSTRACT

A case of Short Root Anomaly (SRA), hypodontia and taurodontism with the associated finding of transverse hemimelia, is presented. Although there is no documented evidence of a link between SRA and hemimelia, the shared genetic, environmental and metabolic factors suggest the possibility of a common underlying mechanism.

Taxonomy Classification: Dentistry, Anatomy

1 | 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 [1]. 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 [2]. 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 [1, 3]. The detection is often incidental and the prevalence of SRA in any teeth type is estimated to be between 0.6% and 2.4% [3, 4], with a higher incidence observed in females, with a ratio of approximately 1:2.6–1:2.7 [3, 4]. Despite the short roots, the affected teeth have normal crown morphology [2, 3, 5] and exhibit normal pulp chambers, root canals, and closed apices, supported by normal periodontal tissues [1–3]. The term "hereditary idiopathic root malformation" has been proposed for cases not associated with any specific syndrome [2].

Certain syndromes and metabolic disorders also present reduced root length. These include Stevens–Johnson syndrome [6–8], Turner syndrome [9], Fraser syndrome [10], Hallermann–Streiff syndrome [11], and Schimke immuno-osseous dysplasia [12]. Furthermore, metabolic disorders such as pseudohypoparathyroidism [13, 14], hypophosphatasia [15], and vitamin D-dependent rickets Type I [16] 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 [17].

It is possible that the factors that contribute to the development of SRA may also influence the proper formation and growth of the skeletal system, given the correlation and shared genetic and metabolic background. Transverse hemimelia is a congenital limb deficiency characterized 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 categorized 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 [18]. 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) [19]. In the majority of cases, the precise etiology remains undetermined, with no discernible genetic or environmental factors [19]. Furthermore, there is no evidence for familial recurrence [20]. 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 [19].

The purpose of this study was 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.

2 | 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. At the time of presentation, there was no significant medical history. The patient was born with incomplete formation of the right upper limb with a deficiency in 2/3 of the forearm (transverse hemimelia). The patient was referred to an orthodontist for further evaluation and diagnosis. The orthodontist advised that a panoramic radiograph be obtained. The panoramic radiograph (Figure 1A) revealed the following:

- Generalized SRA in the permanent dentition. In the majority of cases, the root/crown ratio was approximately 1:1 and apices were slightly rounded. 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) [26].

Developmental disorders are illustrated based on the panoramic radiograph in Figure 1B.

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

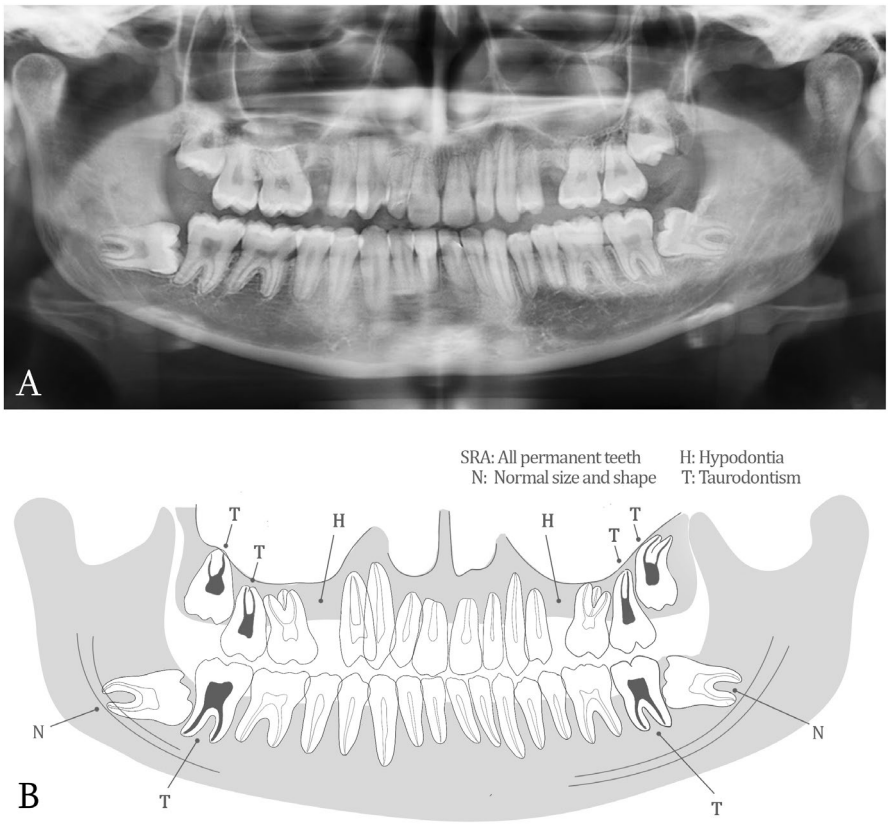

FIGURE 1 | Panoramic X-ray of the patient (A) and the illustration of the panoramic X-ray highlighting the dental anomalies (B). The SRA affects the entire dental arch, while the third molars do not appear to be affected.

for restoration. A comprehensive medical history was obtained from the parents to ascertain any potential contributing factors.

3 | Methods

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. As cases of SRA are challenging for orthodontic treatment due to incomplete root development a systematic reevaluation with intraoral radiographs every 6 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.

4 | Conclusions

A rare distortion that involves the incomplete development of the length of all roots 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.

5 | 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 [21, 22]. 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 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 [23]. 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 [22]. 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 [24]. 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 [25].

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 [2, 19]. Metabolic disorders such as hypophosphatasia, which affects bone mineralization, can also impact dental root development, leading to SRA [15]. 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 [16]. 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 [19]. 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 [19]. 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 [19]. 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. 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. The formation of the roots of permanent teeth is typically completed 4–5 years before the formation of the third molars [29]. It is plausible that the factor influencing the root length of all other teeth did not affect the third molars.

Hypodontia is a common 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 [30]. 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 [26].

SRA primarily raises concerns pertaining to aesthetics and restoration, although the prognosis of the dentition is typically favorable in cases of isolated dental groups. The occurrence of generalized SRA is considered rare [3]. 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 [27, 28]. 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. Further research is required to ascertain whether there are any direct associations between these conditions.

Author Contributions

Emmanuel Mazinis: conceptualization, data curation, formal analysis, funding acquisition, investigation, methodology, project administration, resources, software, supervision, validation, visualization, writing – original draft, writing – review and editing. **Vasilios Thomaidis:** conceptualization, data curation, formal analysis, funding acquisition, investigation, methodology, project administration, resources, software, supervision, validation, visualization, writing – original draft, writing – review and editing.

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The authors have nothing to report.

Consent

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

Conflicts of Interest

The authors declare no conflicts of interest.

Data Availability Statement

The authors have nothing to report.

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