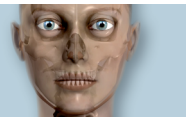
Orthodontics & Craniofacial Research



ORIGINAL ARTICLE

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Characterization of short root anomaly in a Mexican cohort – hereditary idiopathic root malformation

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Structured Abstract

Objective – The purpose of this study was to systematically characterize individuals with short root anomaly (SRA) without any history of orthodontic treatment. The long-term objective of the study was to improve diagnosis and treatment planning and determine risk factors for developing SRA.

Setting and Sample Population – Twenty-seven patients including two families and 16 unrelated individuals from (9–48 years) reported to orthodontic and/or dental practitioners within the USA.

Materials and Methods – Digital panoramic and periapical films were analyzed to document pattern and frequency of SRA-affected teeth. Crown-to-root (CR) ratios of the affected teeth were used to characterize the extent of malformation. Pedigree analysis by inspection was completed for one family to determine pattern of inheritance.

Results – Twenty-six of the twenty-seven individuals were of Latino descent, and one was of Filipino descent. Hard tissues including enamel, dentin, pulp chambers and canals, and surrounding soft tissues were normal. We found that 25 of 27 individuals had localized SRA and two Latino individuals had generalized SRA. Teeth were affected bilaterally with maxillary central incisors (~63%) and mandibular second premolars most commonly involved (~33%). Affected teeth had a distinct, similar radiographic appearance; in the generalized cases, there was a more severe affection with larger (~twice) CR ratios. Ninety-four percent of affected individuals did not show a significant difference in the CR ratios at different ages. Pedigree analysis suggests an autosomal dominant inheritance pattern in one family.

Conclusion – This is the first report to show that SRA occurs more frequently in Latino individuals and has a predilection for anterior teeth.

The occurrence of SRA in two families further confirms a hereditary component and supports a distinct nosology and nomenclature, hereditary idiopathic root malformation (HIRM) and warrants further investigation.

Key words: crown–root ratio; hereditary root malformation; idiopathic root malformation; root malformation; short root anomaly

Introduction

The short root anomaly (SRA) is not well documented likely because it is poorly understood, misdiagnosed, and the etiology remains unknown. SRA was first identified by Volmer Lind (1) in 1972 in a Swedish population. Lind defined SRA as 'abnormally short, plump roots always affecting the maxillary central incisors and rarely any other teeth'. Permanent teeth with short roots greatly impact diagnosis and treatment planning. The prevalence of misdiagnosis of SRA is a significant concern given its influence on treatment planning, especially in the prospective orthodontic patient. There is evidence that unusual root morphology before orthodontic treatment can increase the risk of root resorption (2). Patients with short or blunt roots before treatment underwent significant root shortening during orthodontic treatment (3). Periodontal disease and root resorption will compromise prognosis in an individual with the SRA phenotype. Therefore, the proper diagnosis and clinical management of this condition is critical.

Since its initial documentation, the prevalence in Caucasians has also been reported as 2.4–2.7 and 10% in Mongolian populations (4). However, no epidemiological studies have documented the presence of this anomaly among Latinos. Lack of clinical signs or symptoms of presence of the condition in an individual is the principal reason why SRA had received little attention. SRA is often misdiagnosed as root resorption due to the low frequency among Caucasians. The frequency of the dental anomaly in Caucasians may be low, but the prevalence in Latinos is likely greater. The increase in SRA reports in the USA is juxtaposed with an increase in the Latino population in the USA. The Latino population in the USA is rapidly growing and will likely continue to do so in the future. According to an update from the Population Reference Bureau's, the US population grew by only 9% between 2000 and 2009, while the Latino population alone increased by 37% over the same period of time. As the Latino population has continued to grow, so have the reports of this dental anomaly. The observation of an apparent increase in reported cases, while anecdotal, warrants a formal investigation of both the characteristics and prevalence of idiopathic root formation to provide better diagnosis and management of the condition.

An autosomal dominant inheritance pattern of SRA has also been documented in several individuals with SRA from eight families (4). And while the literature reveals evidence of familial cases of SRA, no study has previously shown an association of SRA with the Latino population. The objective of our study was therefore to characterize SRA, in terms of morphology and segregation in one family and document its occurrence in an orthodontic cohort. We also aim to document the occurrence of this anomaly in a Latino population. These studies will aid in future diagnosis, determine specific genetic etiology, and increase awareness of the anomaly in the dental community.

Materials and methods

Patient identification and characterization

Twenty-seven SRA-affected individuals (age range 9–48 years) referred from orthodontic and/or dental practitioners within the USA were selected for the study. The inclusion criteria

included normal size and shape of the tooth crown. History of trauma, root resorption, and orthodontic treatment was used to exclude any individual from the study. Twenty-six of the 27 SRA individuals were of Latino descent, and one individual was of Filipino descent. Sixteen patients were unrelated, whereas rest were from two families. Pedigree analysis by inspection was performed for two families. The study was approved by the Institutional Review Board for Biomedical Research on Human Subjects and was conducted in accordance with the ethical standards and principles for research involving human subjects at the University of North Carolina at Chapel Hill. All subjects were de-identified, except for age, prior to referral to the study coordinator.

Clinical and radiographic exam

Clinical examination was performed and at the initial visit and subsequently as required on an individual basis. Both digital panoramic and periapical films collected from patient records were analyzed to characterize any pathology including SRA. Surrounding tissue, root morphology, and crown-to-root length (CR) ratio of affected teeth were analyzed to characterize the SRA in the individuals. Radiographic density, amounts of the enamel, dentin, periodontal ligament, and lamina dura were observed. Abnormalities in the pulp and the presence of pathologies were noted in all individuals. The root morphology was then determined. Root was generally classified as having a blunt or round apex and whether or not the root appeared noticeably plump (lack of significant taper toward the apex).

CR ratio calculations

The CR ratio was determined using Lind's method (1). Lind did not use the cemento-enamel junction as the boundary between the crown and root when viewing radiographs because the line does not lie in the same plane as the apex. Therefore, two points were first drawn on the affected tooth. As shown in Fig. 1,

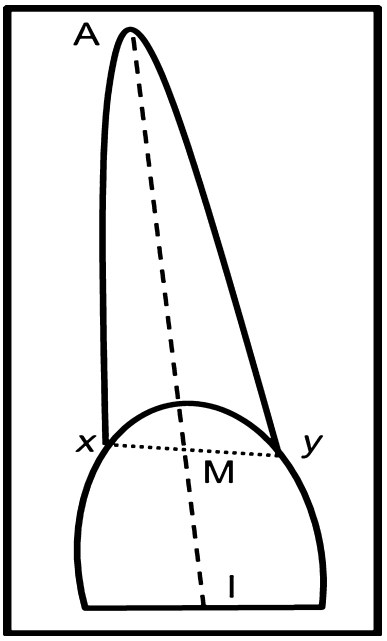

Fig. 1. Two points were placed on the mesial and distal of the tooth at the intersection of the outline of the crown and root, '*y*' and '*x*', respectively. The points were connected horizontally, and the midline of this line was marked 'M'. The root apex and midpoint of incisal edge were marked as 'A' and 'I', respectively. A line was drawn to connect points A-M and M-I. The crown CR ratio was determined using the expression CR ratio = IM length/AM length, where IM and AM both represent the crown and root length, respectively.

the first point was placed on the mesial of the tooth at the intersection of the outline of the crown and root as '*y*'. Similar point was placed on the distal outline as '*x*'. The points were connected horizontally, and the midline of this line was marked 'M'. The root apex was marked as point 'A', and the midpoint of the incisal edge or cusp tip was marked 'I'. A line was drawn to connect points A-M and M-I. The crown CR ratio was determined using the expression CR ratio = IM length/AM length, where IM and AM both represent the crown and root length, respectively. The CR ratio was calculated for each tooth in each affected individual, and the mean CR value was then calculated for each affected tooth in our population of SRA individuals.

Statistical analysis

Descriptive statistics for the population were generated with frequency of affected teeth in localized and generalized SRA-affected individu-

als for comparison with previously reported studies.

Results

Our study documents and characterizes SRA based on identification and referrals from private practitioners. Of those cases referred, 26 were Latino and only one was Filipino. In all individuals, enamel, dentin, pulp chambers and canals, and all surrounding tissues appeared normal. No other dental or craniofacial pathology was noted. Twenty-five of the 27 individuals had localized SRA; two Latino individuals had generalized SRA. In generalized cases, all teeth were affected. Teeth were always affected bilaterally with maxillary central incisors most commonly affected (~63%). Molars and mandibular canines were least commonly affected (~10%). Affected mandibular second premolars as well as maxillary central incisors had a distinct appearance. Maxillary central incisors appeared plump with rounded apices on periapical radiographs. Mandibular second premolars did not appear as plump, but had blunted apices that could easily be misdiagnosed as root resorption. Affected teeth in the generalized cases were more severely affected, having larger CR ratios (~twice as in localized cases).

In a severe manifestation of a 9-year-old female, clinical photos reveal severe malalignment and crowding (Fig. 2). The panoramic radiographs of the same female at age 9 and 12 illustrate the developing premolar roots at 9 years old and the lack of further development at 12 years old (Fig. 3). This manifestation of generalized SRA is characterized by a blunted root and a significantly reduced crown–root ratio.

CR ratios in 94% of the SRA-affected individuals were not significantly different at following visits as compared to the CR ratios at the initial visit. Table 1 summarizes the morphological features of all the affected teeth in localized SRA. Most commonly affected tooth is the maxillary central incisor followed by maxillary first and second premolar and mandibular second premolars. Tables 2 and 3 summarize the CR ratios of each tooth in maxillary and mandibular arch with number of affected individuals. The most commonly affected tooth was the maxillary central incisor. The mean CR ratio for two maxillary central incisors was 1.4 ± 0.8. Similarly, the

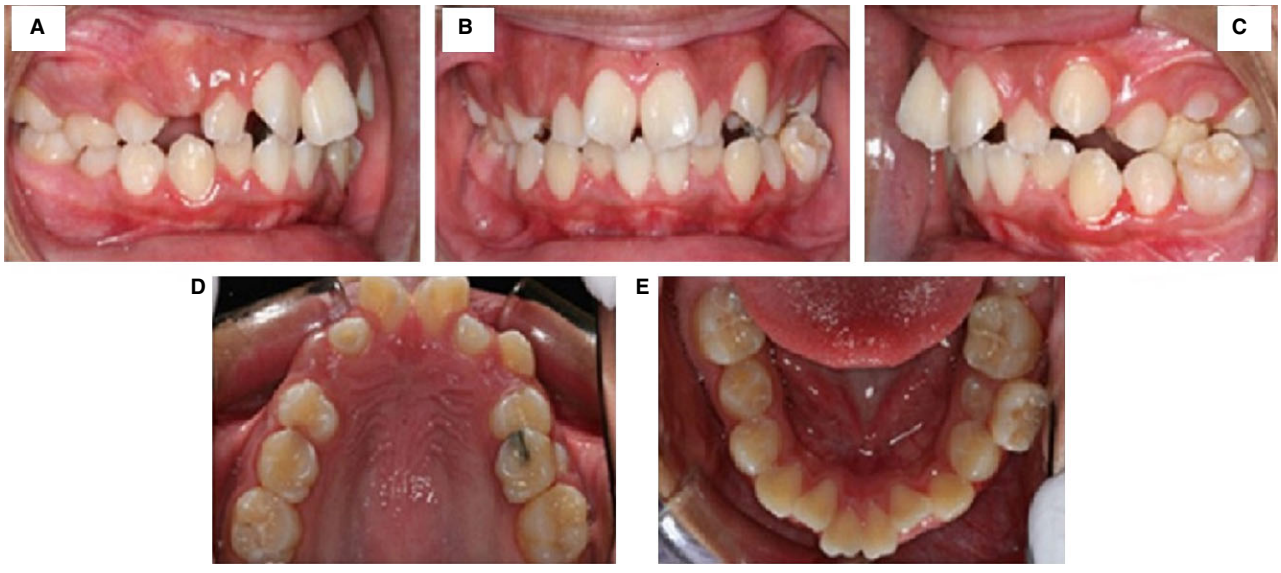

Fig. 2. Clinical photographs of a 9-year-old female with generalized SRA who also presents with a significant malocclusion due to crowding and malalignment shown in (A) right buccal view, (B) frontal view, (C) left buccal view, (D) maxillary occlusal view, and (E) mandibular occlusal view. The orthodontic force needed to correct her teeth may further jeopardize the root length and the longevity of her teeth.

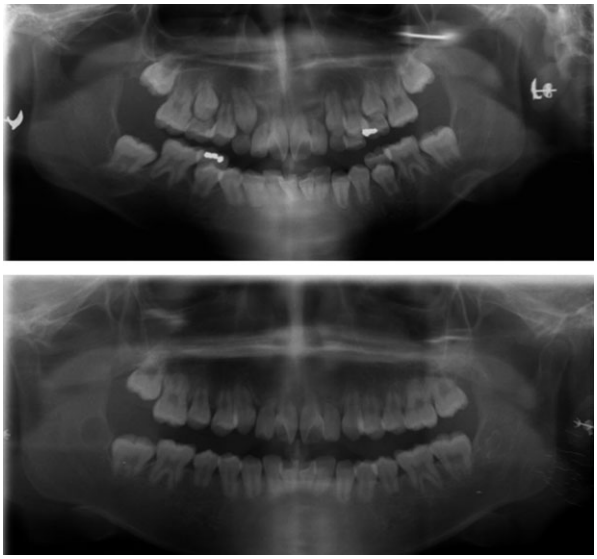

Fig. 3. The panoramic radiographs of the same female reported in Figure 2 at age 9 and 12.

Table 1. **Root morphology of affected teeth in localized cases. (Mandibular first premolars were not affected in localized cases)**

Affected tooth	Root	Apex
Maxillary central incisors	Plump	Rounded
Maxillary first premolars	Normal	Rounded
Maxillary second premolars	Normal	Rounded
Mandibular second premolars	Normal	Blunt

Table 2. **Mean CR ratio ± SD for each maxillary tooth. Maxillary central incisors are commonly affected**

Maxillary tooth, n	No. of affected individuals	Mean CR ratio ± SD
3	2	1.53 ± 0.59
4	3	1.71 ± 0.16
5	2	1.11 ± 0.10
6	2	1.37 ± 0.29
7	2	1.97 ± 1.82
8	10	1.52 ± 0.90
9	10	1.26 ± 0.89
10	2	1.99 ± 0.98
11	2	1.06 ± 0.19
12	3	1.13 ± 0.47
13	3	1.48 ± 0.38
14	2	1.11 ± 0.23

Table 3. **Mean CR ratio ± SD for each mandibular tooth. Mandibular second premolars are commonly affected**

Mandibular tooth, n	No. of affected individuals	Mean CR ratio ± SD
19	2	0.92 ± 0.04
20	7	1.14 ± 0.45
21	1	1.30 ± 0.34
22	2	1.14 ± 0.20
23	2	0.93 ± 0.19
24	2	1.52 ± 0.51
25	2	1.5 9 ± 0.67
26	2	1.25 ± 0.57
27	2	1.38 ± 0.33
28	1	1.32 ± 0.22
29	7	1.33 ± 0.53
30	2	1.01 ± 0.12

mean CR ratio for mandibular second premolars was 1.3 ± 0.4. Table 4 summarizes the comparison of the CR ratio of the SRA-affected teeth in localized and generalized cases. The CR ratio in generalized cases is approximately twice that seen in localized cases.

Finally, we found an autosomal dominant pattern of inheritance in a family of 32 with seven affected individuals (Fig. 4). The affected individuals in this family had localized SRA, and similar to most other SRA cases, the most commonly affected tooth was maxillary central incisor. Periapical radiographs of the 12-year-old daughter of the proband (III:4) and 8-year-old son of the proband (IIII:7) are shown in Fig. 5A, B, respectively.

Table 4. **Comparison of the mean CR ratio for teeth affected with localized and generalized cases. Affected teeth in the generalized cases were more severely affected, having larger crown-to-root ratios**

Affected tooth	Mean CR ratio ± SD for localized cases	Mean CR ratio ± SD for generalized cases
8	1.00 ± 0.16	2.57 ± 0.25
9	1.03 ± 0.14	1.70 ± 1.63
20	0.78 ± 0.18	1.54 ± 0.18
29	0.81 ± 0.08	1.69 ± 0.23

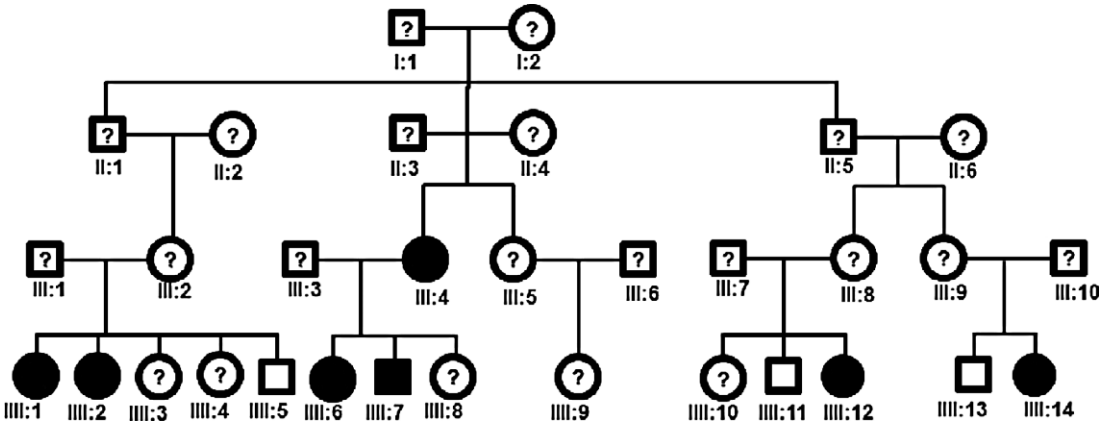

Fig. 4. Pedigree of a family of 32 individuals with seven affected family members (ranging from 8–29 years of age).

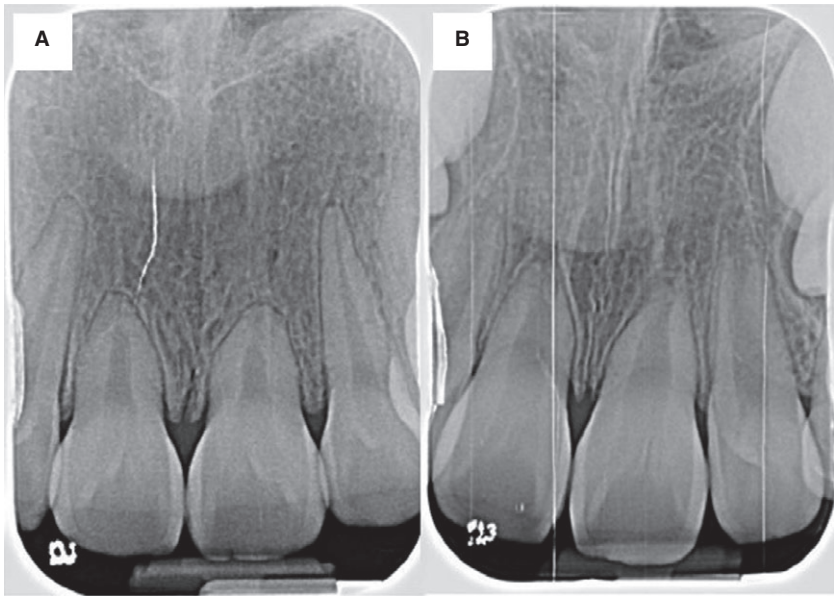

Fig. 5. (A) A periapical radiograph of the 12-year-old daughter of the proband (III:4). (B) A periapical radiograph of the 8-year-old son of the proband (IIII:7).

Discussion

In our study, we found that a previously reported condition, SRA, also has a specific predilection in the Latino population. The importance of this finding, along with its hereditary component in two families, is the contribution to the diagnostic regime for the general dentist and orthodontist. The implication of documenting and understanding this condition is to allow for refinement of clinical management downstream. For instance, the increased risk of root resorption or other adverse effects secondary to orthodontic tooth movement is quite high. Further, with an increasing presence of a Latino population in the USA, SRA is likely to increase in prevalence over time. Previous studies have asserted that the prevalence of this condition ranges from 2.4 to 2.7% in Caucasians, 0.7–1.2% in a Turkish population (5), and 0.3% of a Finnish population (6). Interestingly, those populations of Asian descent have revealed a higher prevalence – 10% in a Mongolian population (1, 7, 8) and 10% in a Japanese population (9). This may be consistent with the admixture of the Mexican native that includes an Asian component. Ongoing studies to complete whole exome sequencing for selected affected individuals will definitively determine the genetic etiology.

In most studies, SRA has been reported as non-syndromic; however, SRA has been commonly reported with hypodontia. Approximately

12% of the SRA-affected individuals in one report had hypodontia (4). Rarely, SRA is associated with syndromes; one report in two primordial dwarf siblings occurred with microdontia (10). In another case report, generalized SRA with obliterated pulp chamber was reported in short-limb dwarf (11). SRA is a reported finding in Steven–Johnson syndrome along with enamel hypoplasia as common finding (12, 13). Dentin dysplasia type I commonly affects root and often demonstrates SRA (14). One of the hallmark features of dentin dysplasia type I is the 'waterfall appearance' of the dentin which separates the dentin dysplasia type I from non-syndromic SRA (15). SRA is also seen in congenital renal disease, the condition which often demonstrates ground glass appearance of the bone (16). Two sporadic cases of SRA were also reported. In one case, SRA was seen along with microdontia, taurodontism, dens invaginatus, and obliterated pulp chamber with no known etiology (17). In second case, SRA was present along with talon cusp, dens invaginatus, microdontia, and affected mandibular incisors with no known etiology (18). One of the case report of Rothmund–Thomson syndrome (characterized by genodermatosis, poikilodermatous rash, juvenile cataracts, skeletal, and dental abnormalities) also demonstrated presence of generalized SRA (19).

In our study, SRA demonstrates autosomal dominant inheritance as previously reported (4). We did not find any microdontia, taurodontism, or hypodontia as shown in other reports (4, 6). The mean age for diagnosis of SRA has been reported to be 14 years which was comparable to the mean age of 17 years seen in our study (4, 7). Similar to the previously reported findings (1, 4, 6), we found that the maxillary central incisors were the most commonly affected tooth. Previous reports suggest that nearly 70% of the reported cases of SRA had affected maxillary central incisors. Our study also demonstrated that maxillary central incisors were affected in more than 63% of the SRA-affected individuals. Generalized and localized SRA affected teeth bilaterally; in other words, affected teeth always were present in pairs in our subjects. This bilateral affectedness of SRA has been previously reported (4, 6). The most commonly affected teeth after maxillary central incisors are maxillary premolars and mandibular premolars. Mandibular incisors, molars, and canines are least affected by SRA (4). A study documenting CR ratio in normal Finnish population reported that mean root length of maxillary central incisors was ~1.8 times that of the crown length. Similarly, mandibular second premolar root length was ~2.2 times that of crown length (20). Although comparing CR ratio between a Finnish and a Latino population would not be valid, it is interesting to note that the maxillary central and premolar root length in our study was ~0.7 and ~0.8 times that of the crowns, respectively. This gives us an approximate idea about the severity of the condition.

According to a retrospective study, nearly 40% of the 900 orthodontic patients demonstrated at least one anomaly affecting the crown or root of the teeth (5). In another study, although the kappa statistic was not reported, it demonstrated that interexaminer reliability in diagnosing SRA is not promising (21). Therefore, the chances of misdiagnosis of SRA are higher. We already know from the large number of reported cases that the resorption potential of SRA-affected teeth after orthodontic movement is very high (2–4, 6, 22, 23). Hence, it is important to detect the condition in the patient at the pre-treatment stage and carefully plan orthodontic treatment. During orthodontic tooth movement, collagenases break down collagen facilitating tooth movement of tooth. The levels of *MMP9*, a collagenase, was increased in the gingival crevicular fluid around the tooth affected with SRA as compared to normal teeth (24). Although complex interplay of cytokines exists for checking the balance of the collagen, increased *MMP9* may be one of the mechanisms involved in increased resorption potential in SRA-affected teeth.

Based on the reviewed literature and findings from our study, it is clear that SRA has an autosomal dominant pattern of inheritance. Also, the etiology of SRA is not clearly understood. It is clear from the characteristics of the SRA described in our study that SRA not only

affects the root length but also affects the overall form and shape of the tooth root. Hence, we propose a new term for non-syndromic SRA as 'hereditary idiopathic root malformation' or 'HIRM'.

Conclusion

The increasing Latino population within the USA warrants a timely documentation and characterization of this condition. We provide here numerical comparisons for calculations of CR ratios that can be applied to diagnostic regimes and therefore proper treatment planning. The apparent familial pattern supports a nomenclature that includes this distinction, hereditary idiopathic root malformation. Future genetic approaches will be focused on understanding the etiology of HIRM and the molecular mechanisms underlying the development of the condition.

Clinical relevance

Short root anomaly (SRA) is not well documented likely because it is poorly understood and the etiology remains unknown. It was first described by Lind in 1972. The anomaly may be misdiagnosed as resorption. SRA has not been reported in a Latino population in the past. To our knowledge, this is the first study characterizing SRA in a Latino population. We further confirm the previously reported autosomal dominant inheritance pattern of SRA, and therefore, we propose that a more accurate name for the anomaly would be 'hereditary idiopathic root malformation' or HIRM.

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Author contribution

AH: collected data. CPP: analyzed data and wrote manuscript. KHJ, SNH, and RWT: provided analytical insight and patient referrals. SAFB: wrote, finalized, and approved manuscript.

Conflict of interest

The authors declare no conflict of interests.

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