

## Syndromic occurrence of short root anomaly with multiple dental anomalies in siblings

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### Abstract

The case report describes two cases of siblings who presented with short root anomaly with oligodontia, dens invaginatus, taurodontism, pulp stones, multituberculism of molars and generalized microdontia. No familial occurrence was seen and the dental anomalies were not associated with systemic manifestations. The sporadic occurrence of a combination of dental anomalies in a single patient can present a diagnostic challenge due to the possibility of their occurrence with various syndromes and systemic conditions. The article also reviews previously reported cases of short root anomaly with other dental anomalies which showed diverse variations in the pattern of clinical presentation. Treatment plan in both the siblings was aimed at preserving existing teeth and early management of oligodontia.

**Key words:** Short roots, Oligodontia, Dens invaginatus

### Introduction

Short root anomaly of permanent teeth is a rare condition where the length of the root is less than or equal to that of the crown<sup>1,2</sup>. The diagnosis is based on relative root length, as defined by Lind,<sup>3</sup> where the relative length of the root is less than 1.6 times of the crown. If short roots are seen in atleast one pair of permanent teeth bilaterally, the dentition is considered to be affected. In teeth with short root anomaly, the crown and root development may proceed normally until tooth eruption and hence the anomaly is apparent to the clinician only one to two years after the emergence of the tooth<sup>1</sup>.

Short root anomaly may be associated with many systemic conditions and syndromes like idiopathic hypoparathyroidism, exposure to x-radiation during periods of odontogenesis, thalassemia, scleroderma, Stevens–Johnson syndrome, Down syndrome and Laurence–Moon–Bardet–Biedl syndrome<sup>4,5</sup>. Conical short roots on teeth with obliterated pulp chambers have been reported in Rothmund–Thompson syndrome<sup>6</sup>. Shortness of the roots of the teeth has also been observed in some short-stature syndromes such as Aarskog syndrome and dwarfism of Seckel<sup>7,8</sup>. Short root anomaly

along with multiple dental anomalies are also found to be associated with microcephalic dwarfism<sup>4,9</sup>. However few reports describe cases where the short roots are neither due to resorption nor any systemic disturbance and found to be associated with non syndromic occurrence of various other dental anomalies such as tooth agenesis, dens invaginatus, supernumerary teeth, generalized microdontia<sup>1,7</sup>. However there is a huge variation in their presentation which can be challenging to the clinician in terms of both diagnosis and management. This article describes the occurrence of short root anomaly along with other dental anomalies in apparently healthy siblings with no evidence of familial occurrence.

### Case Report

Two siblings, a 9 year old male patient and a 10 year old female patient presented with a complaint of several unerupted front teeth. The parent reported missing teeth during the primary dentition period and early exfoliation of the remaining primary teeth in both the siblings. Medical history did not reveal any systemic disease. The children were born to consanguineous parents. Their mother reported uneventful pregnancies. The patients

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had two younger siblings who did not have any missing teeth. No other member of the immediate family had missing teeth.

On examination, their height and weight were at the tenth percentile and the head circumferences of both the siblings were within normal limits. There was neither abnormality of skin, nails and hair nor history of sweating abnormality. On examination of the face, flat-bridged nose and concave profile were noted. Intra oral examination revealed following findings:

**Sibling1: (Male, aged 9 yrs)**

The teeth seen in the oral cavity were: 17, 16, 26, 36, 43, 46. (Fig 1) The permanent molars were smaller in size (microdontia) and were cup shaped with multituberculism and protosylid. The occlusal table was reduced in area due to small tooth size and was essentially flat because of the absence of normal cusp morphology. The color of the teeth was normal. Eruption bulge of erupting 33 was seen. 43 had Grade III mobility. All the other teeth showed grade I mobility. The remaining alveolar ridge was very thin. Orthopantomogram (Fig 2) revealed that all upper and lower anterior teeth were congenitally missing, except 23, 33 and 43. All the unerupted premolars present had dens invaginatus. The lower permanent molars exhibited taurodontism, short root anomaly and pulp stones were present in the pulp chamber. All the molars had single root. Tooth buds of all the unerupted second and third molars were present.

**Sibling 2: (Female, aged 10 years)**

The teeth seen in the oral cavity were: 16, 15, 14, 13, 23, 24, 25, 26, 36, 35, 43, 45 and 46. (Fig 3) The lower canine exhibited grade III mobility. The morphology of the erupted molars was similar to the first sibling. The teeth were cup shaped with multituberculism and exhibited microdontia. Orthopantomogram (Fig 4) revealed congenitally missing 12, 11, 21, 22, 33, 32, 31, 41, and 42. All the premolars seen in the radiograph had dens invaginatus. Short root anomaly was present in all the posterior teeth as well as the lower canine. In addition, the molars showed obliteration of pulp chamber with pulp chamber represented only by a crescent shaped radiolucent line. The short roots of molars did not show any bifurcation.

Patients were referred to the pediatrician to rule out any systemic disorders. Investigations revealed that blood profile, hormonal and serum levels were within normal limits. 43 was extracted in both the individuals due to grade III mobility and sent for histopathological examination. The crown root ratio of the extracted canine was approximately 1:1. Ground sections revealed the presence of normal appearing enamel and dentin with a well formed DEJ (Fig 5). Rehabilitation of both the siblings was done with removable partial dentures. Besides, preventive measures were taken to maintain the existing teeth.

**Table 1:** Clinical details of cases with multiple dental anomalies but without systemic manifestations, reported in literature.

Author/ year	Age/ Sex	Dental anomalies	Mode of inheritance
Robbins and Keene (1964) <sup>5</sup>	19yrs/ male	Microdontia, Conical canines and premolars, Short conical roots, Cup shaped molars, Multiple dens invaginatus, Multituberculism of molars	nil
Casamassimo et al (1978) <sup>14</sup>	9yrs/male	Microdontia, Taurodontism, Dens invaginatus, Conical incisors, Cup shaped molars and rosette like occlusal surface, impacted teeth, Short roots, Pulp stones	X-linked
Ireland et al (1987) <sup>7</sup>	Siblings 14yrs/male 12yrs/female	Short roots, Taurodontia, Multiple dens invaginatus	Autosomal dominant
Edwards et al (1990) <sup>2</sup>	10yrs/ female	Short roots, Microdontia, Hypodontia, Dens invaginatus	Autosomal dominant
Apajalahti et al (1999) <sup>1</sup>	Series of eight families involving 15 individuals	Short anomaly with hypodontia with one or two missing teeth in most cases. Taurodontism, Peg shaped laterals, Dens invaginatus, Supernumerary teeth seen in some affected familymembers	Autosomal dominant inheritance in three cases. Two cases were of siblings
Desai et al (2006) <sup>15</sup>	20yrs/ male	Short roots, Microdontia, Taurodontia, Dens invaginatus, hypodontia	nil



Fig 1: Intraoral view of sibling 1

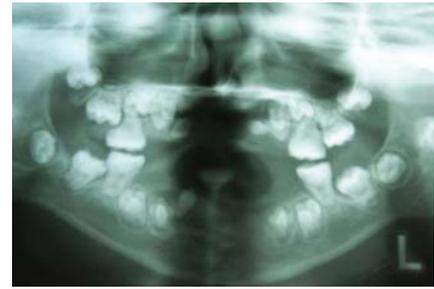


Fig 2: Orthopantomogram of sibling 1



Fig3: Intraoral view of sibling 2



Fig 4: Orthopantomogram of sibling 2



Fig 5: Ground sections taken from extracted canines of sibling 1&2 respectively at 10X magnification showing normal enamel and dentin (extracted tooth in the inset)

## Discussion

The two cases presented above were of siblings who presented with multiple dental anomalies such as oligodontia, short root anomaly, dens invaginatus, multituberculism of molars and generalized microdontia. According to our knowledge, no scientific literature is available on similar pattern of dental anomalies in a single individual. Variations were noted in the intraoral findings even between the siblings. While the older sibling had obliterated pulp chambers in the molars, the younger sibling had pulp stones and taurodontism of lower permanent molars.

The differential diagnosis included syndromes with manifestations of one or more dental anomalies seen in the above cases such as Russell silver syndrome, Williams' syndrome, psuedohypoparathyroidism which present with generalized microdontia<sup>10</sup> and those associated with oligodontia such as ectodermal dysplasia, Downs' syndrome, Ellis-van Creveld and incontinentia pigmenti<sup>11</sup>. In the reported cases, no

systemic abnormalities were detected which ruled out association of dental anomalies with any syndrome or systemic conditions associated with short root anomaly. The presence of short roots and pulp calcifications were suggestive of dentin dysplasia, however histological evidence ruled out dentinal defect<sup>12, 13</sup>.

Occurrence of multiple dental anomalies with short root anomaly has been reported previously in isolated cases or within families without evidence of systemic manifestations. From Table1, it is evident that short root anomaly can occur with multiple varied combinations of dental anomalies. A positive family history was seen in some of the previously reported cases, however, in the present case, the family history was negative. The bizarre combination of dental anomalies which occurred in these cases as well as our case demonstrates the profound morphogenetic influence that disturbances in tooth development may have on dental morphology and the protean manifestations those disturbances may

elicit<sup>4</sup>. The genetic origin of these constellation of dental anomalies is yet to be determined and is an area for further research.

The regulation of tooth development is by inductive interactions between epithelial and mesenchymal tissues which are mediated by signaling pathways. It is these signaling interactions which determine the location, identity, size and shape of teeth during early stages of tooth development. Mutations in developmental regulatory genes associated with tooth development are known to cause a variety of dental defects. For example, mutations in *Msx1*, *Pax9* and *Axin2* are known to cause non syndromic tooth agenesis<sup>16,17</sup>. The combination of dental anomalies as seen in this case probably indicates a mutation in developmental regulatory genes giving rise to different phenotypic manifestations.

The treatment plan in this case aimed at preserving existing teeth, and enhancing occlusion, mastication, and esthetics as both the siblings had severe hypodontia. It is important to preserve the existing teeth in such cases though their prognosis is poor due to short roots and grade I mobility so as to prevent resorption of alveolar processes, maintain the vertical dimension and proprioception mechanism of bone<sup>13</sup>. A recall protocol was set for these patients as every three months to make way for tooth eruptions. Dentures are to be replaced once a year to accommodate for changes in the alveolar ridge dimensions due to skeletal growth<sup>18</sup>.

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### Conclusion

The sporadic occurrence of a combination of dental anomalies in a single patient can present a diagnostic challenge due to large variation reported in literature. The clinician can arrive at a diagnosis by ruling out defects in dental hard tissues by histological methods. It is also important to rule out systemic conditions which are known to present with any one of these dental anomalies.

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