DOI: 10.1111/j.1365-263X.2007.00876.x

Assessing the proposed association between tooth agenesis and taurodontism in 975 paediatric subjects

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International Journal of Paediatric Dentistry 2008; 18: 231–234

Background. An association between tooth agenesis and taurodontism has been suggested. The identification of subpopulations with specific associated dental anomalies (subphenotype) would allow testing of the specific hypothesis that certain genetic factors contribute to the specific subphenotype.

Aim. This work aims to assess a large cohort to verify if the association between tooth agenesis and taurodontism is present.

Design. Panoramic radiographs of 1002 patients were examined and 975 were used in this study. The presence of tooth agenesis and taurodontism was assessed in the study population.

Results. The frequency of tooth agenesis was 4.6% and the frequency of taurodontism was 1.6%. There were, however, no observations of concomitant tooth agenesis and taurodontism.

Conclusions. Our data do not support the hypothesis that isolated tooth agenesis is associated with isolated taurodontism.

Introduction

Tooth agenesis is the most common congenital anomaly in humans, and most cases are likely to be the result of many genes acting alone or in combination. Tooth agenesis has been associated with 49 syndromes. Of particular interest is the association of tooth agenesis with clefts of the lip and palate, which suggests that, in some instances, tooth agenesis and clefts share the same genetic factors[1]. Tooth agenesis has also been reported in association with other dental anomalies, including taurodontism[2–6]. Taurodontism is a condition where in the tooth shape varies, characterized by an apical dislocation of the pulpal chamber of a molar, also commonly showing bifurcated individual roots[7–9].

Both tooth agenesis and taurodontism are reported to be common defects. Tooth agenesis has a frequency of between 1.6% and 10%[10,11], excluding third molars that are absent in 20% of the general population[11]. The frequency of taurodontism varies from 0.3% to 11.3%, depending on the definition of taurodontism[2,3,9,12–14].

The identification of subpopulations with specific associated dental anomalies (subphenotype) would allow testing of the specific hypothesis that certain genetic factors contribute to the specific subphenotype. Therefore, the aim of this work was to verify if tooth agenesis and taurodontism were associated and could define a subphenotype for future genetic studies of dental development.

Methods

Panoramic radiographs from 1002 patients treated at the Federal University of Rio de Janeiro's Continuing Education Clinical Program in Pediatric Dentistry were examined. All radiographs were taken in the same clinic, by the same radiology technician, and developed using the same protocol. All patients, whose ages ranged from 6 to 12 years and were seen between January 1999 and July 2006, were included in this study. Table 1 describes the distribution of the subjects by age and gender. All radiographs were examined by the same professional (E.C.K.) using the same

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protocol. In brief, radiographs were examined in a dark room over a light box that had a frame to avoid light passing on the sides of the radiograph. The protocol of this study was reviewed by the Institutional Review Board of the Federal University of Rio de Janeiro, and each subject in the project signed an age-appropriate informed consent form along with his or her parents who signed a detailed informed consent form.

Tooth agenesis was defined based on the age of the subjects and when initial tooth formation would be visible radiographs[8]. For example, second premolar agenesis was only defined in children older than 8 years. Taurodontism was assessed in the first and second lower permanent molars (multiradicular teeth) and was defined by the apical enlargement of the pulpar chamber associated with shorter roots[15]. The presence of taurodontism was determined when pulpar chamber enlargement is visible in the radiographs. Patients whose with radiographs are of poor quality were excluded. No patient was found to have an underlying syndrome.

Results

From the radiographs from 1002 patients, 27 were excluded due to poor quality; therefore, 975 radiographs were available for the study. The average age of the population studied was 8.0 years (standard deviation = 1.7).

Forty-five patients (19 boys and 26 girls) presented with tooth agenesis (4.6%). Sixteen patients (7 boys and 9 girls) presented with taurodontism (1.6%) (Table 2).

In the 45 tooth agenesis patients, 3 had four congenitally missing teeth, 4 had three missing teeth, 11 had two missing teeth, and the remaining 27 patients had just one missing tooth. Sixteen patients had maxillary missing teeth, 23 patients had mandibular missing teeth, and 6 patients had tooth agenesis in both arches. The most commonly absent tooth was the second mandibular premolar (Table 3).

Taurodontism affected 40 mandibular molars, 28 first molars, and 12 second molars. No tooth agenesis patient presented with taurodontism. The average age of children with taurodontism was 7.93 years (standard deviation = 1.72).

Table 1. Distribution of the population studied according to age and gender.

	Gender					
	Male		Female		Total	
Age (years)	*n*	%	*n*	%	*n*	%
6	61	13.1	102	19.9	163	16.7
7	109	23.4	123	24.0	232	23.8
8	131	28.2	134	26.2	265	27.2
9	71	15.3	71	13.9	142	14.5
10	61	13.1	51	10.0	112	11.5
11	13	2.8	20	3.9	33	3.4
12	18	3.9	10	2.0	28	2.9
Total	464	100	511	100	975	100

Table 2. Frequency of tooth agenesis and taurodontism.

	Frequency					
	Males		Females		Total	
Dental anomaly	*n*	%	*n*	%	*n*	%
Not present	438	94.4	476	93.1	914	93.8
Tooth agenesis	19	4.1	26	5.1	45	4.6
Taurodontism	7	1.5	9	1.8	16	1.6
Total	464	100	511	100	975	100

Note: The frequency of tooth agenesis and taurodontism is not statistically different between males and females [χ^2-test; $P = 0.95$ (taurodontism) and $P = 0.46$ (tooth agenesis)].

Table 3. Teeth affected by agenesis.

Teeth	Frequency *n*	%
Second mandibular premolar	27	34.6
Second maxillary premolar	18	23
Upper lateral incisor	14	17.9
Lower lateral incisor	10	12.8
Lower central incisor	4	5.2
First mandibular premolar	2	2.6
Lower canine	2	2.6
Upper canine	1	1.3
Total	78	100

Discussion

Our results do not support the hypothesis that tooth agenesis is associated with taurodontism. We found that the frequency of taurodontism (1.6%) is within the range reported in the literature (0.3–11.3%)[2,3,9,12–14]. We also found that the frequency of tooth agenesis (4.6%) is

within the range described in the literature[10,11]. Previous studies have reported that second and third molars are more commonly affected by taurodontism[9,12]. We found more first-molar cases of taurodontism, and this difference may be due to the younger population in this study and the impossibility of examining third molars.

The studies that suggested association between tooth agenesis and taurodontism imply that the association is more likely in cases of severe tooth agenesis (oligodontia or six or more absent teeth)[2–4]. The most severe cases we found had four missing teeth only and this may explain our findings. Severe tooth agenesis cases, however, are much less common in the population (frequency of 0.25%)[16], and the reports of families with known gene mutations segregating with tooth agenesis did not describe associated taurodontism[1].

A *DLX3* frameshift mutation was identified in the amelogenesis imperfecta hypoplastic–hypomaturation with taurodontism syndrome[17]. One can argue that variation in *DLX3* or other *DLX* family gene members could contribute to isolated forms of taurodontism. This model of using syndromes as models for isolated traits that are part of the syndromic spectrum have being successfully used in the case of isolated cleft lip and palate, isolated tooth agenesis and *IRF6* (which mutations in *IRF6* cause Van der Woude syndrome, which presents with oral clefts, lower lip pits, and in 40% of the cases tooth agenesis)[18,19].

There is evidence that taurodontism could be linked to oral clefts. A Finish study that investigated 39 twins with cleft lip (13 monozygotic and 26 dizygotic) reported that 4 of 13 monozygotic pairs with taurodontism were concordant for the trait, as were 9 of 12 dizygotic pairs. Taurodontism was symmetric in 91% of the affected molar pairs[20]. Concomitant taurodontism and tooth agenesis was observed in 8 of 16 taurodontic twin pairs, which leads us to believe that tooth agenesis and taurodontism associated with clefts may have different genetic contributing factors than the isolated forms.

In summary, our data do not support the hypothesis that tooth agenesis is associated with taurodontism and that this association could constitute a subphenotypic craniofacial trait.

What this paper adds

- It appears that the suggested association between tooth agenesis and taurodontism may not be real.

Why this paper is important to paediatric dentists

- Tooth agenesis is the most common congenital anomaly in humans and is associated with more severe conditions such as cleft lip and palate.
- Clinical markers such as preferential associations of dental abnormalities can help the understanding of the underlying cause leading to craniofacial defects.

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