Original Research Article

Oral bony outgrowths: Prevalence and genetic factor influence. Study of twins

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ARTICLE INFO

Article history:
Received 24 March 2015
Received in revised form
22 May 2015
Accepted 14 July 2015
Available online 29 July 2015

Keywords:
Genetic factor
Oral bony outgrowths
Torus mandibularis
Torus palatinus
Twins

ABSTRACT

Objective: The aim of the study was to verify the influence of a genetic factor on the etiology of oral bony outgrowths and to determine the prevalence and type of oral bony outgrowths (tori and exostoses) among a group of Lithuanian twins.

Materials and methods: In total, 162 twins (81 twin pairs) were analyzed for the presence or absence, type, and size of oral bony outgrowths. Statistical analysis was carried out to find the prevalence of bony protuberances and the relationship between zygosity and occurrence of oral bony enlargements. Zygosity of twins was confirmed by DNA analysis.

Results: 59.9% of the subjects had oral bony outgrowths. Mandibular tori were found in 56.8% and palatal tori in 1.8% of the sample. Palatal exostoses and mandibular exostoses were present in 1.8% and 3.1% of the sample, respectively, whereas maxillary exostoses were not found. A higher percentage of tori and exostoses were found in the group of older subjects (≥18 years old, p = 0.025). No significant difference was found between men and women in the prevalence of bony outgrowths. High \( \phi \) and \( r \) values (0.91 ± 0.062) showed very good concordance of oral bony outgrowths between monozygotic and moderate concordance (0.58 ± 0.141) between dizygotic co-twins (p < 0.001). The calculation of heritability estimate verifies dominant influence of genetic factor on the etiology of oral bony outgrowths (\( h^2 = 0.658 \)).
1. Introduction

Tori and exostoses are non-pathological and asymptomatic bony enlargements in the jaw [1]. Depending on their anatomic localization, 5 forms of oral bony outgrowths (OBOs) are defined: torus mandibularis (TM), torus palatinus (TP), palatal exostoses (PE), maxillary exostoses (MxE), and mandibular exostoses (ME) [2]. TM tends to occur bilaterally in the lingual surface of the mandible, above the mylohyoid line in the region of canines or premolars. TP are located along the midline of the hard palate. Exostoses appear on the vestibular surfaces of the jaws (maxillary and mandibular exostoses) or on the palatal face of the maxilla in the molar region (palatal exostoses) and consist of multiple bony tissue nodules [1,2].

The prevalence is quite diverse, the frequency depends on the ethnic group and race and varies according to population and sample: from 0.9% to 61.7% for TP and from 0.54% to 64.4% for TM [1].

The diagnosis of oral bony outgrowths is based on clinical and radiographic findings. Histologically, tori and exostoses consist of compact bone. Trabecular bone with a small amount of fibro-fatty marrow is sometimes visible [3]. The gingiva overlying bony outgrowths is thinner than normal and may ulcerate during mastication. It remains important to distinguish between benign bony protuberances, malignant lesions (osteosarcomas or chondrosarcomas), and systemic diseases like Gardner’s syndrome [4].

OBOs usually do not produce any symptoms, are painless, and do not require any treatment. Occasionally, problems due to OBOs are described, but the cases of removal are very rare. Some cases report the removal due to prosthesis reasons [5,6], intubation difficulties [7], limited tongue movement [8], or obstructive sleep apnea [9]. Bony protuberances may be used as autogenous bone in implantology or periodontology [10]. Some research shows that occlusal force becomes stronger in patients with the presence of tori (specifically TM) [11].

The exact cause of OBOs presence is unclear and remains highly controversial. In previous studies, genetic [11–13], functional (e.g. occlusion hyperstress) [11,14,15], and environmental (such as diet and use of drugs) [16–18] factors have been discussed as possible reasons for origin and development, but none have been defined as dominant. Scholars agree on the multifactorial nature of the causes [19,20], but few have addressed such analysis in their studies [21]. However, multifactorial needs to be proven by a series of investigations.

Previous studies have analyzed the influence of genetics in OBOs etiology mostly focusing on familial studies [12,22] and regional studies [18], or were conducted comparing ethnic groups [10,14,17]. The aim of our study was to confirm or deny the importance of genetic factor scoring concordance values of OBOs presence or absence in monozygotic (MZ) and dizygotic (DZ) twin pairs.

2. Materials and methods

In total, 162 plaster casts of individuals (or 81 pairs of twins) were surveyed, using the database of Orthodontic Department and Scientific Twin Center at the Lithuanian University of Health Sciences. Plaster casts were randomly selected from the nationwide population-based database. The criteria for inclusion were as follows: (a) subjects with permanent dentition; (b) no orthodontic treatment performed; and (c) DNA-confirmed zygosity. The sample consisted of 47 monozygotic and 34 dizygotic twin pairs. The age of the subjects ranged from 12 to 51 years (mean age, 20.3 ± 0.9 years); and the subjects were divided into 2 age groups: <18 and >18. There were 100 women and 62 men in the sample.

Two external examiners (periodontologist and dentist) were calibrated. To calibrate the examiners, 10 plaster casts which did not belong to the study sample were evaluated. The kappa (κ) index for inter-rater agreement and intraclass correlation coefficient were calculated (κ = 0.82; ICC = 0.87).

The examiners evaluated the data manually analyzing the plaster casts and looking for any type of bony protuberance (TM, TP, PE, MxE, ME) (Fig. 1). Questionable tori or exostoses were recorded as not present. In case of disagreement, the case was discussed and the consensus was reached.

We classified TM size according to the classification used by Haugen [23] and Eggen et al. [13,18] as follows: an outgrowth of less than 2 mm was considered small, 2 to 4 mm – medium, more than 4 mm – large. The distance between the alveolar process and the highest elevation of tori was measured with a digital caliper. Each protuberance was measured 3 times and the average was recorded.

To achieve the accuracy in determination of twin zygosity, DNA analysis was employed. Zygosity determination was carried out using a DNA test. The polymerase chain reaction set AmpF/STR® Identifier® (Applied Biosystems, USA) was used to amplify short tandem repeats and 15 specific DNA markers (D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D22S138, D19S433, vWA, TROX, D18S51, D5S818, FGA) and the Amel fragment of the amelogenin gene were used for comparison of genetic profiles. The zygosity determination using this molecular genetic technique reaches 99.9% accuracy.

The obtained data were analyzed using the IBM SPSS Statistics 22.0 package. The significance level was set at p < 0.05. The interdependence of qualitative evidence was evaluated by chi-square (χ²) criteria. The strength of agreement between the measurements was evaluated using the kappa (κ) coefficient. K values ≤0.2 were considered as poor agreement, 0.21–0.40 as fair, 0.41–0.60 as moderate, 0.61–0.80 as good, and 0.81–1.00 as very good [24].

The Spearman’s intra-pair correlation coefficient was calculated for MZ (tMZ) and DZ (tDZ) twins. The genetic determination was assessed using Lundstrom’s approach and
3. Results

In 162 subjects studied, 59.9% had OBOs. TP were found in 1.8% of the subjects, PE and ME were present in 1.8% and 3.1%, respectively, whereas MxE were not found. The most frequent type of bone protuberance was TM (56.8%). The results demonstrated that 51.6% of all the TM were small in size, 42.3% were medium, and 6.1% were measured as large. In the majority of the cases, the subjects had more than 1 bony outgrowth (Fig. 2).

Tori and exostoses were more often diagnosed in women, but gender difference showed no statistical significance. In addition, a lower incidence of OBOs was found in the group of younger subjects (≤18), whereas the group of the subjects over 18 displayed a higher frequency of osseous outgrowth occurrence (p = 0.025) (Table 1).

To evaluate the influence of genetics in the etiology of OBO, we calculated the concordance values for the presence or absence of OBO between the first and the second twin (co-twin) in MZ and DZ pairs. The analysis demonstrated that 95.7% of all the MZ and 79.4% of the DZ co-twins exposed concordance in the occurrence of bony outgrowths, i.e. both individuals in the pair had or did not have OBO (p = 0.021). High χ and r values (0.91 ± 0.062) showed very good concordance of OBO between co-twins in the MZ pair and moderate concordance of OBO (0.58 ± 0.141) between DZ co-twins (p < 0.01). The calculation of heritability estimate verifies dominant influence of genetic factor on OBO etiology (h² = 0.658) (Table 2).

4. Discussion

According to our data, the prevalence of OBOs is 59.9%. As found in the studies worldwide, the occurrence of OBOs has a wide range: between 0.54% and 64.4% [1]. These differences

![Figure 1](image1.png)

**Fig. 1 – Diagnosis of oral bony outgrowths using plaster casts.**

![Figure 2](image2.png)

**Fig. 2 – Distribution of oral bony outgrowths. Abbreviations:** TM, torus mandibularis; TP, torus palatinus; PE, palatal exostoses; MxE, maxillary exostoses; ME, mandibular exostoses; dex., right side; sin., left side.

| Table 1 – Prevalence of oral bony outgrowths according to gender and age. |
|-----------------------------|--------------------------|--------------------------|
|                            | Oral bony outgrowths     | Difference p-value       |
|                            | n    | %    | (chi-square test) |
| Gender                     |      |      |                |
| Females                    | 63   | 63   | p = 0.221      |
| Males                      | 34   | 54.8 |                |
| Age                        |      |      |                |
| ≤18                        | 43   | 52.4 | p = 0.025      |
| >18                        | 54   | 67.5 |                |
depend on the population studied, methods of data collection, and examination. Previously, researchers found a 0.73% occurrence of oral bony outgrowths in Lithuania [27], but the results reported diagnoses from medical records, where osseous protuberances that needed removal were counted.

Based on our research, TM dominates in the investigated sample of the Lithuanian twins. Many studies indicate ethnicity as an important factor in the etiology and the prevalence of the dominant type of OBOs [10,17,18,28]. According to the results of the conducted surveys, TP was commonly found in German, Norwegian, Croatian, Thai, and Malaysian populations [14,15,23,29]; meanwhile TM dominated in Japanese, Spanish, and Ghanian populations [11,30,31].

In our study, 51.6% of TM were smaller than 2 mm, 42.3% were between 2 and 4 mm, and only 6.1% were larger than 4 mm. This finding is in agreement with previous studies, which report small OBOs dominance [29,32]. However, this leads to the question of the precision in data collection, as it is very difficult to detect small OBOs covered with fibromucosa during intraoral examination. The strength of our study is the analysis of plaster casts, since we had the possibility to calibrate the examiners, recalculate and compare the cases in order to achieve maximum accuracy. The inter-rater agreement (weighted kappa) was 0.82 and intraclass correlation coefficient (ICC) was 0.97.

Our data agree with prior studies analyzing the correlation between OBOs and gender, saying that women tend to have any kind of OBO more often than men [18,29]. However, no significant difference between men and women was found, and this finding agrees with previous studies [11,23,33]. The findings do not confirm the X chromosome-linked heritability of OBOs.

In our study, OBOs were also diagnosed in 12- to 13-year-old individuals (6 cases). Consequently, our findings show that OBO is not an age-dependent characteristic common only in older subjects. With this data, we support previous research, which shows presence of OBOs in childhood and early adolescence [34]. Nevertheless, general analysis of the data demonstrated that OBOs were less frequently found in the younger subjects (≤18). These findings, together with other studies, indicate that OBOs grow continuously [17,30] and commonly are found later in life [11,14,18,23].

Until now, the importance of genetics in OBOs etiology has been analyzed using familial studies, regional studies, or ethnicity research [10,12,14,17,22,28]. This research gives a new perspective since it takes the sample of twins as an object to verify the influence of genetic factors on the occurrence of OBOs.

MZ twins are of the same gender and share a single genotype. Such twins are genetically identical and are more likely to share common environment; therefore, they express more similar traits than DZ twins [35]. The calculation of heritability estimate provides a means of quantifying the extent of the genetic contribution to phenotypic variation, with proportion ranging theoretically from 0 (no genetic contribution) to 1 (variation entirely attributed to genetic influence) [25]. High rate of heritability estimate \( h^2 = 0.658 \) proves the dominant influence of genetic factor in OBO etiology.

The results of our study prove the heritability of OBOs. Heritability, however, does not explain the occurrence of OBOs in all the cases or their continuing lifetime growth. A multifactorial etiology of OBOs was hypothesized before, but no consensus was found [11,19,21].

### 5. Conclusion

Our results verify dominant influence of genetic factor in OBOs etiology. The most common OBO in the studied sample of Lithuanian twins was TM. Overall, future studies in this field may give valuable knowledge about clinical variables, which stimulate bone remodeling and strengthening over time.

### Conflict of interest

The authors declare no conflict of interest.

### Acknowledgments

We are grateful to Prof. Dr. Georg Meyer for scientific consultation and to Drs. Vilma Švalkauskiénė, Mantas Šidlauskas, Eglė Rupšytė, Saulėnė Guobytė for the help in data collection.

This research was partly funded by the Research Foundation of the Lithuanian University of Health Sciences.

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