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Microdontia and hypodontia in two female skeletons from the Rovinj – St. Euphemia site*

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Abstract

This case study focuses on two rare dental anomalies – unilateral microdontia and bilateral hypodontia – identified in two female skeletons excavated in a shared burial from the medieval and early modern cemetery of St. Euphemia in Rovinj, Croatia. Detailed anthropological and radiological analyses revealed the presence of microdontia in the right maxillary third molar of individual SU 298 and hypodontia affecting the maxillary lateral incisors of individual SU 299. These findings contribute to the understanding of dental variation in archaeological contexts and highlight the influence of genetic and environmental factors influencing these anomalies. The co-occurrence of anomalies and the shared burial context suggest a potential familial link, underlining the importance of integrating bioarcheological with molecular studies. This article provides deeper insight into the health of individuals and the wider population in Rovinj between the 15th and 17th centuries.

Keywords: hypodontia; microdontia; dental anomalies; Rovinj – St Euphemia; Croatia

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Introduction

Hypodontia, also referred to as a congenital absence of teeth, predominantly affects the third molars, second premolars, maxillary lateral incisors, and mandibular mesial incisors (1). However, the term is also applied to describe the absence of other teeth, although such cases are encountered less frequently. This condition is alternatively referred to as tooth agenesis, a term that emphasizes the developmental failure of tooth formation (2). Microdontia describes a dental anomaly characterized by abnormally small tooth size. This condition can appear in an isolated form, most often affecting the lateral incisors or third molars, or in a generalized form, which is considerably rarer. Notably, microdontia exclusively affects permanent dentition, and it is widely believed to be genetically predetermined (1).

The genetic etiology of both hypodontia and microdontia has been suggested by many familial case reports (3) and corroborated through twin studies (4). Additionally, chromosomal anomalies, such as trisomy 21, have been implicated in the occurrence of these dental conditions (5). Hypodontia is rarely an isolated phenomenon as it is frequently reported with other dental anomalies, including aberrations in tooth size, further highlighting the interconnected nature of these conditions (6).

This study focuses on closely related dental anomalies identified in two female individuals excavated from a graveyard carbon-dated to 15th - 17th centuries (1480-1640 cal AD), calibrated through radiocarbon analysis of grave SU 247. It is one of the few studies in Croatia that aims to provide detailed analysis of these anomalies in an archaeological context.

Materials and methods

Protective archaeological excavations were conducted by the Department for Archaeology of the Croatian Conservation Institute between 2018 and 2023. The primary aim of the research was to gain a deeper understanding of burial practices and the daily lives of the inhabitants of Rovinj during the medieval and modern periods. The excavation campaigns uncovered various types of burial structures, including interconnected carved "chambers" with partially preserved side walls, tombs defined by burials in the parent rock with walls formed using stone, brick and lime binder, as well as burial "chambers" carved directly into the parent rock. Most burials exhibited a supine east-west orientation, with the head at the western end of the grave. While individual burials predominated, some instances of adults buried with children were observed. A small number of ceramic items were discovered before the cemetery's formation, i.e., from the prehistoric, late antique, and early medieval periods, as well as from the period of the high and late Middle Ages. Few grave goods were found in older graves, whereas younger graves contained various religious items such as rosary beads, medals, crosses, and pendant-reliquaries. Recovered numismatic finds date from the 12th to the 17th centuries (7). Some burials displayed characteristics of deviant burial practices. In the late medieval period, deviant burials were defined as those deviating from burial rites normalized by the Catholic Church (8). In this context, SU 298 burial was identified as deviant, as the individual was found with a stone block over the skull and between her legs (Figure 1).

Anthropological analysis of individuals recovered during the 2022/2023 excavation campaign was performed at the Anthropological Center in Zagreb, a unit of the Croatian Academy of Sciences and Arts. A total of 142 individuals were analyzed, including 110 adults and 32 subadults. It was possible to analyze the remains of 58 males, 46 females and all subadults, while due to the poor preservation of bones, determining the sex and age-at-death of other individuals was not possible. Biological sex was determined using morphological traits of the skull (9) and pelvis (10). Age-at-death was estimated based on examination of the pubic symphysis (11, 12), the auricular surface of the ilium (13), and dental attrition (14).

Radiographic imaging was employed for detailed analysis of dental anomalies. Digital X-rays were captured at the Zadar Veterinary Center using the Mindray VetiX P8 device, optimized for imaging bone structures. The goal of imaging was to visualize fine bone details, with device settings optimized for the absence of soft tissues. The kilovolt potential (kVp) was set to 50–60, ensuring good contrast between structures, while a current of 5–10 mAs provided adequate exposure quality. The exposure time was adjusted to 0.1–0.2 seconds, and the focal distance was maintained at 30–40 cm to achieve sharpness and even distribution of X-rays. Precise collimation was directed at the maxillae, reducing radiation scatter, while standard filtration further enhanced image clarity (information obtained at the Zadar Veterinary Center). Analysis of the images confirmed the presence of hypodontia of the lateral incisor and microdontia. These

findings were crucial for the accurate interpretation of dental anomalies in the analyzed individuals.

Results

SU 298

Individual SU 298 was identified as a female aged between 25 and 30 years, with an estimated preservation state of approximately 50%. Macroscopic examination revealed several notable findings, including a bilateral septal aperture, six lumbar vertebrae (representing a benign numerical variation without signs of transitional morphology), porosity around the external auditory meatus and on the promontory (suggestive of otitis media), and evidence of a healed fracture on the distal right fibula, indicative of an antemortem ankle sprain. Notably, microdontia was observed in the right maxillary third molar (Figure 2a).

Figure 1. Deviant burial of a female individual in SU 298. The rock was placed on her skull and between her legs (photograph: Josip Višnjić).

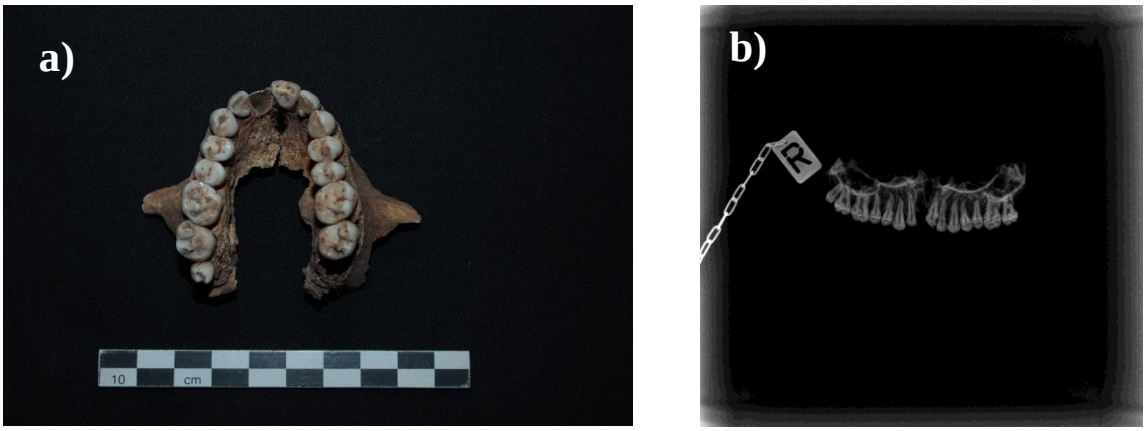

Figure 2. SU 298 (Female, 25-30) a) Inferior view of the maxilla exhibiting unilateral microdontia of the third molar (photograph: Laura Vranješ); b) Radiographic image of a maxilla showing unilateral microdontia of the third molar.

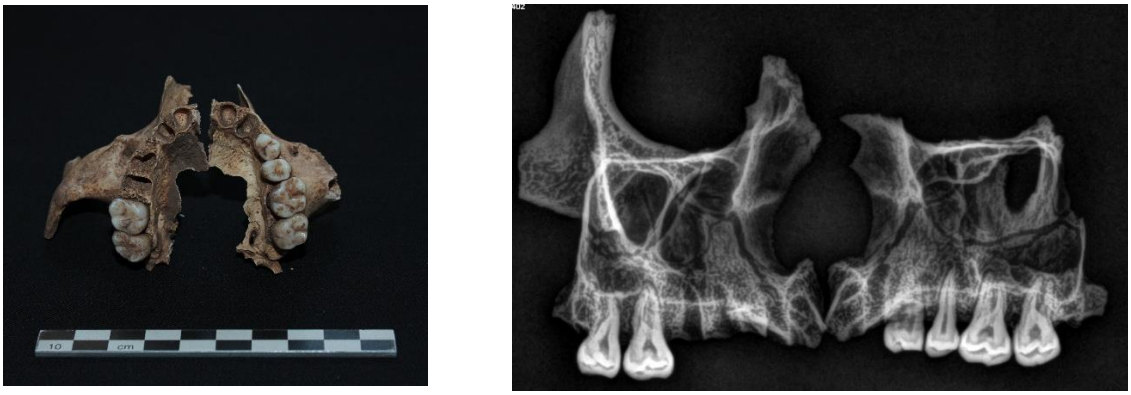

Figure 3. SU 299 (Female, 40-45) a) Inferior view of the maxilla exhibiting bilateral hypodontia of lateral maxillary incisor hypodontia; (photograph: Laura Vranješ); b) Radiographic image of a maxilla showing bilateral hypodontia of lateral maxillary incisors.

All maxillary teeth were found in situ, except for the right central incisor and left third molar, which were lost postmortem. Similarly, all mandibular teeth were in situ, with the exception of the third molars, whose status could not be determined. Slight calculus was present on the mandibular central and lateral incisors, the right canine, and both premolars, while maxillary teeth showed no such deposits apart from moderate dental wear. High levels of wear were observed on the right canine, left central incisor, and first premolar. The third molar affected by microdontia displayed minimal wear.

This young adult female presents a case of unilateral microdontia affecting the right maxillary third right molar. Radiographic imaging confirmed that the microdontia involves the entire tooth, indicating a generalized reduction in size rather than localized anomalies in specific tooth structures such as the crown or root. In contrast, the left maxillary third molar appeared to exhibit normal morphology, as indicated by the dimensions of the left alveolus (Figure 2b). Apart from this anomaly, the individual's overall dental health was good. Dental wear was minimal across the dentition, consistent with the individual's young age.

SU 299

Individual SU 299 is a female aged between 40 and 45 years, with an estimated preservation status of 60%. Notable features included bilateral porosity surrounding the external auditory meatus and promontories, consistent with otitis media, and Schmorl's nodes on the superior surfaces of two thoracic and one lumbar vertebra. Dental anomalies included abnormal positioning of the left maxillary first premolar, which exhibited a vertical orientation, and the rotation of the right maxillary first premolar by approximately 45 degrees. Additionally, this individual presented bilateral hypodontia of the maxillary lateral incisors, and both maxillary third molars were absent (Figure 3a).

This female individual presents a clear case of bilateral hypodontia affecting the maxillary lateral incisors. Radiographic imaging confirmed the congenital absence of these teeth, with canines located adjacent to both maxillary central incisors. The identification of the canines was substantiated by their characteristically longer and more robust roots, distinguishing them from lateral incisors (Figure 3b). The absence of the lateral incisors resulted in morphological changes to the surrounding dentition. First premolars exhibited rotation, likely caused by the empty space on the alveolar ridge left by the missing lateral incisors. Specifically, the left first premolar displayed complete vertical rotation, whereas the right first premolar was rotated approximately 45 degrees. The underlying factors contributing to the differing degrees of rotation between the two premolars remain unclear. Despite these dental anomalies, the individual's teeth demonstrated overall good health. No evidence of carious lesions was observed across the dentition, and only minor dental wear and slight calculus deposition were noted. The latter was primarily localized and did not appear to compromise the individual's oral health significantly.

Discussion

Hypodontia can manifest in either syndromic or non-syndromic forms. Diagnosis typically arises when teeth fail to erupt within their expected timeframe (15). In the case of the individual from

SU 299, the absence of maxillary lateral incisors, which normally erupt between the ages of 8 and 9, strongly suggests hypodontia. Reported prevalence rates for hypodontia vary across populations, ranging from 2.6% to 11.3% (16).

The etiology of hypodontia is multifactorial, involving both genetic predispositions and environmental influences (16). A key theoretical framework used to explain hypodontia used to be Butler's Field Theory, initially developed in 1939 for mammalian teeth and later adapted by Dahlberg for human dentition. This theory categorizes teeth into distinct fields, each anchored by a genetically stable "key" tooth located at the first position within the group. Teeth positioned distally in the field, such as maxillary lateral incisors, are classified as genetically unstable, due to their higher susceptibility to developmental anomalies. Such an explanation aligns with the findings in SU 299, where these genetically unstable teeth (maxillary lateral incisors) are absent.

Microdontia can also occur in syndromic or non-syndromic forms depending on its association with birth defects (17). Some authors consider microdontia to be a mild form of hypodontia (18, 19, 20). Shafer et al. (1993) classify microdontia into three types: true generalized microdontia, relative generalized microdontia, and microdontia of individual teeth. True generalized microdontia, commonly observed in conditions like congenital hypopituitarism, ectodermal dysplasia, and Down syndrome, involves teeth that have a normal shape but are smaller than usual. Relative generalized microdontia features normal or slightly smaller teeth within disproportionately large jaws. Finally, microdontia of individual teeth – noted in the individual from SU 298 – typically affects a single tooth and can involve the lower incisors, canines, premolars, and upper molars (19, 21). Microdontia of a single tooth is further categorized into three types: microdontia of the entire tooth, microdontia of the tooth crown, and microdontia of the root (18, 19). The observed microdontia of individual teeth in SU 298 appears isolated and unassociated with syndromic presentations, as the skeletal morphology shows no abnormalities indicative of conditions like Down syndrome or ectodermal dysplasia.

Genetic factors

Both hypodontia and microdontia are often influenced by mutations in genes involved in dental development, such as MSX1 and PAX9. These genes regulate key processes in tooth formation, and their mutations can result in varying dental anomalies among family members due to differential gene expression. MSX1 is a homeobox gene that plays a regulatory role by influencing the transcription of other genes essential for tooth development. Mutations in MSX1 are associated with both hypodontia and microdontia (16). Mutations in PAX9 represent one of the most common genetic causes of tooth agenesis. Studies have documented more than 50 mutations linked to this gene (22). Interestingly, PAX9 mutations are associated with the agenesis of maxillary teeth more frequently than mandibular teeth (22). In addition to the effect PAX9 mutation has on agenesis of teeth, it has been shown that tooth size can also be influenced through that mutation (23). Brook's (1984) work highlights the genetic basis of hypodontia and microdontia, emphasizing that these anomalies occur more frequently among first degree relatives than in the general population, supporting a hereditary component. Brook also notes that females are more likely to exhibit the aforementioned conditions. Importantly, hypodontia and microdontia often co-occur, suggesting shared underlying genetic mechanisms. The clustering of hypodontia and microdontia in SU 298 and 299 raises the possibility of a familial relationship, supported by their burial in the same grave. However, without DNA analysis, this remains speculative.

Syndromic forms of these conditions often involve mutations in specific genes, such as the EDA gene (MIM# 300451), associated with X-linked hypohidrotic ectodermal dysplasia (XLHED; MIM# 305100). XLHED is a rare disorder characterized by sparse hair, underdeveloped sweat glands, tooth agenesis, and microdontia (24, 25, 26).

Environmental factors

Studies on monozygotic twins indicate that environmental factors, in addition to genetic predispositions, affect the expression of hypodontia (16). These influences may include intrauterine conditions, nutrition, or early developmental factors.

Absence of syndromic indicators

Both individuals exhibit normal skeletal morphology, with no signs of syndromes commonly associated with hypodontia or microdontia. All visible epiphyses are fused, indicating normal growth and development, and stature appears within the expected range of the population. The absence of craniofacial malformations, vertebral anomalies, or osseous

destruction further rules out conditions like Down syndrome or ectodermal dysplasia (21, 27, 28). In addition to the dental abnormalities, a benign variation was observed in individual SU 298, characterized by the presence of a sixth lumbar vertebra. This anomaly is worth noting regardless of whether the genetic background is known, and despite the fact that it cannot be linked to the dental anomalies. Such variations may still hold relevance in the broader context of morphological assessment and paleopathological interpretation.

Conclusion

This research provides a valuable contribution to the understanding of rare dental anomalies, such as microdontia and hypodontia, in archaeological contexts. The analysis of two female individuals (SU 298 and SU 299), discovered in a grave at the site of St. Euphemia in Rovinj, offers detailed insights into their dental health and anomalies. Identification of unilateral microdontia in SU 298 and bilateral hypodontia of the lateral incisors in SU 299 highlights not only the rarity of these conditions, but also their potential interconnection. These two individuals represent a unique example at this site, raising the possibility that they were biologically related.

Although supported by meticulous anthropological and radiological analyses, the absence of molecular and genetic studies limits the ability to draw definitive conclusions about the causes of these anomalies. Even so, the study associates microdontia and hypodontia with known genetic factors, such as mutations in the MSX1 and PAX9 genes, which play a key role in dental development. This research emphasizes the complex interplay between genetic and environmental factors in influencing the development of dental anomalies. Environmental influences, in particular, may contribute to the expression of these conditions.

Emphasizing an interdisciplinary approach in archaeological studies enhances our ability to identify and interpret dental anomalies. The combination of archaeological, anthropological, and radiological methods enables the precise identification and interpretation of dental anomalies, thereby expanding the boundaries of our understanding of the lives of past populations. The broader archaeological context of the site, which includes evidence of deviant burial practices and a variety of grave goods, enriches interpretations of the social and cultural dynamics in Rovinj between the 15th and 17th centuries.

Documentation and analysis of dental anomalies, such as those found in this case, not only offer data on the biological characteristics of individuals, but also provide a broader perspective on the genetic and environmental context in which they lived.

Declaration of Interest

None

Author Contributions

LV contributed to the conceptualization, data analysis, manuscript writing, visualization, and supervision of the study.

AU contributed to data analysis, manuscript writing, manuscript review and editing, as well as visualization.

Statement on the use of artificial intelligence in manuscript preparation

Artificial intelligence was not used in the preparation of this manuscript.

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