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TWIN STUDIES IN ORTHODONTICS: UNRAVELING THE GENETIC AND ENVIRONMENTAL ETIOLOGY OF MALOCCLUSION

Orthodontics

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ABSTRACT

Twin studies have long been a cornerstone in understanding the relative contributions of genetics (nature) and environmental factors (nurture) to various traits and conditions, including those in orthodontics. This review explores the role of twin studies in orthodontic research, highlighting key findings on the heritability of craniofacial morphology, malocclusion, and other dentofacial characteristics. The implications of these findings for clinical practice and future research are also discussed. Malocclusion, a deviation from the normal alignment of teeth, is a multifactorial condition influenced by genetic, environmental, and epigenetic factors. Twin studies have been instrumental in elucidating the relative contributions of genetics and environment to the etiology of malocclusion. This review explores the findings from twin studies on the etiology of malocclusion and discusses how these insights inform orthodontic treatment strategies. By understanding the genetic and environmental underpinnings of malocclusion, orthodontists can tailor treatments to address the specific causes of malocclusion in individual patients, leading to more effective and personalized care.

KEYWORDS

Twin Studies, Genetics, Monozygotic twins, Nature Vs Nurture

INTRODUCTION

The exploration of dentofacial development and malocclusion has long been framed by the classical debate of genetic inheritance versus environmental influence. Within orthodontic research, twin studies especially those comparing monozygotic (MZ) and dizygotic (DZ) twins have provided a robust methodological framework to distinguish the contributions of heritable and non-heritable factors. Through these comparisons, researchers have been able to estimate the heritability of various craniofacial and occlusal characteristics with increasing precision.

Initial investigations emphasized a dominant genetic role in the development of malocclusion, particularly for skeletal Class III patterns and specific cephalometric traits [1]. However, emerging evidence from more recent studies suggests that environmental and epigenetic factors—including prenatal conditions like chorionicity play a substantial role in shaping dentofacial features, particularly in dental alignment and occlusal discrepancies [2][3].

This review synthesizes current findings from the literature, with emphasis on recent twin studies and longitudinal comparisons of identical twins subjected to different treatment regimens. For instance, the case report by Sugawara et al. examined identical twins managed with one-phase versus two-phase orthodontic treatment and highlights on the predominant influence of inherent growth patterns over the modifying effects of treatment timing suggesting that morphogenetic factors remain the primary drivers of jaw development [4].

Parallel findings from systematic reviews reinforce that while certain intra-arch traits such as arch width, length, and mandibular crowding show moderate to high heritability ($h^2 = 0.35–1.0$), inter-arch traits like overjet, overbite, and molar relationships are more heavily influenced by environmental factors [3]. This distinction is clinically significant, as it provides insight into which traits may be more amenable to orthodontic intervention and which may require more comprehensive or surgical approaches. By delineating the genetic and environmental etiologies of dentofacial anomalies, twin studies continue to serve as an essential tool in orthodontic research.

Design Approaches In Twin Studies

Twin studies are essential for discerning genetic and environmental influences on dentofacial traits. The classical twin design compares monozygotic (MZ) and dizygotic (DZ) pairs to quantify heritability through structural equation modeling, analyzing additive genetic, shared environmental, and unique environmental components [3].

A critical methodological advancement accounts for chorionicity—accounting for placental sharing (monochorionic) in MZ twins, which otherwise inflates genetic estimates. This adjustment is particularly important for prenatal environment-sensitive traits like craniofacial development [2]

Longitudinal twin studies provide additional insight by tracking phenotypic changes across time. These designs are instrumental in distinguishing static genetic effects from dynamic environmental influences and are particularly valuable in orthodontics for evaluating age-related progression of malocclusion and skeletal development.

Twin-reared-apart studies provide a unique natural experiment where shared genetics but distinct environments enable clear attribution of dentofacial traits to heredity, offering strong evidence for genetic influence on craniofacial development [4]

Understanding Zygosity And Phenotypic Expression In Twin Research

Zygosity forms the basis of classification in twin studies and plays a pivotal role in interpreting heritability estimates. Monozygotic (MZ) twins, arising from a single fertilized ovum that divides into two embryos, share nearly 100% of their genetic material. This genetic uniformity makes them particularly useful for estimating the influence of environmental variables. In the referenced study, MZ twins displayed minimal intrapair variation in key cephalometric parameters, such as total anterior facial height (TAFH) and mandibular plane angle, supporting a strong genetic component in vertical skeletal traits [5](Figure1).

By contrast, dizygotic (DZ) twins, formed by the fertilization of two separate ova, share approximately 50% of their segregating genes—similar to ordinary siblings. The inclusion of DZ twin pairs in research facilitates comparisons that help differentiate environmental

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influences from genetic contributions. In the current study, DZ twins demonstrated greater variability in linear and angular cephalometric traits, such as the lower incisor to A-Pog distance and interincisal angle, pointing to the involvement of both heredity and environment [3].

In addition to genetic classification, twins are often categorized based on their phenotypic concordance. Concordant twins both exhibit the same trait—for example, identical malocclusion class—implying a stronger genetic underpinning. Discordant twins, where only one sibling expresses the trait, provide evidence for environmental or epigenetic modulation(Figure 2).A study found that MZ twins had high concordance for skeletal features, whereas discordance was more pronounced in DZ pairs, particularly in dental and occlusal traits [3].

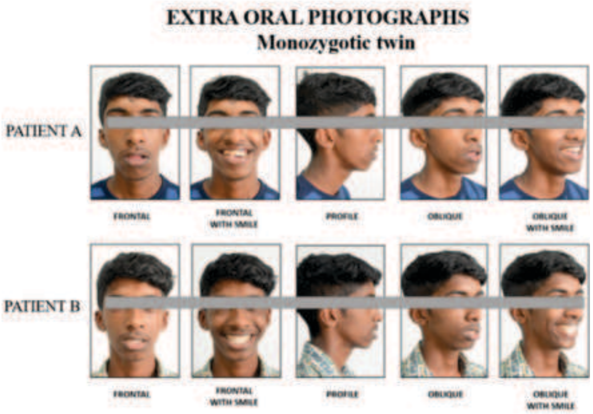

Figure 1: Comparison of Extra-oral Photographs of Monozygotic Twins

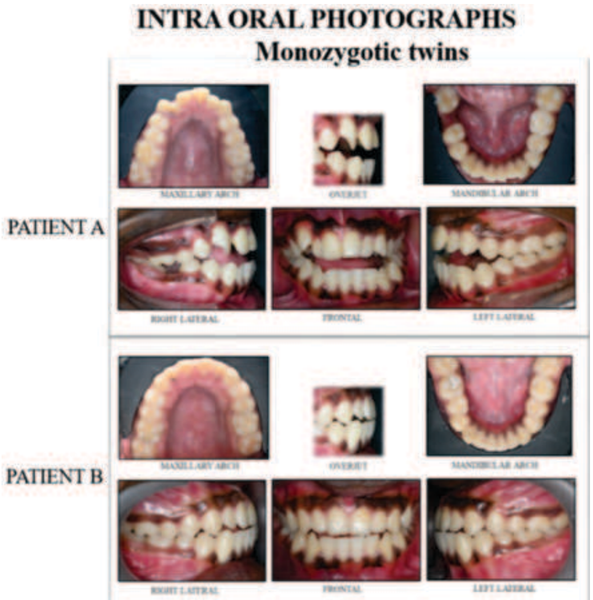

Figure 2: Comparison of Intra-oral Photographs of Monozygotic Twins

The impact of chorionicity further enhances the interpretation of twin study data. Among MZ twins, MC pairs—who share the same placenta—experience more homogenous prenatal environments, which can influence craniofacial symmetry and dental arch form[6]. However, these conditions may also introduce variations due to unequal nutrient distribution through shared placental circulation. Notably, MC twins in the study showed greater discordance in dental arch asymmetry, highlighting the complex interplay of prenatal factors on postnatal dentofacial development [2].

Heritability of Dentofacial Phenotypes

Intra-Arch Traits

Maxillary arch width and length demonstrate high heritability, with estimates ranging from $h^2 = 0.16$ to 1.0 [1,3]. Mandibular crowding exhibits moderate heritability ($h^2 = 0.35–0.81$), while maxillary crowding appears to be more environmentally influenced [3].

Inter-Arch Traits

Overjet and overbite are largely governed by environmental factors, with low heritability values ($h^2 < 0.2$) [1]. Posterior crossbite shows conflicting data; some studies report high heritability ($h^2 = 1.0$), while others find no significant genetic correlation [1,3].

Malocclusion Classes

Class II Division 1 malocclusion demonstrates moderate heritability ($h^2 = 0.3–0.5$), with notable environmental contributions, such as thumb-sucking [3]. Class II Division 2 displays a stronger genetic component ($h^2 = 0.6–0.7$), particularly in retroclined incisors and deep bite patterns [1]. Class III malocclusion is highly heritable, especially with mandibular prognathism ($h^2 = 0.5–0.8$) [3].Comparison of monozygotic twin's orthopantogram depicts the variations in the dentition (Figure 3)

Cephalometric Comparisons In Monozygotic Twins

Cephalometric evaluation offers a precise method to quantify craniofacial morphology and differentiate between genetic and environmental contributions. Standard parameters include linear dimensions such as total anterior facial height (TAFH), upper and lower anterior facial heights (UAFH, LAFH), and posterior facial height (PFH). Angular measurements include the saddle angle (N-S-Ar), mandibular plane angle (SN-MP), and interincisal angle (IIA). Dentoalveolar indicators like upper incisor to SN (UI-SN) and lower incisor to mandibular plane (LI-MP) angles, as well as vertical molar position, are also commonly analyzed.

Vertical skeletal parameters such as TAFH and LAFH consistently show higher heritability ($h^2 > 0.7$) than horizontal measurements, according to Manfredi et al. [5]. Sreedevi et al. further supported this with findings indicating high heritability in mandibular parameters, such as the mandibular plane to Frankfort plane (MP-FP) angle ($h^2 = 0.71$), while maxillary anterior heights were more influenced by environmental factors (UAFH, $h^2 = -0.36$) [5].

The distinction between genetic and environmental control of cephalometric traits is essential for treatment planning. High heritability in features like the Y-axis angle ($h^2 = 0.94$) suggests strong genetic influence, while traits like UAFH show greater susceptibility to environmental modulation. Dental occlusion tends to vary even ingenetically identical twins, likely due to environmental factors like muscle activity, oral habits, and functional adaptations [3](Figure 4).

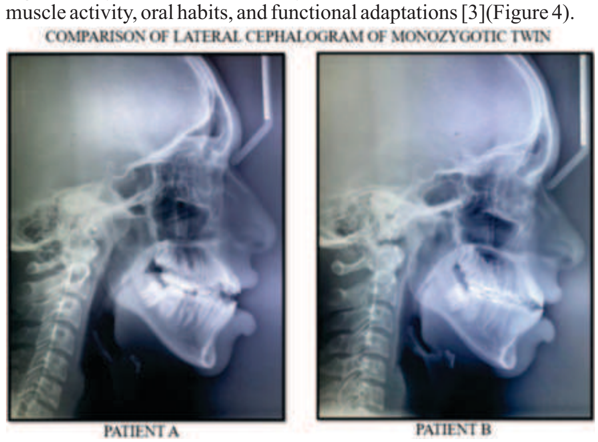

Figure 3: Comparison of Orthopantomogram of Monozygotic Twin

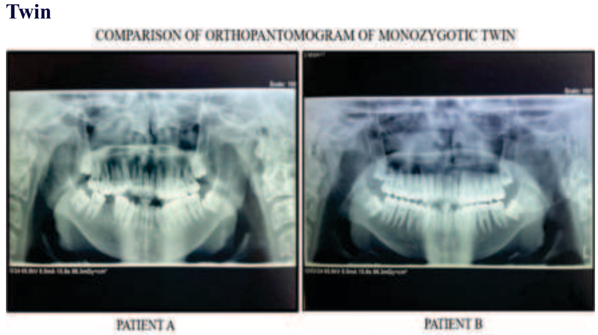

Figure 4 Comparison of Lateral Cephalograms of Monozygotic Twins

Clinical Implications of Heritability in Orthodontic Diagnosis and Treatment

Understanding the heritability of dentofacial traits has profound clinical relevance for orthodontic diagnosis, treatment planning, and patient counseling. Twin studies, by distinguishing genetic influences from environmental ones, provide an evidence-based framework for predicting treatment response and identifying traits that may require individualized intervention strategies.

Highly heritable craniofacial parameters, such as mandibular plane angle and total anterior facial height, often show limited response to orthopedic modifications due to their strong genetic basis. This suggests that patients exhibiting such traits may benefit more from surgical or skeletal anchorage-based interventions rather than conventional growth modification techniques. For example, vertical growth patterns with high heritability require early identification and may necessitate aggressive control measures or even orthognathic intervention in non-growing patients.

Conversely, traits with low heritability—such as overjet, overbite, and maxillary incisor inclination—are more environmentally influenced and, therefore, more amenable to orthodontic correction through habit elimination, functional appliances, and timely intervention. Recognizing these distinctions allows clinicians to tailor treatment plans based on the likelihood of modifying a given trait, thereby enhancing treatment predictability and long-term stability.

Additionally, the study of craniofacial asymmetry through cephalometric evaluations in twins reveals that such variations are predominantly environmental in origin. This reinforces the potential for successful correction using functional therapy and orthodontic appliances, particularly when identified early in development. Insights into the environmental modulation of dental traits further underscore the importance of monitoring oral habits, muscular function, and early developmental factors[7].

Ultimately, the integration of heritability data into clinical orthodontics not only informs more precise diagnosis and prognosis but also fosters a move toward personalized treatment strategies. Future incorporation of genetic and epigenetic testing may further refine this approach, paving the way for a more biologically informed and patient-specific model of orthodontic care.

CONCLUSION

Twin studies reveal that dentofacial phenotypes arise from complex interactions between genetic and environmental factors. While skeletal features such as mandibular length and vertical facial dimensions are predominantly influenced by genetic components, dental traits and occlusal characteristics are more susceptible to environmental modulation. Accounting for variables such as chorionicity and incorporating longitudinal data can further refine heritability estimates. As orthodontics moves toward more personalized approaches, insights from twin studies will continue to inform evidence-based diagnostics and individualized treatment planning.

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