
Non- Sindromic Oligodontia : A Case Report Of A Patient With Multiple Missing Teeth And A Literature Of Review

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Abstract

Agenesis of teeth is a developmental anomaly of the stomatognathic system characterized by the congenital absence of one or more teeth due to the failure of tooth germ formation during odontogenesis. This condition may affect both primary and permanent dentition and is classified according to the number of missing teeth into hypodontia, oligodontia, and anodontia. The method used in this report was a case report accompanied by a literature review based on relevant data sources. The case involved a 16-year-old female patient who presented with complaints of multiple spaces between her teeth and dental caries. Further examination, including panoramic radiography, revealed the congenital absence of nine permanent teeth: 17, 15, 24, 25, 27, 37, 35, 44, and 47. Clinical examination also showed the persistence of several primary teeth (55, 63, 65, and 75) with dentinal carious lesions but without mobility. This condition may result from mutations in genes involved in tooth development, such as MSX1, PAX9, and AXIN2. Mutations in these genes can disrupt epithelial-mesenchymal interactions during odontogenesis, leading to failure of tooth germ formation and resulting in multiple tooth agenesis. Oligodontia requires individualized management because its clinical manifestations vary considerably depending on the number of missing teeth, the condition of the alveolar bone, occlusal relationships, and the patient's age. Therefore, a multidisciplinary approach involving orthodontists, oral and maxillofacial surgeons, prosthodontists, and, when necessary, other dental specialists is essential to achieve optimal functional and esthetic outcomes.

Keywords: Oligodontia, Hypodontia, Agenesis, Gen mutation, Panoramic, Management

INTRODUCTION

Tooth agenesis is a developmental anomaly of the stomatognathic system characterized by the congenital absence of one or more teeth due to the failure of tooth germ formation during odontogenesis. This condition may affect both primary and permanent dentition and is classified according to the number of missing teeth into hypodontia, anodontia, and oligodontia.

Hypodontia is characterized by the congenital absence of one to five teeth, excluding the third molars. This condition occurs due to disturbances during odontogenesis that result in failure of tooth germ formation, causing permanent teeth to fail to develop normally. Hypodontia is the most commonly encountered form of tooth agenesis. The diagnosis of hypodontia is established through clinical examination and radiographic assessment, particularly panoramic radiography, to confirm the presence or absence of permanent tooth germs. Early detection is essential for determining an appropriate treatment plan because the management of hypodontia generally requires a multidisciplinary approach involving pediatric dentists, orthodontists, prosthodontists, and oral surgeons. Treatment options may include preservation of retained primary teeth with favorable prognosis, orthodontic treatment for space management, and prosthetic rehabilitation or dental implants after completion of craniofacial growth (Meade, 2023).

Anodontia is the most severe form of tooth agenesis, characterized by the complete failure of tooth development in both primary and permanent dentitions due to the absence of all tooth germs during odontogenesis. This condition differs from hypodontia, which involves the absence of one to five teeth, and oligodontia, which is characterized by the absence of six or more teeth (excluding third molars), as anodontia involves the complete failure of development of all dental elements. Anodontia is considered a very rare developmental dental anomaly compared with other forms of tooth agenesis. Etiologically, anodontia is primarily associated with genetic factors that disrupt the regulation of tooth development during odontogenesis. Tooth formation involves complex interactions between the oral

epithelium and ectomesenchymal tissues, which are regulated by various molecular signaling pathways, including WNT, Bone Morphogenetic Protein (BMP), Fibroblast Growth Factor (FGF), and Sonic Hedgehog (SHH) pathways. Alterations or mutations in genes involved in tooth development can lead to failure of dental lamina and tooth bud formation, resulting in the absence of all teeth. Anodontia is also frequently associated with genetic disorders such as hypohidrotic ectodermal dysplasia (HED), particularly due to mutations in the EDA, EDAR, and WNT10A genes, which play important roles in the development of ectodermal structures, including teeth, hair, skin, and sweat glands (Meade, 2023). The diagnosis of anodontia is established through a combination of clinical and radiographic examinations, particularly panoramic radiography, to confirm the absence of tooth germ development. Management of anodontia requires a multidisciplinary approach involving pediatric dentists, orthodontists, prosthodontists, and oral surgeons. Treatment aims to restore masticatory function, improve esthetics, enhance the patient's quality of life, and maintain the development of craniofacial structures. In young patients, removable prostheses are often used as an interim treatment option because skeletal growth is still ongoing, whereas definitive rehabilitation using dental implants may be considered after completion of craniofacial growth and when adequate alveolar bone conditions are achieved (Varghese et al., 2024).

Oligodontia is a more severe form of tooth agenesis, defined as the congenital absence of six or more teeth, excluding the third molars, and is associated with significant clinical consequences. This condition not only affects masticatory function but also has a considerable impact on esthetics, phonation, and the development of occlusion. Furthermore, oligodontia is frequently associated with genetic factors, including mutations in the PAX9, MSX1, and WNT10A genes, and may occur as either a non-syndromic condition or as part of a syndrome (Meade, 2023). The pathogenesis of oligodontia is closely associated with disturbances during odontogenesis, particularly during the initiation and early tooth germ formation stages. Normal odontogenesis involves complex interactions between the oral epithelium and neural crest-derived ectomesenchymal tissue, regulated by multiple molecular signaling pathways. Disruption of these epithelial–mesenchymal interactions may result in the failure of dental lamina formation or tooth bud development, ultimately preventing normal tooth germ formation. At the molecular level, oligodontia has been linked to abnormalities in several key signaling pathways, including the WNT/ β -catenin, Bone Morphogenetic Protein (BMP), Fibroblast Growth Factor (FGF), and Sonic Hedgehog (SHH) pathways, all of which play essential roles in regulating tooth cell proliferation, differentiation, and morphogenesis. Mutations in genes involved in these pathways, such as WNT10A, PAX9, and MSX1, can impair the expression of proteins required for normal tooth development, leading to arrested tooth formation during the early stages of odontogenesis (Yu M et al., 2022).

The management of non-syndromic oligodontia requires an individualized and multidisciplinary treatment approach because the severity of the condition varies depending on the number and distribution of missing teeth, the patient's age, craniofacial growth status, occlusal relationship, alveolar bone availability, and esthetic demands. The primary treatment objectives are to restore oral function, improve facial esthetics, maintain periodontal health, preserve alveolar bone, and enhance the patient's psychosocial well-being. Treatment planning should therefore be based on comprehensive clinical and radiographic examinations and coordinated among multiple dental specialties. Orthodontic treatment is often the initial phase of management to align the existing dentition, redistribute or close spaces, correct malocclusion, and establish an ideal occlusal relationship before definitive prosthetic rehabilitation (Aronovic et al., 2022).

Based on the description, this review focus on the clinical patient, radiographic, diagnosis. Accordingly, the purpose of this literature review is to describe the type of tooth agenesis, identify a case of agenesis, characteristic radiographic, and review effective management methods based on relevant scientific sources.

RESEARCH METHODS

This study employed a case report method accompanied by a literature review. The case report aimed to provide a detailed description of the clinical characteristics, supporting examinations, diagnosis, and management of a patient with non-syndromic oligodontia. Meanwhile, the literature review was conducted to compare the findings of the present case with current scientific evidence, thereby providing a more comprehensive understanding of the etiology, clinical characteristics, diagnosis, and management of non-syndromic oligodontia.

The case report data were obtained from the patient's medical records, including history taking, intraoral and extraoral clinical examinations, and supporting examinations using panoramic radiography. All clinical findings were analyzed to establish the diagnosis and determine an appropriate treatment plan based on the patient's clinical condition.

The literature review was conducted by searching scientific articles from several electronic databases, including PubMed, Google Scholar, ScienceDirect, and Wiley Online Library. The search strategy employed keywords relevant to the research topic, including *"tooth agenesis," "oligodontia," "non-syndromic oligodontia," "clinical features of oligodontia," "radiographic findings of oligodontia," "genetics of oligodontia,"* and *"management of oligodontia,"* either individually or in combination using the Boolean operators AND and OR.

The inclusion criteria consisted of scientific articles published within the last ten years, available in full text, written in English or Indonesian, and specifically addressing non-syndromic oligodontia in terms of etiology, clinical characteristics, radiographic findings, diagnosis, and management. The exclusion criteria included articles that were irrelevant to the research topic, unavailable in full text, editorial articles, commentaries, opinion papers, and studies containing incomplete or unreliable data.

The retrieved articles were screened based on the relevance of their titles, abstracts, and full-text content to the research topic. Articles that met the inclusion criteria were then subjected to a comprehensive full-text review, followed by data extraction on the etiology, clinical characteristics, radiographic findings, diagnosis, and management of non-syndromic oligodontia. This approach was intended to provide a comprehensive, evidence-based discussion of the diagnosis and management of non-syndromic oligodontia.

RESULTS AND DISCUSSION

Case Report

A 16-year-old female patient presented with a chief complaint of multiple spaces between her teeth and dental caries. The patient reported that the condition had been noticed since she entered junior high school and initially assumed that the spaces were due to delayed tooth eruption; therefore, she did not seek further dental examination. The patient stated that she had previously experienced tooth mobility, which resulted in tooth extraction, and that several teeth had carious lesions but were left untreated. Over time, the patient noticed that the spaces between her teeth became increasingly wider and that no replacement teeth erupted. The condition became more disturbing as she grew older, particularly affecting masticatory function, esthetics, and self-confidence.

Based on the anamnesis, the patient reported having a history of gastritis as a systemic condition and denied any history of allergies to medications, food, or environmental factors. The patient had previously visited a dentist during elementary school and underwent tooth extraction. She reported that her last dental visit was approximately seven to eight years ago. The patient brushed her teeth two to three times daily. The patient's parents had no history of systemic diseases; however, they reported dental complaints involving multiple carious teeth and tooth sensitivity, particularly when experiencing fatigue.

Intraoral examination revealed the presence of erupted permanent teeth in the maxillary right quadrant (region one), including teeth 11, 12, 13, 14, and 16. In the maxillary left quadrant (region

two), the erupted permanent teeth were 21, 22, 23, 25, and 26. In the mandibular left quadrant (region three), the erupted permanent teeth were 31, 33, 34, and 36. In the mandibular right quadrant (region four), the erupted permanent teeth were 41, 42, 43, 45, and 46. Further examination using panoramic radiography revealed congenital absence of nine permanent teeth, including teeth 17, 15, 24, 25, 27, 37, 35, 44, and 47. Clinical examination also showed the presence of retained primary teeth, including teeth 55, 63, 65, and 75, with dentinal-depth carious lesions but without tooth mobility. Percussion, probing, and palpation tests showed negative results. The patient reported no pain or discomfort; however, the condition interfered with mastication.



figure 1. Panoramic radiographic appearance



figure 2. Anterior intraoral photograph



figure 3. Maxillary occlusal intraoral photograph figure



figure 4. Mandibular occlusal intraoral photograph

Based on the clinical examinations performed, the patient was diagnosed with non-syndromic oligodontia, a condition characterized by the congenital absence of more than six teeth without any associated systemic disease. This condition is likely related to genetic factors, particularly mutations in genes involved in tooth development, such as *MSX1*, *PAX9*, and *AXIN2*. Mutations in these genes may disrupt epithelial–mesenchymal interactions during tooth development, resulting in failure of tooth germ formation and leading to multiple tooth agenesis (Biedziak et al., 2022).

Review of Literature

Hypodontia or tooth agenesis is defined as a common anomaly of human dentition characterized by the congenital absence of one or more teeth due to failure of tooth development. The term oligodontia is also used to describe the developmental absence of multiple teeth, which was previously considered to be commonly associated with systemic manifestations. However, according to the currently accepted definition, oligodontia is not always accompanied by systemic abnormalities and can be classified into syndromic oligodontia (associated with systemic manifestations) and non-syndromic oligodontia (without systemic abnormalities) (Meade, 2023).

Schalk-van der Weide stated that the congenital absence of six or more permanent teeth (excluding the third molars) can be used as a diagnostic criterion for oligodontia. According to the Consensus Conference on Oral Implants in Young Patients in 1996, the following definitions were established: hypodontia is defined as the absence of one to five permanent teeth, oligodontia refers to the absence of six or more permanent teeth, and anodontia refers to the absence of all permanent teeth. Therefore, oligodontia is generally defined as the congenital absence of six or more permanent teeth, with third molars (wisdom teeth) excluded from the calculation. Oligodontia is also referred to as partial anodontia, severe hypodontia, or advanced hypodontia. Some authors also describe this condition as selective tooth agenesis.

To date, no epidemiological studies have specifically reported the prevalence of oligodontia in the Indonesian population. This may be attributed to the rarity of oligodontia as a developmental dental anomaly. Available publications from Indonesia are still predominantly limited to case reports and reports on patient management, which are insufficient to represent the prevalence rate at the population level. Therefore, discussions regarding the prevalence of oligodontia generally refer to epidemiological data from various countries. Based on several international studies, the prevalence of oligodontia in the general population ranges from 0.08% to 0.36%. The reported prevalence is approximately 0.08% in the Netherlands, 0.10% in Finland, 0.19% in Sweden, 0.14% among Caucasian populations, and approximately 0.25% in Asian populations, particularly China (Castilho et al., 2023). Several studies have indicated that oligodontia tends to occur more frequently in females than in males. A retrospective study conducted in Japan reported that the prevalence of oligodontia was approximately 1.8% in females and 0.9% in males, suggesting a higher tendency of occurrence among females. However, other studies have not identified significant differences in the prevalence of oligodontia based on sex (Ariizumi et al., 2026).

Most cases of oligodontia are caused by genetic factors. Tooth development is regulated by complex interactions among various genes and signaling pathways during odontogenesis. Mutations in *WNT10A*, *PAX9*, *MSX1*, *AXIN2*, *EDA*, *EDAR*, and *EDARADD* genes have been shown to disrupt tooth germ formation, resulting in tooth agenesis with varying degrees of severity, ranging from hypodontia to oligodontia. Among these genes, *WNT10A* is the most frequently reported gene associated with mutations in oligodontia cases, whereas *PAX9* is considered the most common genetic cause identified in non-syndromic oligodontia (Kouroupakis et al., 2025). Non-syndromic oligodontia is predominantly inherited in an autosomal dominant pattern and is associated with mutations in genes involved in odontogenesis, particularly *PAX9* and *MSX1*. Among the various genes implicated in tooth agenesis, *PAX9* is one of the most frequently identified genetic causes of non-syndromic oligodontia. A systematic review published in 2023 reported that *PAX9* mutations are the most common genetic cause of tooth agenesis and that null mutations, including frameshift and nonsense mutations, result in a greater number of missing teeth compared with in-frame mutations. This finding

indicates that frameshift mutations lead to reduced or complete loss of PAX9 protein function, thereby disrupting odontogenesis and resulting in a more severe oligodontia phenotype (Intarak et al., 2023).

Oligodontia requires individualized management because its clinical manifestations vary widely, including the number of missing teeth, alveolar bone condition, occlusal relationship, and the patient's age, resulting in the need for different treatment plans for each case. Therefore, the management of oligodontia cannot be performed by a single dental discipline alone but requires a multidisciplinary approach involving orthodontists, oral and maxillofacial surgeons, prosthodontists, and other relevant specialists when necessary to achieve optimal functional and esthetic outcomes (Aronovich et al., 2022).

CONCLUSION

Oligodontia is a severe developmental dental anomaly characterized by the congenital absence of six or more permanent teeth, excluding the third molars, which can significantly affect masticatory function, esthetics, phonation, and occlusal development. Based on the case report using panoramic radiography revealed congenital absence of nine permanent teeth, including teeth 17, 15, 24, 25, 27, 37, 35, 44, and 47. The patient was diagnosed with non-syndromic oligodontia. Due to the diverse clinical manifestations involving the number of missing teeth, alveolar bone condition, occlusal relationships, and patient age, oligodontia requires individualized treatment planning. Successful management relies on a multidisciplinary approach involving orthodontists, oral and maxillofacial surgeons, prosthodontists, and other specialists to achieve optimal functional and esthetic outcomes.

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