Non-syndromic Oligodontia in Three Healthy Siblings With Decreasing Severity: A Case Series

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BACKGROUND

Oligodontia, defined as the congenital absence of **six or more** permanent teeth, is a highly heritable dental anomaly. Strong familial clustering has been reported, with a sibling recurrence risk of **43.8%**, supporting a predominantly **autosomal-dominant inheritance pattern with reduced penetrance**. While tooth agenesis overall shows a slight female predominance, this trend varies across cohorts. The teeth most frequently affected in oligodontia are the **second premolars**, followed by the **maxillary lateral incisors**, whereas the **maxillary central incisors** are rarely absent.

GENETIC BASIS

The genetic basis of oligodontia is heterogeneous, with pathogenic variants identified in genes such as ***WNT10A*** (~25% of cases), ***MSX1***, ***PAX9***, ***AXIN2***, ***LRP6***, ***WNT10B***, ***EDA***, ***EDAR***, and ***EDARADD***, all of which are integral to the ***WNT*** and ***EDA* signaling pathways** essential for tooth development.

Despite identifiable mutations, oligodontia demonstrates **marked variable expressivity and reduced penetrance,** even among relatives carrying the same variant. Monozygotic twins with identical ***MSX1*** mutations may present with different patterns of missing teeth, underscoring the role of epigenetic or environmental modifiers. Additionally, genetic synergism (e.g., ***PAX9*** with ***WNT10A***) can produce more severe phenotypes, whereas single heterozygous variants frequently show incomplete penetrance.

CLINICAL SIGNIFICANCE

Functional impairment is the most consistently documented impact of oligodontia. Affected patients scored significantly lower in the "eating and drinking" domain, and the more severe the condition is, the greater the resulting functional disability.

Psychosocial burden is substantial and multifaceted. Patients report that oligodontia negatively impacts making friends (45%), school participation (45%), public speaking (45%), and overall life satisfaction (64.5% reported life less satisfying pre-treatment).

Salivary dysfunction is prevalent, particularly in patients with ectodermal dysplasia features.

Associated dental anomalies (frequency):

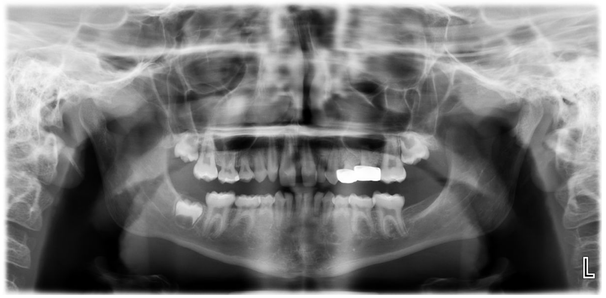
- **Primary molar ankylosis** (65.7%)
- **Taurodontism of mandibular first molars** (34.3%)
- **Enamel hypoplasia** (11.9%)**, conical incisors** (8.9%)**, and altered tooth size/shape**

CASE SERIES

Three siblings, ages **7** to **11 years**, presented to our clinic for routine initial examinations. Intraoral evaluation and subsequent radiographic analysis revealed a significant incidental finding: **nonsyndromic oligodontia** affecting all three children. Interestingly, the siblings demonstrate a striking gradient of **variable expressivity**; the eldest sibling is the most severely affected, while the youngest presents with a milder phenotype.

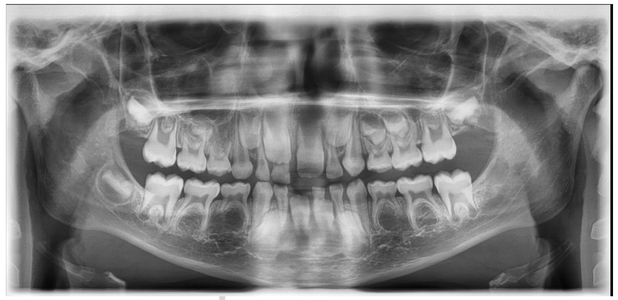
Sibling 1 - 11y 7m Male (Proband)

Dental Findings: Severe oligodontia, microdont #7,
Restorations present: zirconia crowns on #I and #J



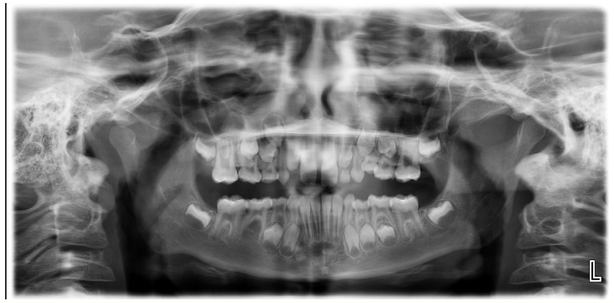
Sibling 2 - 9y4m Male

Dental Findings: Moderate oligodontia
Restorations present: None



Sibling 3 - 7y9m Male

Dental Findings: Mild hypodontia, Ankylosis of #I and #J
Restorations present: None



DENTAL TREATMENT

Despite the varying number of missing teeth, all three siblings share a synchronized clinical management plan focused on tooth preservation and long-term rehabilitation:

- **Aggressive Prevention:** All siblings are maintained on a **strict 3-month recall** schedule.
- **Minimal Intervention:** High-risk pits and fissures have been treated with **resin sealants**.
- **Home Care:** All siblings follow a rigorous, standardized oral hygiene regimen supervised by guardians.
- All three cases have been integrated into a **multidisciplinary "watch-list"** involving the Orthodontic and Prosthodontic departments. This proactive collaboration ensures that once dental development and skeletal growth allow, a seamless transition to functional and aesthetic rehabilitation can begin.

CONCLUSION

- This case series highlights the **critical role of early radiographic screening** in identifying incidental familial oligodontia, while illustrating how a possible shared genetic mutation can manifest as a broad clinical spectrum
- Oligodontia requires a **comprehensive interdisciplinary team approach** involving pediatric dentists, orthodontists, prosthodontists, oral and maxillofacial surgeons, and periodontists, with the prosthodontist or restorative dentist often taking the lead role in orchestrating treatment.
- **Management is staged** according to growth and development, beginning in early childhood and continuing through skeletal maturity.

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