10-year-old Male with Ectodermal Dysplasia 3 aka Wiktop Syndrome: A Case Report

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Resident: Jasmine Sandhu, DDS Faculty: Rodney Vergotine, DDS
University of Michigan School of Dentistry. Ann Arbor, Michigan – Mott's Children Hospital

BACKGROUND

Ectodermal dysplasias (EDs): Heterogeneous group of nearly 50 genetic disorders affecting the development of two or more ectodermal derivatives including hair, teeth, nails, sweat glands. and certain other tissues.

Causative genes and developmental pathways in all the Eds have been identified.

Classic EDs are caused by genetic variants affecting the ectodysplasin signal transduction pathway (*EDA*, *EDAR*, *EDARADD*, and *IKBKG*), *GJB6*, and *WNT10A*.

These include:

X-linked hypohidrotic ectodermal dysplasia, Ectodermal dysplasia and immunodeficiency 1, Ectodermal dysplasia 2 (Clouston type). More rare forms of EDs include tumor protein p63-related disorders and Focal dermal hypoplasia (Goltz syndrome).

The focus of this report is a case of Ectodermal Dysplasia 3, also known as Wiktop Syndrome or Tooth and Nail Syndrome (TNS). Inherited in an autosomal dominate pattern, 50% chance of passing on this mutation to an offspring; women and men affected equally. Witkop syndrome is caused by a change in the MSX1 gene located on chromosome 4p16. Characterized by both hypodontia, affecting both dentitions and nail dysplasia. Typical hair and sweat gland function. The primary teeth may be conical or narrow crowns and show prolonged retention. Secondary dentition may be partially or totally absent, especially the mandibular incisors, maxillary canines, and second molars

TNS may be suspected at birth if one or more toenails and/or fingernails are absent. The condition is diagnosed at around four or five years of age, when certain primary teeth are missing and underdevelopment of nails may be noted.

Clinical Presentation

A 10-year-2-month male presented to University of Michigan Pediatric Dental clinic with father.

Chief concern: Second opinion because previous dentist stated that patient needed "multiple extractions."

Medical history: Arthrogryposis multiplex congenita and a family history of ectodermal dysplasia.

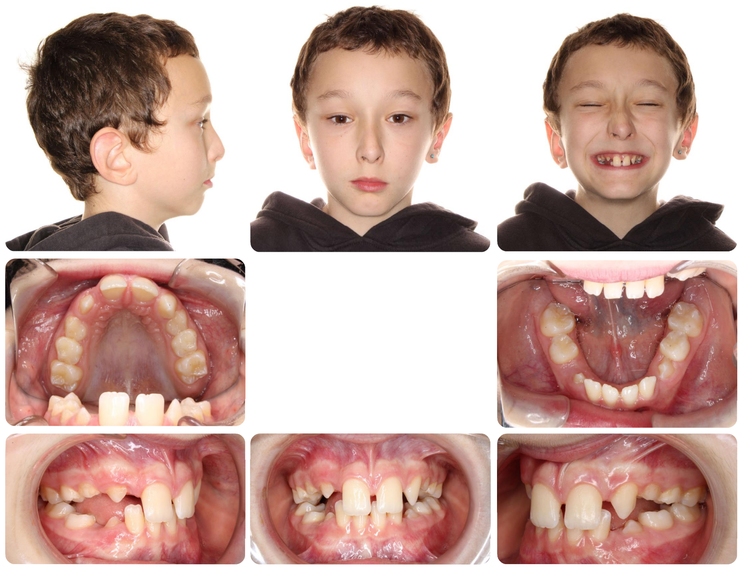
Father stated that both himself and patient's younger brother have been diagnosed with ED.

Treatment completed: Comprehensive oral exam, PANO, child prophy, and fluoride varnish.

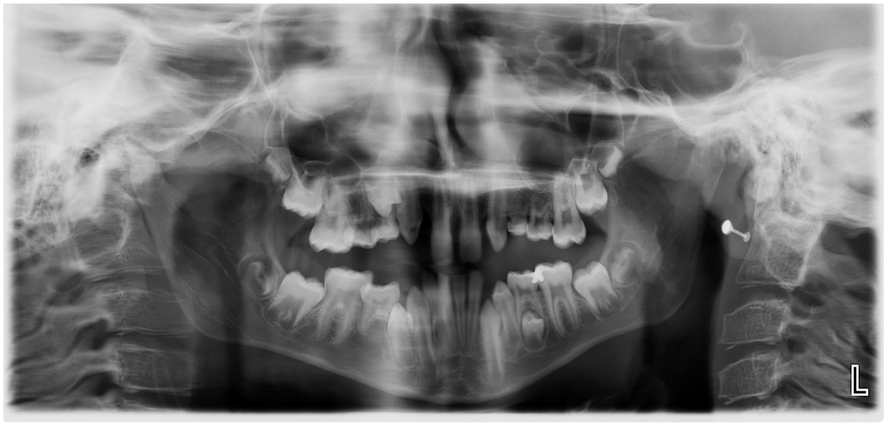
Carious lesions: #19-mesial and #K-distal.

Treatment recommended: #19-MO, #K-DO, graduate orthodontic referral to generate multi-displinary treatment goal/plan.

Extra Oral and Intra Oral Records



Radiographic Records



Record Analysis

Extra Oral Examination: No palpable lymphnodes, normal hair texture, volume, and pigmentation. Koilonychia (spoon) nails. Convex profile with a posterior facial divergence. Nose and Nasal dorsum are within normal limit. Upturned columella and obtuse nasolabial angle, with a retrusive lip position and upper lip curvature. Patient's upper lip curvature is thin and lower lip thickness is within normal limits. Patient's chin prominence is deficient and chin-throat line is weak.

Transverse Profile: Asymmetric midlines with the maxillary midline shifted 4mm to the left; a narrow nasal proportion, full commissure width, and wide smile esthetic.

Vertical Profile: Patient's proportions are short, incisal display is within normal limit, insical display at rest is 3mm and at smile is 12mm (100%), gingival display is excessive, with more gingival display on right compared to left. Gingival display at rest is 0mm and at smile is 4mm. No occlusal cant present. No TMJ or muscle pain. No popping, clicking, or crepitus noted. Patient has a maximal opening of 45mm, a right lateral excursive movement of 10mm, a left lateral excursive movement of 11mm, and a protrusive movement of 6mm.

Intra Oral Examination: Mixed dentition and has fair oral hygiene. No soft tissue lesions are present. Congenitally missing teeth #4, #7, #10, #12, #13, and #29.

Sagittal Profile: Bilateral class 1 molar, an overjet of 4mm

Transverse Profile: No posterior crossbite present

Vertical Profile: 50% overbite and a 6mm posterior open bite

X-Ray Analysis: Patient has all four developing third molar buds present. Patient's condyles and mandibular boarder are within normal limits. No bone loss, periapical lesions, endodontically treated teeth, pathological conditions, or supernumerary teeth present. Patient is at CVM stage 2.

Cast Analysis: Patient's arch form is tapered and upper arch width is narrow. Patient has spacing in both upper and lower arches of 7 to 8 mm, with a mild curve of Wilson and severe cure of spee. Tooth form is ovoid and tooth size is within normal limits.

Treatment / Management

Treatment plan options presented:

1. Non-surgical, Non-extraction, observe for pattern of eruption: wait for eruption of U3s, UR4, LL4 and decide on occlusal scheme after; fabricate activated lower lingual holding arch (LLHA) to help upright L6s; recall in 6 months to 1 year
2. Non-surgical, non-extraction of U3s, UR4, LL4 and decide on occlusal scheme after; fabricate activated LLHA to help upright L6s, 2x4 bracketing to close diastema between U1s and gain some initial alignment

Treatment objectives:

Facially: Maintain convex profile, maintain mentolabial fold, maintain slight chin deviation to the R

Skeletally: Maintain skeletal class 1, maintain Frankfort-mandibular plane angle (FMA)

Dentally: Maintain class 1 molars, maintain OJ and OB, achieve coincident midlines, and level lower curve of Wilson

Treatment of choice:

1. Non-surgical, non-extraction, observation and 6 month recall; in 6 months take another PANO (once L3s eruption), if any teeth are blocked out then patient will be sent to graduate pediatrics for extraction of LR-E and UR-D.

Since patient is missing multiple adult teeth, ortho and pediatrics should prioritize preserving the primary teeth that not caries free and have stable roots with no succedanous permanent tooth.

Ortho will work closely with prosthodontics to determine restorations with their combined goal to minimize amount of implants patient will need. Goal is to have 1 premolar is each arch, therefore UL quadrant will need **implant** since patient does not have premolar in that arch.

Ortho plans on doing **caning substitution** in upper arch to replace congenitally missing lateral incisors.

References

Majmundar VD, Baxi K. Ectodermal Dysplasia. [Updated 2023 Jul 24]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan-. https://rarediseases.org/rare-diseases/ectodermal-dysplasias/