文題目(出處)：	A case of Lacrimo-Auriculo-Dento-Digital Syndrome with multiple congenitally missing teeth. Case Rep Dent Volume 2016, Article ID 8563961
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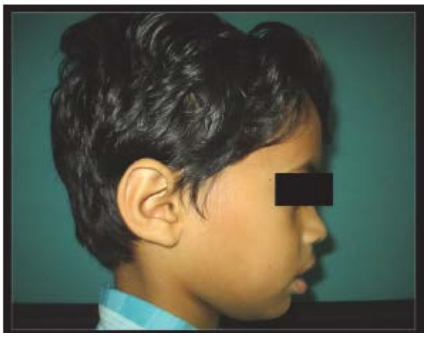
內文：

Introduction

1. Lacrimo-auriculo-dento-digital (LADD) syndrome, also known as Levy-Hollister syndrome, is a rare genetic disorder characterized by anomalies affecting the lacrimal and salivary glands and ducts, ears, dentition, and extremities.
2. Lacrimal gland
   (1) Most common : malformations including hypoplastic or aplastic lacrimal puncta and/or an obstruction of the nasolacrimal duct, excessive tearing (epiphora), inflammation of the tear sac (dacryocystitis), and dryness and inflammation of the cornea and conjunctiva (keratoconjunctivitis)
   (2) Less common : underdeveloped or missing lacrimal glands may result in lack of tears (alacrima) and dry eyes (xerophthalmia)
3. Slivary glands
   Undevelopment or absence may result in dry mouth (xerostomia) and vulnerability to severe dental caries.
4. Oral findings
   small teeth (microdontia), enamel hypoplasia, belated eruption, missing teeth .
5. Ears
   Cup-shaped, low-set ears maybe present associated with mild to severe hearing loss.

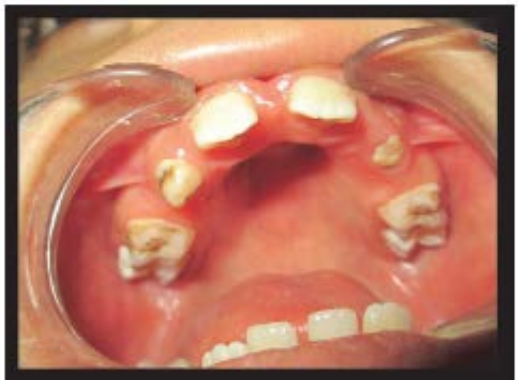
Case Report

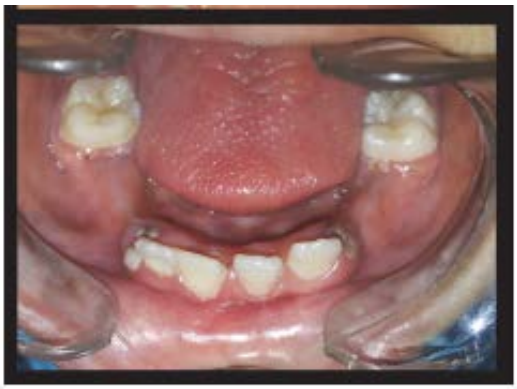
1. A 7-year-old boy complained of dryness of mouth since birth and decayed teeth. Additional concerns included dryness and itchiness in the eyes. His ears appeared to be normally located but were small and cup-shaped. Palpation of soft tissues in the submandibular and parotid regions revealed no perceptible enlargement.



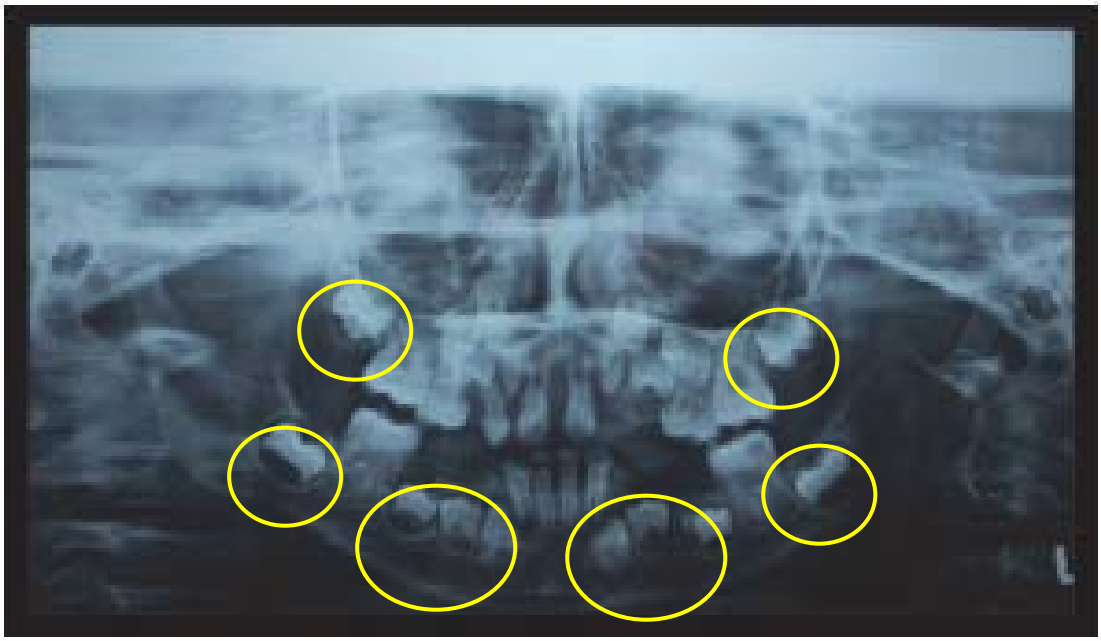
Intraoral examination :

Reduced saliva with multiple carious lesions and enamel dysplasia were observed. Several primary teeth were absent as per their chronological age of eruption.

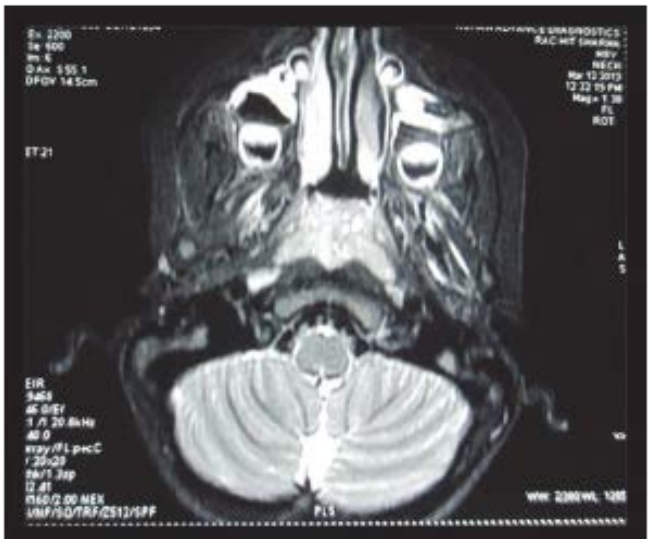




2. Panoramic radiograph finding :
   Several primary teeth were absent and tooth buds of unerupted permanent teeth were evident



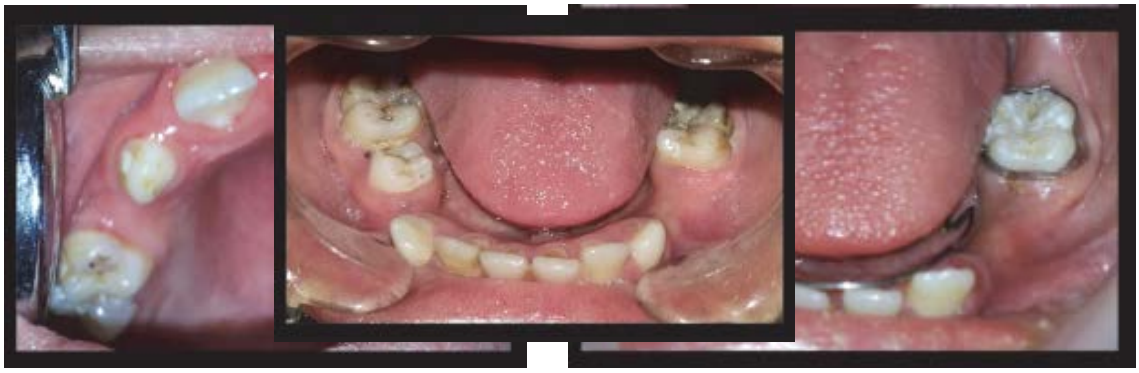
3. MRI scan :
   (1) aplasia of bilateral parotid and submandibular salivary glands and hypertrophied minor salivary glands along oropharyngeal wall.



(2) Lacrimal apparatus agenesis was also evident.

4. Treatment plan :
   (1) Restorations in relation to primary maxillary canines and second molars
   (2) Extraction of root stumps of mandibular canines
   (3) Band adaptation was done in relation to permanent molars
   (4) Alginate impressions for fabricating space maintainers
   (5) Prescribed fluoride mouthrinse and salivary substitute, and recalled.
   (6) The second visit :
   A lingual arch space maintainer was given in the lower arch, and topical fluoride varnish and resin-based pit and fissure sealants were applied
   (7) Patient refused an upper arch space maintainer due to apparent discomfort
   (8) Eruption of lower permanent canines and right second premolar after 3 years

After 3 years



Discussion

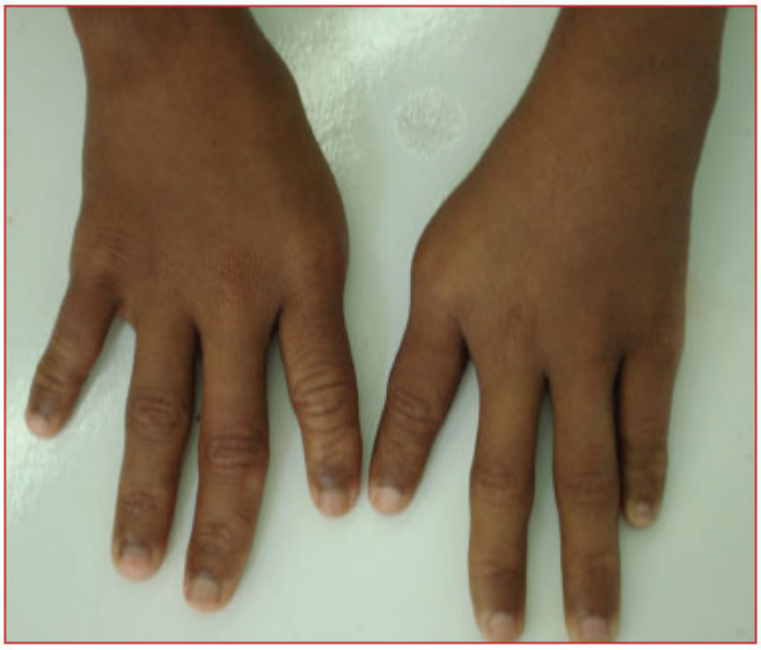
1. The absence of several primary teeth of this case
   (1) Maintenance of arch space is difficult due to increased caries risk and discomfort in wearing appliances because of diminished salivary flow.
   (2) Prescription of salivary substitute may enable the patient to wear the appliance comfortably
   (3) The appliance for a period sufficient to prevent pathological migration and provided space for eruption of permanent teeth.
2. LADD syndrome
   (1) an autosomal dominant disorder due to mutations in one of at least three genes, the fibroblast growth factor receptor 2 (FGFR2), fibroblast growth factor receptor 3 (FGFR3), and fibroblast growth factor
3. Overlapping clinical features and similar genetic etiology but are yet distinct from LADD syndrome.
   (1) Aplasia of the lacrimal and salivary glands (ALSG) presents with symptoms including xerophthalmia, xerostomia, scarring of the conjunctiva, dental erosion, periodontal disease, and increased risk of dental caries. ALSG is inherited as an autosomal dominant disorder caused due tomutation in the FGF10 gene.
   (2) Ectrodactyly-ectodermal dysplasia-cleft lip/palate (EEC syndrome) is another autosomally dominant syndrome and characterized by digital malformations, cleft palate, and cleft lip. Overlapping features with the LADD, including abnormalities of lacrimal ducts, chronic conjunctivitis, hypodontia, and/or microdontia .
   (3) Labyrinthine aplasia, microtia, and microdontia (LAMM) syndrome presents with Michel aplasia (complete bony and membranous aplasia of the inner ear) in association with microdontia andmicrotia. LAMM is

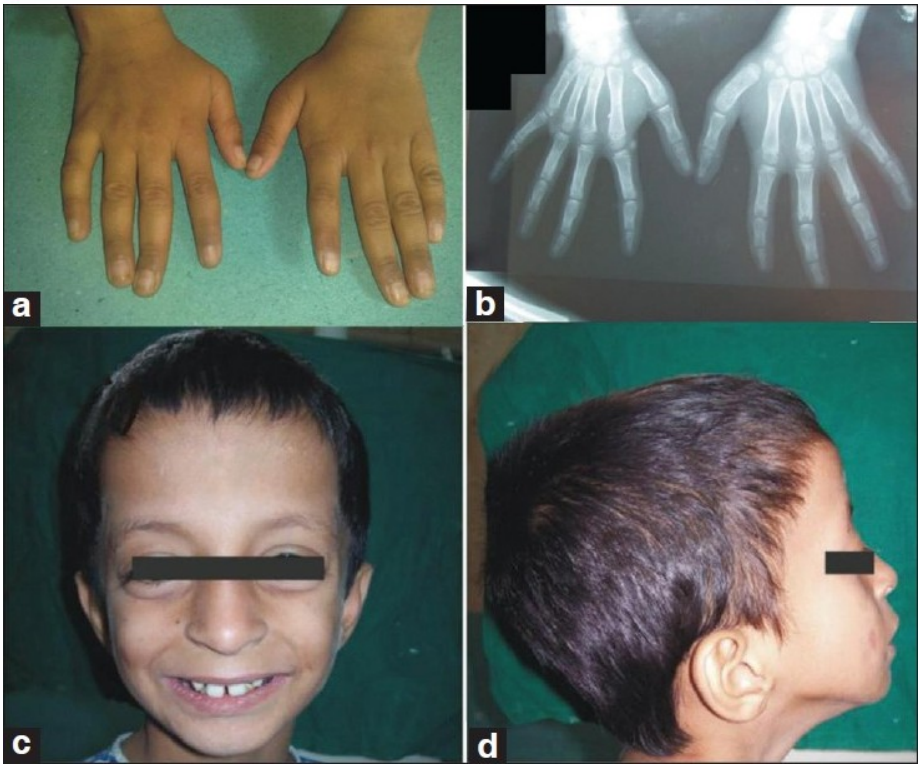
characterized by an autosomal recessive pattern of inheritance involving mutations in the FGF3 gene.

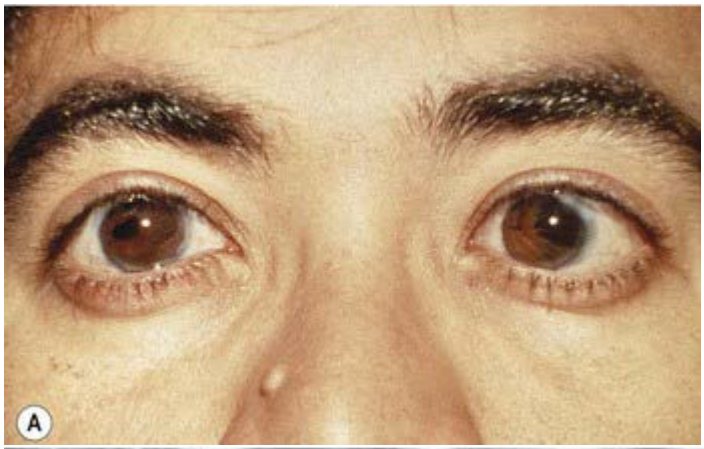
Conclusion

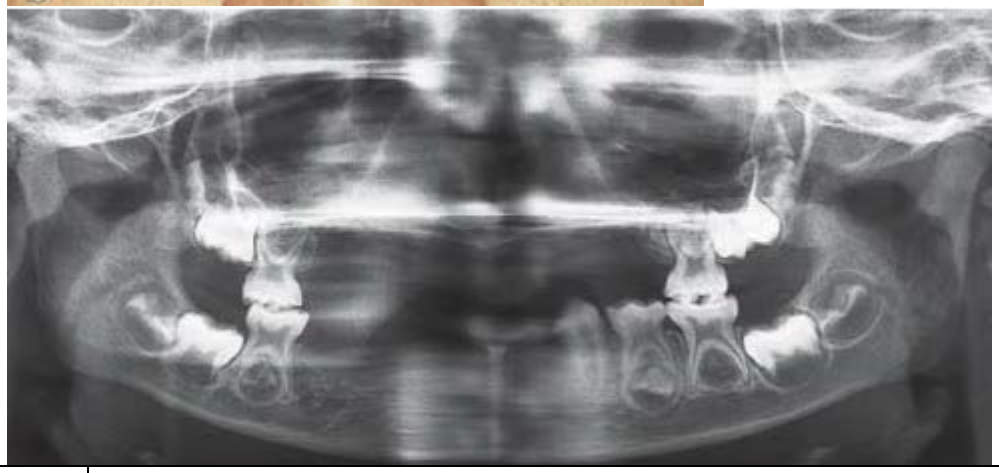
1. LADD syndrome is an extremely rare condition with characteristic oral, lacrimal, and auditory clinical findings. It requires a thorough evaluation to rule out the abovementioned similar conditions and determine presence of associated systemic manifestations.
2. Multiple missing primary dentition is an unusual association that requires comprehensive dental therapy.

Others









題號	題目
1	Absence of the major salivary glands can't be confirmed by (A) technetium-99m pertechnetate scintiscan (B) CT (C) MRI (D) Panoramic radiograph
答案(D)	出處：Oral and Maxillofacial Pathology, 3e ,p 453
題號	題目
2	Which is not mutated gene associated with LADD syndrome ? (A) FGFR2 (B) FGF11 (C) FGFR3 (D) FGF10
答案(B)	出處：Oral and Maxillofacial Pathology, 3e ,p 454