Ectrodactyly and Oligodontia a Rare Association

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ABSTRACT

A 29-year-old male patient reported with diastemata in the maxillary and mandibular arches since childhood. On general physical examination, a cleft was seen in place of the middle finger which was missing on both right and left hand. On intra-oral examination, multiple teeth were missing. Further, the orthopantomograph and the wrist radiograph revealed multiple missing teeth with the absence of a middle finger. There are many syndromes associated with partial anodontia and ectrodactyly, but the isolated occurrence of these two entities is not reported in the literature. Here, we present a case of a rare association of ectrodactyly and partial anodontia.

Key words: Ectrodactyly; missing middle finger; oligodontia

Introduction

Ectrodactyly is a rare congenital malformation of limbs with median clefts of hand, feet, and aplasia/hypoplasia of phalanges, metacarpals, and metatarsals.[1] It was first described in 1575 by Pare, and in 1884 Pott reported it in children from normal parents.[2] A large number of genetic defects can cause ectrodactyly, autosomal dominance with reduced penetrance being the most common mode of inheritance.[3]

Partial anodontia refers to agenesis of permanent teeth. An isolated association of ectrodactyly and partial anodontia is very rare.

Case Report

A 29-year-old male patient reported to the clinic with diastemata in the maxillary and mandibular arches. On general physical examination both right and left hands had a cleft in the center with the thumb, index finger, and the missing middle finger on the radial side while ring and little finger on the ulnar side were normal. Lower extremities and other parts of the body were normal, and on intra-oral examination multiple teeth were missing. Further, the patient was subjected for orthopantomogram (OPG) and radiographs of the hand and wrist [Figures 1 and 2]. On the OPG, multiple teeth were missing, and wrist radiograph of the right hand revealed a normal thumb and index finger on the radial side with normal ring finger and little finger on the ulnar side. The middle finger showed only the metacarpal with a complete absence of the phalanges-proximal, middle, and distal. The left hand showed normal thumb and little finger. The middle digit had a metacarpal with two proximal phalanges, one fused to index finger at the metacarpophalangeal, and the other one to the ring finger at the middle of the proximal phalanx.

Access this article online	
Quick Response Code:	Website: www.wajradiology.org
	DOI: 10.4103/1115-1474.164867

Differential diagnosis

Orodigital syndrome, acro-dermato-ungual- lacrimal-tooth (ADULT) syndrome and other syndromes associated with partial anodontia and ectrodactyly were considered under differential diagnosis.

Treatment

The patient was referred for orthodontic and prosthetic consultation for partial anodontia and to an orthopedician for correction of the ectrodactyly. The patient is under follow-up.

Discussion

An isolated association of ectrodactyly and partial anodontia together is very rare. Many cases have been reported about split hand-split foot malformations, orodigital syndrome, ADULT syndrome, and many other syndromes associated with partial anodontia and ectrodactyly, which also manifest with other oral and extraoral features. In our case, there were no other features except partial anodontia and wrist

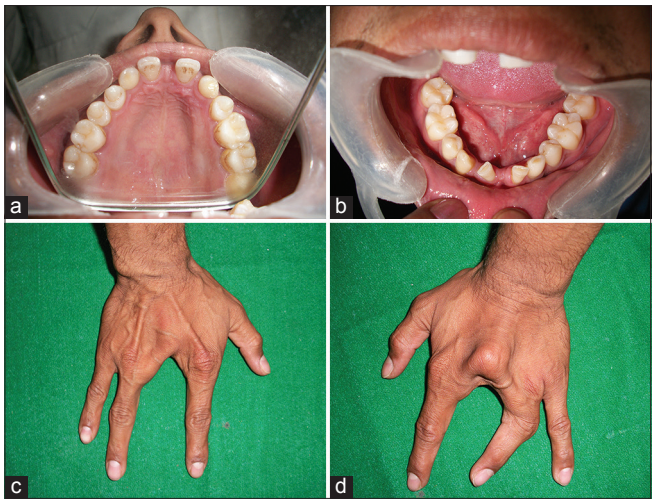

Figure 1: (a) Patient's intraoral photograph showing Maxilla with congenitally missing 13, 23, and 24. (b) Patient's intraoral photograph showing Mandible with congenitally missing 31, 34, 41, and 44. (c) Clinical appearance of right hand with cleft. (d) Clinical appearance of left hand with cleft

malformation. Ectrodactyly is a genetic defect due to deletion or translocation of chromosome 7. A study on mice reported that failure to maintain median apical ectodermal ridge signaling is the main cause of ectrodactyly. In humans, the only mutations known to underlie ectrodactyly is the TP63 gene.[3] The incidence is reported to be 1 in 90,000 babies without any sex predeliction.[1]

The etiology of partial anodontia is believed to be associated with genes,[4,5] and studies have shown that MSX1 and PAX9 genes play a role in early tooth development.[5] It is transmitted as an autosomal dominant, recessive or X-linked condition. Other factors such as endocrine, local, and environmental have also been suggested to play a role in the etiology of partial anodontia.[4]

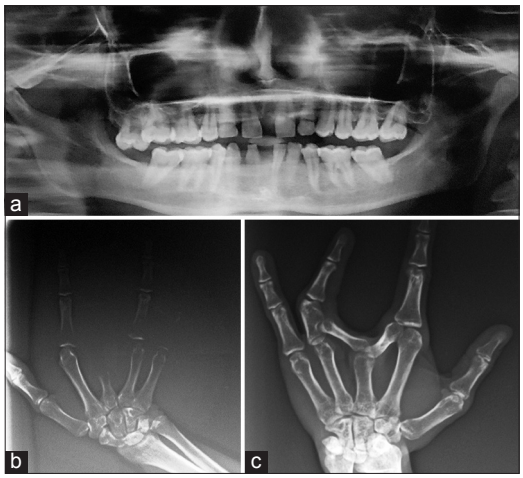

Figure 2: (a) Orthopantomogram showing missing 13, 23, 24, 31, 34, 41, and 44. (b) Posterior-anterior radiograph of the right hand showing absent middle finger phalanges and a partially absent metacarpal. (c) Posterior-anterior radiograph of the left hand showing the double proximal phalanx and anomalous fusion with adjacent bones

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How to cite this article: Najmuddin M, Saheb SA, John T, Parveen S. Ectrodactyly and oligodontia a rare association. West Afr J Radiol 2016;23:130-1.

Source of Support: Nil, **Conflict of Interest:** None declared.