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RESEARCH ARTICLE

ECTERODACTYLY ECTODERMAL DYSPLASIA CLEFTING SYNDROME – A CASE REPORT

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ABSTRACT

The ectodermal dysplasias (EDs) are a large and complex group of diseases. More than 170 different clinical conditions have been recognized and defined as ectodermal dysplasias. Commonly involved ectodermal-derived structures are hair, teeth, nails, and sweat glands. In some conditions, it may be associated with mental retardation. We report a case of 10-year-old male child with ectrodactyly, syndactyly, ED, cleft lip/palate, hearing loss, and mental retardation.

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INTRODUCTION

A 13 year old male patient had come to the op for correcting his bilateral cleft lip, He had already undergone a surgery for cleft palate 3 years back. A bilateral cleft was evident on his lips extending posteriorly to the alveolus region (Fig 1). The cleft was W-shaped (Fig 3). The philtrum was seen protruding, resembling a beak like appearance (Fig 2). Patient had an expressionless face, constant lacrimation from his eyes and also pus discharge from his left ear which he says is only present since the surgery. Patient also experiences water seeping out of his nose while drinking water. He was slightly built under nourished, his hairs and eyebrows were sparse, absence of hair follicles in the skin. His skin appeared dry and scaly (Fig 5). His upper and lower extremities showed split hand and foot appearance respectively (lobster claw like appearance) (Fig 4). Syndactyly was also noticed in both the extremities (Fig 5). There was constant lacrimation from his left ear and there was pus discharge from his left ear. A provisional diagnosis of Ectodermal Dysplasia with cleft lip was given. Upon referring the literature a Differential Diagnosis of AEC syndrome, Limb-Mammary Syndrome, ADULT Syndrome, Ecterodactyly Ectodermal Dysplasia Clefting Syndrome was given.

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Photographs Profile

Investigations

OPG: OPG reveals retained deciduous in relation 14, 15, 24, 25, 35 and 45 with the underlying primary tooth bud missing. Impacted 34 and 44 with incomplete root formation evident. The coronal morphology of the teeth are altered with large pulp canals evident in all the first molars teeth. A discontinuity is seen the alveolar bone in relation to 12 and 22. And the discontinuity in 12 is seen extending to the right maxillary sinus trabaecular pattern evident in maxilla and mandible Generalized loss of lamina dura, evident (Fig 7).

RT LAT CEPH: Lat Ceph reveals hypoplastic maxilla and slightly prognathic mandible (Fig 8).

Final diagnosis: Ecterodactyly Ectodermal Dysplasia Clefting Syndrome.

Treatment Plan: The cleft lip was corrected by cheiloscopy procedure. But the other features were just recorded just for its interesting nature and not addressed. Patient was subjected to scaling and also RCT was done in the molars. Patient came back for review after 2weeks.But did not show up for subsequent follow up.

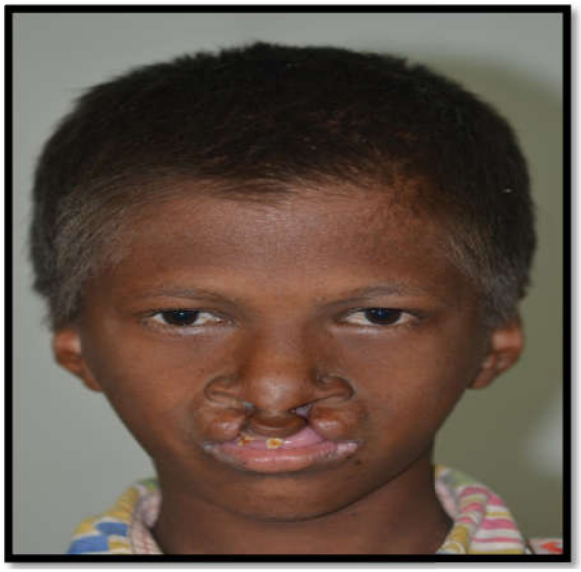


Figure 1. Frontal view

Figure 2. Lateral view

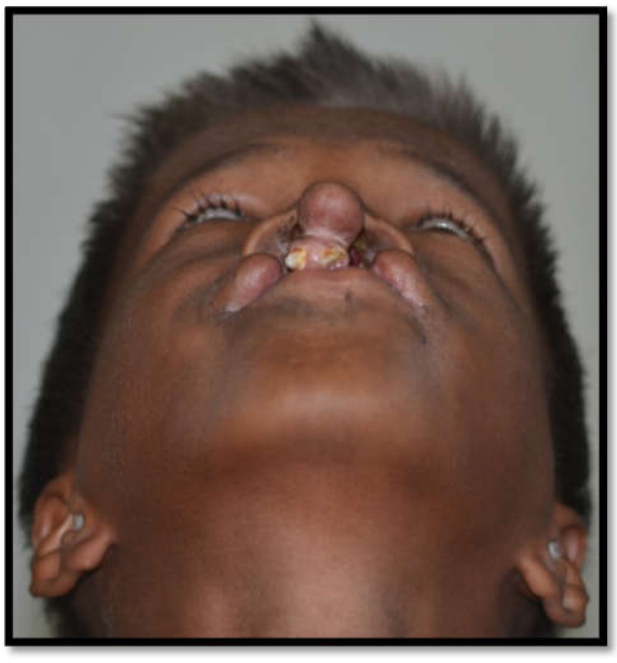
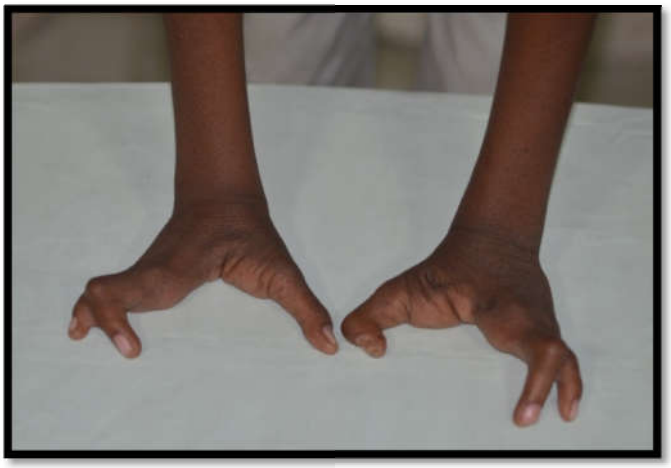


Figure 3. Upper & Lower Extremities



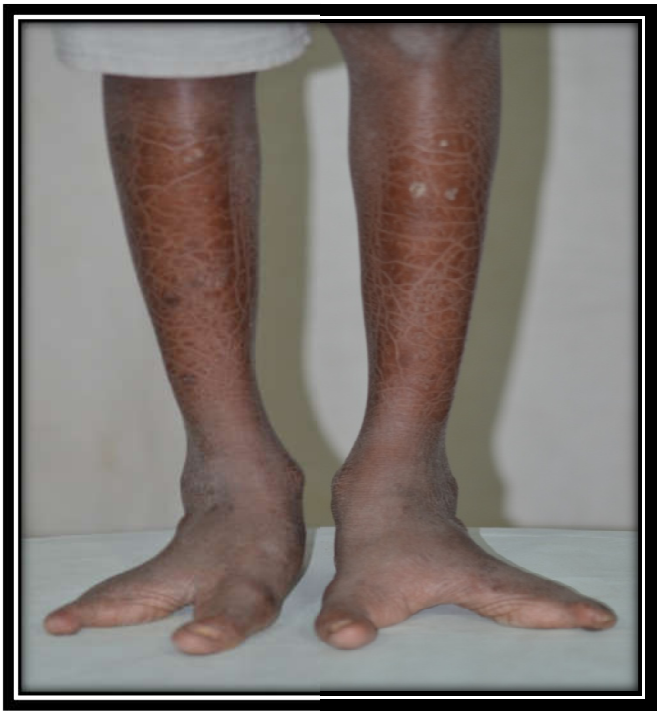


Figure 4 and 5 lobster claw with dry scaly skins

Intra Oral

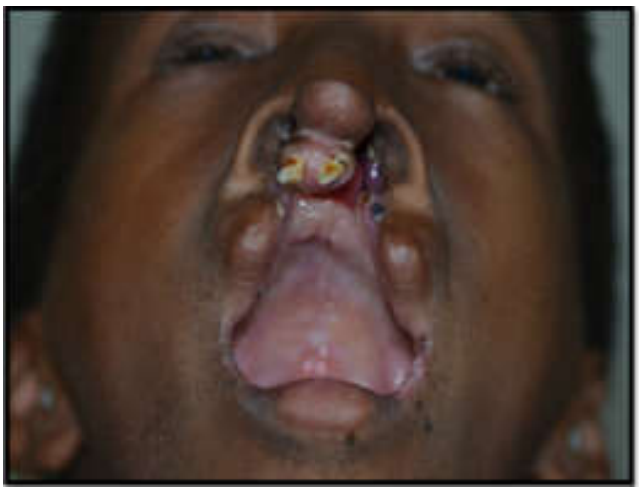


Figure 6.

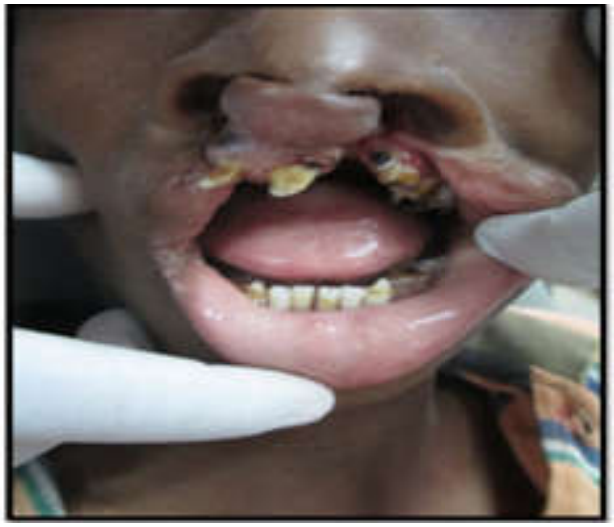
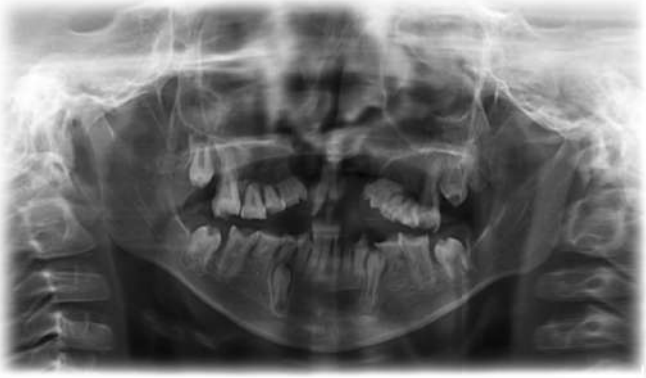


Figure 7

Figure 6 and 7 Opg and lat ceph x rays.

Lateral Cephalogram



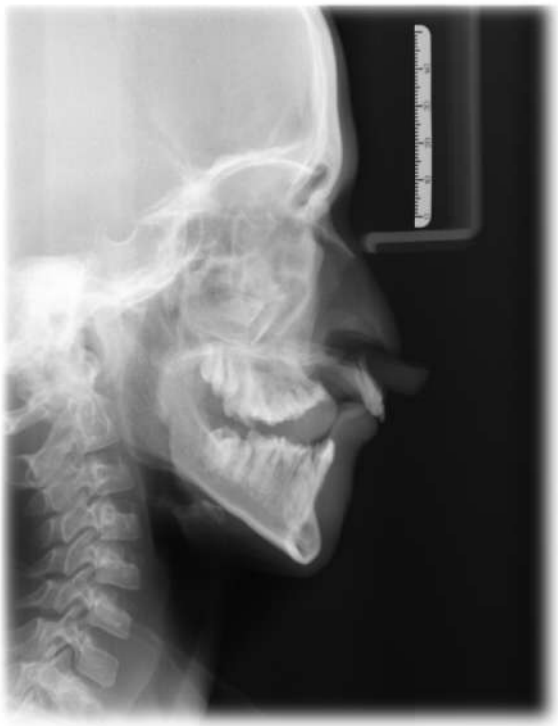


Fig 7 and 8 Opg and lat ceph.

Post OP

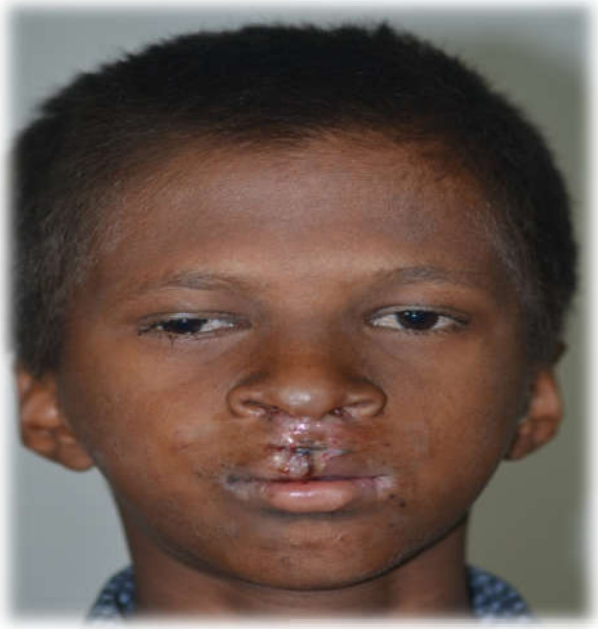


Figure 9 post op

DISCUSSION

EEC Syndrome

Synonyms

- Ectrodactyly-ectodermal dysplasia-clefting syndrome
- Ectrodactyly-ectodermal dysplasia-orofacial clefts

Ectrodactyly ectodermal dysplasia cleft lip/palate (EEC) syndrome is a rare genetic disorder. Symptoms can vary greatly from one person to another. Affected individuals often have abnormalities affecting the limbs including ectrodactyly, a condition in which part or all of the central digits (fingers or toes) are missing (Rosselli, 1961; Rudiger *et al*., 1970). Ectrodactyly often affects the middle fingers or toes, but can present differently in different people (or be absent altogether). A groove or gap in the upper lip (cleft lip) and a groove or gap in the roof of the mouth (cleft palate) may also occur. The ectodermal dysplasia component refers to abnormalities to structures that arise from the outermost layer of the embryo (ectoderm). In EEC syndrome, this generally affects the hair, teeth, nails, skin and sweat glands (Rudiger, 1970). Individuals with EEC syndrome can also develop a variety of additional symptoms including abnormalities of the genitourinary system and the eyes. Intelligence does not seem to be affected. Most cases of EEC syndrome are caused by mutations of the p63 gene and are either new (spontaneous) mutations or are inherited as autosomal dominant disorders (Rudiger, 1970; Freire-Maia, 1970).

Symptoms: Ectrodactyly, which is also known as split hand/foot malformation (SHFM), is a condition characterized by absence or malformation of one or more of the fingers or toes. Usually, the middle fingers or toes are affected Affected individuals have cleft lip with or without cleft palate. Additional distinctive facial features can occur including an undeveloped upper jaw (maxillary hypoplasia), a broad nasal tip, an abnormally long groove (philtrum) between the nose

and the upper lip, and narrowing or blockage of the nasal airways (choanal atresia) (Freire-Maia, 1970). The skin, hair, teeth, and sweat glands are commonly affected. Affected individuals may have dry, discolored (hypopigmented) skin. The skin may also be itchy. In some cases, mildly thickened, scaly patches of skin (hyperkeratosis) may also develop. Individuals tend to be fair skinned and have sparse, coarse, slow-growing scalp hair. Eyelashes or eyebrows may be sparse or absent. Additional symptoms can include slow-growing, thin, malformed (dysplastic) nails and missing, malformed or underdeveloped teeth (hypodontia). Tooth decay (dental caries) is common and often severe. Tooth enamel may be abnormal.[4] Abnormality of the sweat glands can lead to a reduced ability to sweat (hypohidrosis),. Abnormality of the salivary glands can lead to dry mouth (xerostomia) (Thurnam, 1848).

Genitourinary anomalies - Symptoms can include absence of the kidneys (renal agenesis), narrowing of the tubes that carry urine out of the body from the bladder (urethral atresia), and obstruction of the tubes (ureters) that carry urine from the kidney to the bladder, resulting in the accumulation of urine in the pelvis and kidney duct (hydronephrosis). An extremely uncommon genitourinary complication known as atrophic/dysplastic bladder epithelium has been reported in individuals with EEC syndrome. Intelligence is usually unaffected in children with EEC syndrome. Language development, however, may be delayed due to certain associated abnormalities such as cleft lip/palate or hearing impairment (Weech, 1929).

Etiology: EEC syndrome is inherited as an autosomal dominant trait. Some cases occur sporadically with no previous family history of the disorder (i.e., new mutations). Caused by mutations of the TP63 gene In rare cases, individuals with EEC syndrome carry chromosomal disruptions (deletions, translocations) on the long arm of chromosome 7 (7q11.2-q21.3) (Clarke, 1987). When EEC syndrome is caused by mutations of the TP63 gene it is sometimes referred to EEC syndrome type 3 (EEC3); when it caused by chromosomal abnormalities of chromosome 7 it is referred to as EEC syndrome type 1 (EEC1). A disorder formerly designated EEC syndrome type 2 no longer exists. In some cases EEC syndrome may be due to gonadal mosaicism, a condition in which some of a parent's reproductive cells (germ cells) carry the p63 or mutation, while others contain a normal cell line (mosaicism). The other cells (non-reproductive or somatic cells) in a parent's body do not have the mutation (Pinheiro, 1982). As a result, one or more of the parent's children may inherit the gene mutation, potentially leading to development of EEC syndrome, while the parent does not have the disorder (asymptomatic carrier) (Clarke, 1987; Pinheiro, 1982; Pinheiro, 1994).

Treatment: Treatment may require the coordinated efforts of a team of specialists. Pediatricians, pediatric surgeons, plastic surgeons, orthopedic surgeons, orthopedists, dentists, speech therapists, specialists that are trained to deal with abnormalities of the eyes (ophthalmologists), ears (audiologists), and skin (dermatologists), and other healthcare professionals may need to systematically and comprehensively plan an affect child's treatment (Rosenmann, 1976; Bowen *et al*., 1976). Reconstructive surgery may be beneficial for individuals with all defects causing functional disability such as ectrodactyly, syndactyly, cleft lip or palate and other associated facial anomalies (e.g., underdeveloped jaw, malformed ears). Dental surgery and corrective devices may be used to treat misshapen teeth. If teeth are missing, dentures may be necessary. Affected individuals should pay particular attention to dental health to prevent tooth decay. Artificial tears may be necessary for individuals with lacrimal duct obstruction. Surgery may also be necessary for blocked lacrimal ducts. Emollients may be used to treat dry skin. If hearing impairment is present, hearing aids may be beneficial. Children with hypohidrosis should be monitored closely for signs of hyperthermia, particularly during periods of prolonged activity and or during summer months. When hydronephrosis is present, temporary drainage of the urine may be necessary. Surgery may be indicated when pain or infection is present or when kidney function is compromised. Genetic counseling may be of benefit for affected individuals and their families.

Conclusion

Ectrodactyly—ED—cleft lip/palate (EEC syndrome) is a rare form of ED, the symptoms of which can vary from mild to severe. The most common symptoms found in patients with EEC syndrome are as follows: missing or irregular fingers and/or toes (ectrodactyly), abnormalities of hair and glands, cleft lip and/or palate, or unusual facial features, as well as abnormalities of eyes and urinary tract. For accurate diagnosis of patients with this rare syndrome, a careful and thorough examination should be carried out (Brill, 1972).

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