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Case Report

Ectrodactyly-ectodermal dysplasia clefting syndrome (EEC syndrome)

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ABSTRACT

Ectrodactyly-ectodermal dysplasia- clefting syndrome (also k/a. split hand- split foot malformation <SHFM>/split hand-split foot ectodermal dysplasia- cleft syndrome/ectodermal dysplasia cleft lip/cleft palate syndrome) a rare form of ectodermal dysplasia, is an autosomal dominant disorder inherited as a genetic trait and characterized by a triad of (i) ectrodactyly, (ii) ectodermal dysplasia and, (iii) & facial clefts.

1. Introduction

Ectodermal dysplasia is not a single disorder but a group of syndromes deriving from ectodermal structures such as skin, hair, nail and teeth. "Thurman" in 1848 published a first case report of dysplasia but the term " ectodermal dysplasia" was coined by "Weech" in 1929 and suggested 3 features **(1)** tissues affected are ectodermal in origin.**(2)** the defects are developmental anomalies.**(3)** and the hereditary tendencies are strongly developed.[1] More than 170 different syndromes have been identified. Despite some of the syndromes having different genetic causes, the symptoms are sometimes similar. The most common syndromes within this group are hypohidrotic and hidrotic ectodermal dysplasias. Several ectodermal dysplasia syndromes may manifest with mid-facial defects, mainly cleft lip and palate. Rudiger et al and Freire – Mai in 1970 reported a clinical condition called EEC syndrome .The two forms of EEC are found (a) cleft lip with or without cleft palate and (b) with cleft palate alone.[2] Both are inherited with variable expressivity i.e. partial penetrance in the former and complete penetrance in the latter.[3,4] More than

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300 cases have been reported in the literature, affects both males and females but the prevalence and incidence is not known.[5] Here is a case of EEC syndrome in a 5 year old male patient.

2. Case report

A 5 year old male patient reported to the department of Paedodontics and preventive dentistry in CPGIDSH, Lucknow, with a chief complaint of multiple decayed teeth. The family history was non-contributory and gave no history of consanguineous marriage between parents.

On general physical examination, the child was moderately built, had ectrodactyly of hands and feet showing lobster claw- like deformity, (also k/a split – hand and split –foot malformation SHFM) with difficulty in walking and holding with hands, presence of hypernasal voice and dry skin. Figs. 1 and 2. The parents also reported about reduced sweating, intolerance to heat, intermittent fevers and reduced amount of tears while crying.

Extraoral examination showed thin and sparse hair on the scalp that was lighter in color, frontal bossing, depressed nasal bridge, sparse eyelashes on the lower eyelids, dry and warm facial skin, and a surgical scar on the upper lip that was operated. Fig. 3.

Intraorally there were multiple grossly decayed and hypoplastic teeth, pointed crowns of canines, absence of maxillary primary lateral incisors and a repaired cleft palate. Fig. 4.

2.1. P/D

Considering the clinical signs and symptoms a provisional diagnosis of Ectrodactyly-ectodermal dysplasia clefting syndrome or EEC syndrome was made.

3. Investigations performed

(1) Radiographs
  (a) Orthopantomogram (OPG) Fig. 5(a)
  (b) A- P view of hands. Fig. 5(b)
  (c) A- P views of feet. Fig. 5(c)
(2) Sweat pore count Table 1
(3) Schirmer test Table 2

3.1. D/D

1. Rapp – Hodgkin syndrome.
2. Hay–Wells syndrome (AEC syndrome)
3. Adult syndrome
4. Non- syndromic split- hand and split foot malformation (SHFM)

4. Discussion

EEC syndrome a genetic developmental disorder distinctly featuring ectrodactyly, ectodermal dysplasia and facial clefts may also be noticed with characterstics like recurrent urinary tract infections, vesiculioureteral reflux, photobia, anomalies of kidney, hearing loss and speech defects.[6] During reproduction each parent contributes 23 chromosomes: 22 autosomal and one sex chromosome. EEC is autosomal which means that there is an abnormal gene on one of the autosomal (non-sex) chromosomes from either parent. The gene is dominant, only one parent must contribute the abnormal gene for the child to inherit the disease and the contributing parent will usually have the disease, due to the expression of the dominant gene in the parent. Although, it is an autosomal dominant, some cases occur sporadically without previous history of the disorder (i.e. new-mutations.) The present case

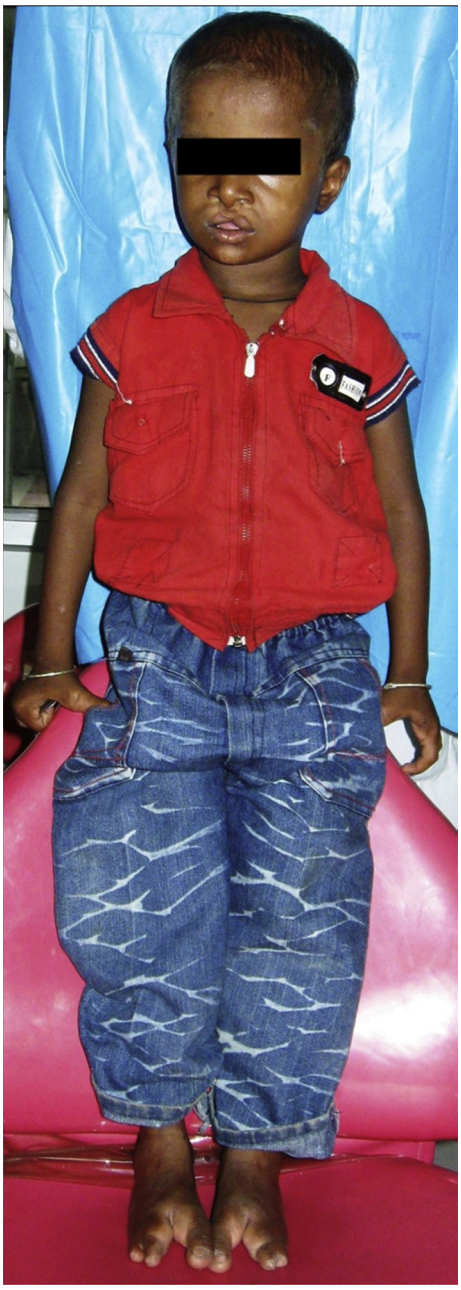

Fig. 1 – Patient with ectrodactyly of hands and feet.