Journal of Advances in Medicine and Medical Research

24(11): 1-4, 2017; Article no.JAMMR.37990
ISSN: 2456-8899
(Past name: British Journal of Medicine and Medical Research, Past ISSN: 2231-0614, NLM ID: 101570965)

An Incomplete Ectrodactyly Ectodermal Cleft Syndrome – A Case Report

Sudakshina Chakrabarti[1*] and G. Karthikeyan[1]

[1]Department of Anatomy, Saveetha Medical College, Chennai, India.

Authors' contributions

This work was carried out in collaboration between both authors. Author SC studied the case and findings in detail. Author GK helped with the write up and collecting references for this report. Both authors read and approved the final manuscript.

Article Information

DOI: 10.9734/JAMMR/2017/37990
Editor(s):
(1) Nurhan Cucer, Department of Medical Biology, Erciyes University, Turkey.
Reviewers:
(1) Murat Varli, Ankara University School of Medicine, Turkey.
(2) Neema Tiwari, King George Medical University, India.
(3) P. S. G. Prakash, SRM Dental College, India.
(4) Somchai Amornyotin, Mahidol University, Thailand.
Complete Peer review History: http://www.sciencedomain.org/review-history/22454

Short Communication

Received 6th November 2017
Accepted 14th December 2017
Published 26th December 2017

ABSTRACT

Ectrodactyly ectodermal dysplasia and clefting syndrome (EEC syndrome) is a rare genetic anomaly involving multiple structures and organ systems. It can be due to spontaneous mutation or autosomal dominant inheritance. The prevalence of this condition is 1-9 / 100 000 population. The common structures affected in this condition are the hair, nails, skin, eye, ear and the skeletal system.

This is the report of a 12 year old male child presenting to the paediatric OPD with complaints of hypohidrosis and febrile episodes past few months. There was no history of breathlessness or chest pain. On examination he was found to have alopecia, misalignment of teeth, clefting of both the lower extremities (absence of the second and the third toe on the right foot and the third toe on the left foot with syndactyly in both feet). There was no involvement of the eye and ear. There were no facial deformities. No cleft lip or cleft palate. External genitalia appeared normal. No abnormalities were found on renal ultrasound. The child was born of non consanguineous marriage. Younger sibling and parents are asymptomatic. He was provisionally diagnosed to have EEC syndrome.

**Corresponding author: E-mail: sudhianat@gmail.com;*

Keywords: Autosomal dominant; hypohidrosis; clefting; syndactyly.

1. INTRODUCTION

Ectrodactyly ectodermal cleft syndrome is a rare form of ectodermal dysplasia. The symptoms can varies from mild to severe and most commonly include missing or irregular fingers and or toes (ectrodactyly or split hand or foot malformation); abnormalities of the hair and gland, cleft lip and or palate, distinctive facial features; and abnormalities of the eyes and urinary tract [1,2]. This syndrome is inherited in an autosomal dominant manner though some authors have declared this as a X- linked recessive inheritance [3,4]. Approximately 90% of individuals with EEC syndrome have a mutation identified in the *TP63* gene. The *TP63* gene codes for the p63 protein, which plays a important role in early development of the ectoderm. The p63 protein is additionally thought to play a role in the development of the limbs, facial features, urinary system, and other organs. Individuals that have EEC syndrome due to a mutation in the *TP63* gene are classified as having EEC syndrome type 3 (EEC3). Whereas EEC 1 is caused by the mutation of long arm of chromosome 7 and this constitutes 10% of cases [5,6]. Though most of the cases of EEC are due to autosomal dominant inheritance there are instances of acquiring this syndrome as a denovo mutation in the germ cells of the unaffected parents a phenomena known as the germline mosaicism and this has lead scientist to think of a X linked recessive disorder in causing EEC syndrome [5,4]. EEC syndrome can result in a multitude of signs and symptoms in its expression in affected individual i.e. it has a variable expressivity. EEC syndrome has a reduced penetrance, all individuals carrying the affected gene will not have the disease and the penetrance is 93% to 98% thus 2 to 7% will be unaffected [5]. Approximately 170 or more variety of clinical presentations is reported in this complex and varied syndrome [3].

2. CASE PRESENTATION

A 12 year old boy presented to pediatrics outpatient department was referred to the Department of Anatomy of Saveetha Medical College for case study. The boy is the first child born to a non consanguineous couple with no history of similar complaints in any of his paternal or maternal ancestors. Patient presented with complaints of decreased sweating on and off febrile episodes with discomfort from past few months. A detailed general physical examination was performed and the findings are listed below. An informed consent was received from the patient's parents regarding publishing this case.

2.1 Skeletal Abnormalities

Bilateral ectrodactyly of the feet were present with absence of 2nd and 3rd toes on the right foot and 3rd toe on the left foot. The right foot had syndactyly of 4th and 5th toes and the left foot had syndactyly of 1st and 2nd toes and also of 4th and 5th toes. This is a classical feature of bilateral lobster feet deformity as captured in Fig. 1.

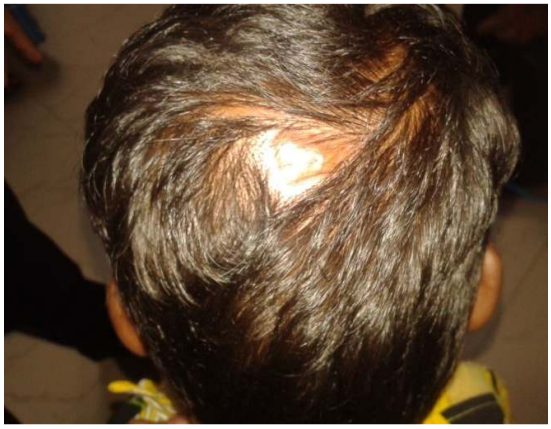

Fig. 1. Alopecia Areata

2.2 Skin Abnormalities

- Patient s skin has a dry scaly appearance with diffuse hypo pigmentation. The scalp hair appeared dry with presence of one patch of alopecia areata over the parieto-occipital region measuring about 4 cm by 3 cm as shown in Fig. 2. Nails were appearing normal. Sweat pore counts on the tips of digits were within normal limits [7]. Body hair was sparse to normal.

2.3 Oral Cavity

Patient had xerostomia with opening of Stenson's duct not visible clearly. Teeth count was normal but there was enamel hypoplasia with numerous caries and misalignment of teeth and abnormal small sized teeth were present as shown in Fig. 3. Patient was referred to orthodontic department for further examination and management.

No abnormality was detected in systemic examination. Heart sounds were normal and per abdomen examination was normal. Lung fields were clear. A diagnosis of EEC syndrome was made and patient was referred for investigations to confirm diagnosis by genetic testing.

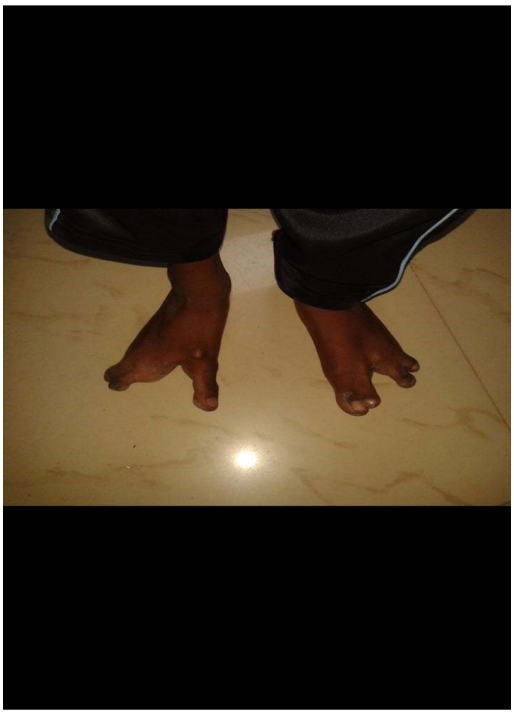

Fig. 2. Lobster deformity

3. DISCUSSION

Ectrodactyly is usually seen as either complete absence of or malformation of one or more fingers or toes. Patients generally have median cleft in upper and lower limbs, which makes the affected limbs look like a lobster claws and hence the name given. This lobster claw is thought to arise as a result of a wedge-shaped defect of the apical ectoderm of the limb buds [8]. Sometimes all four limbs involvement may be seen, even though this is a rare phenomenon. The patients of EEC may sometimes have webbing or fusion (syndactyly) of the fingers and/or toes as present in the present case [9]. The spectrum of dermatological manifestation associated with ectodermal dysplasia is variable and include hyper keratosis, thickened scaly skin to hypo pigmented dry skin with poor hair growth. Scalp hair as well as eyebrows may be sparse, wiry, and with hypo pigmented hair [10]. Other symptoms can include dysplastic nails and peg-shaped teeth with misalignment with are present in the above case. Tooth decay is very common clinical finding as seen in our patient and is often very severe and sometimes tooth enamel may be abnormal [11,12]. Other associated anomalies of urinary system, hearing loss, abnormalities of sweat, lacrimal and endocrine glands have also been reported [13,14,15].

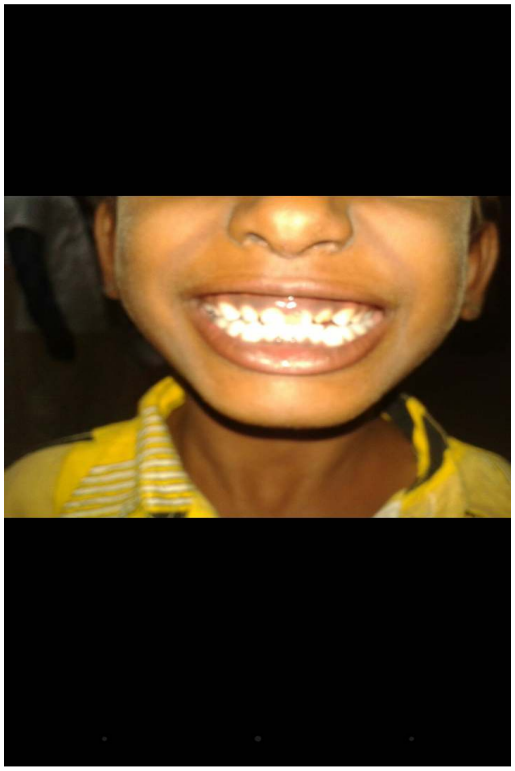

Fig. 3. Misalignment of teeth

The management of this condition starts with a multiple investigations as USG scanning for renal anomalies, 2D echocardiography, skin biopsy, thyroid function test, hearing tests, opthalmological evaluation, imaging techniques for affected limbs, and confirmation of this syndrome can be obtained by genetic testing for detection of mutation. The treatment is primarily supportive involving multidisciplinary approach. Limb reconstructive surgeries for ectrodactyly, syndactyly can be considered, artificial dentures for missing teeth, usage of artificial tears are some of the treatment options [13,14,15,7].

4. CONCLUSION

EEC syndrome is a multiple congenital anomaly syndrome with a varied presentation. The above case is a classical example of incomplete EEC syndrome that requires a multispeciality

approach in its management. Early detection and counseling can help to improve the mental IQ in these cases and parental genetic counseling and testing can be helpful to prevent recurrences in future pregnancies.

CONSENT

As per international standard or university standard, patient's written consent has been collected and preserved by the authors.

ETHICAL APPROVAL

Ethical approval for presenting and publishing this case has been obtained from the institutional ethical committee. Informed consent has been taken from the parents of the patients to publish the findings after maintaining confidentiality by not disclosing any personal information of patient or his relatives.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

REFERENCES

1. Ectrodactyly Ectodermal Dysplasia Cleft Lip/Palate. NORD; 2012. Available:http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/935/viewAbstract
2. Reid Sutton V, Hans van Bokhoven. TP63-related disorders. GeneReveiws; 2015. Available:http://www.ncbi.nlm.nih.gov/books/NBK43797/
3. Mohita Marwaha, Kanwar Deep Singh Nanda. Ectrodactyly, ectodermal dysplasia, cleft lip, and palate (EEC syndrome). Contemp Clin Dent. 2012;3(2): 205-208.
4. Shafer, Hine, Levy. Chapter 19 – Diseases of the skin. 5th ed. New Delhi: Elsevier Publications. Shafer's Textbook of Oral Pathology. 2006;1099–102.
5. Didier Lacombe. EEC syndrome. Orphanet; 2011. Available:http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=1896
6. TP63. Genetics home reference; 2011. Available:https://ghr.nlm.nih.gov/gene/TP63
7. Frias JL, Smith DW. Diminshed sweat pores in hypohidrotic ectodermal dysplasia: A new method for assessment. J Pediatr. 1968;72:606-610.
8. Agrawal A, Agrawal R, Singh R, Agrawal R, Agrawal S. Lobster claw deformity. Indian J Dent Res. 2014;25(2):243–7. DOI: 10.4103/0970-9290.135935
9. Patel A, Sharma D, Yadav J, Garg E. Split hand/foot malformation syndrome (SHFM): Rare congenital orthopaedic disorder. BMJ Case Rep; 2014. DOI: 10.1136/bcr-2014-204731
10. Thakkar S, Marfatia Y. EEC syndrome sans clefting: Variable clinical presentations in a family. Indian J Dermatol Venereol Leprol. 2007;73(1):46–8. DOI: 10.4103/0378-6323.30653
11. Knaudt B, Volz T, Krug M, Burgdorf W, Röcken M, Berneburg M. Skin symptoms in four ectodermal dysplasia syndromes including two case reports of Rapp-Hodgkin-syndrome. Eur J Dermatol. 2012;22(5):605–13. DOI: 10.1684/ejd.2012.1787
12. Joseph R, Nath SG. Association of generalized aggressive periodontitis and ectrodactyly-ectodermal dysplasia-cleft syndrome. Indian J Hum Genet. 2012;18(2):259–62. DOI: 10.4103/0971-6866.100793
13. Kaercher T. Ocular symptoms and signs in patients with ectodermal dysplasia syndromes. Graefes Arch Clin Exp Ophthalmol. 2004;242(6):495–500. DOI: 10.1007/s00417-004-0868-0
14. Kumar HN, Kugar TS, Rao RJ, Kodkany S. EEC syndrome. Indian J Ophthalmol. 2007;55(2):162–3. DOI: 10.4103/0301-4738.30725
15. Roelfsema NM, Cobben JM. The EEC syndrome: A literature study. Clin Dysmorphol. 1996;5(2):115–27. DOI: 10.1097/00019605-199604000-00003

Peer-review history:
The peer review history for this paper can be accessed here:
http://sciencedomain.org/review-history/22454