CHRIST SIEMEN SYNDROME - A REPORT OF TWO CASES

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Ectodermal Dysplasia(ED) is a rare hereditary disorder which is characterized by triad of signs comprising sparse hair, abnormal or missing teeth and inability to sweat. This article comprises of 2 such cases one of a 5 year old child with hypohydroic ectodermal dysplasia and a 15 year old female patient of clustons syndrome. This paper also highlights about history, clinical features, differential diagnosis and treatment options of such patients

Keywords: Ectodermal dysplasia, Christ –Siemens Tourine syndrome, Anodontia, hypohydrosis, hypotrichosis, clustons syndrome.

Ectodermal dysplasia (ED) represents a rare group of inherited disorders characterized by dysplasia of tissues of ectodermal origin (hair, nails, teeth, skin) and occasionally, dysplasia of mesodermally derived tissues[1]. The condition is though to occur in approximately 1 of 100,000 live births.[1,2] Freire - Maia – Pinheiro have described 154 patterns of ED divided them into 11 subgroups.[3]

Clinically ED may be divided into 2 broad categories. The X linked hypohydrotic form characterized by classical triad of hypodontia (abnormal or missing teeth), hypohidrosis (inability to sweat due to lack of sweat glands), hypotrichosis (sparse hair) and by characteristic dysmorphic facial features, which is also termed Christ –Siemens Tourine syndrome. The hydrotic form of ectodermal dysplasia that usually spares the sweat glands, can affect the teeth, hair, nails. This second category was described by Clouston in 1929 and Lowry et al in 1966 as autosomal dominant, which is found primarily is Canadian families of French descent. The most frequently form in the christen Siemen Taurine Syndrome.[4,5]

The other manifestation associated with this disease include; skin changes, recurrent ocular infection, chronic rhinitis, dystrophic nails, epitaxis, dysphagia, dyaphonia, alopecia and atypical facies.[6] Extramedullary haematopoiesis of cranial dura,[7] diminished resistance to respiratory infections[8], and nasopharyngeal rhabdomyosarcoma 9 are some of the other disorders that can be present in patient with hypohydrotic or anhidrotic ectodermal dyplasia.

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Case Reports

Case – 1

A 5 year old boy reported to the department of Oral medicine and Radiology with complaint of absence of teeth, inability to eat and difficulty in speech. Medical history revealed that the child had delayed milestones and has hearing defect and rarely communicated with other children and mother had consanguineous marriage. On examination he was febrile with prominent forehead, ears placed obliquely and protruberant lips .The patient also presented with sparse hair in the scalp and eyebrows. Hyperpigmentation was evident in mid face region. On intraoral examination revealed complete absence of primary dentition. OPG was taken which revealed complete absence of primary dentition with presence of developing tooth buds of permanent canine and under developed maxillary and mandibular alveolar ridges. Based on the clinical and radiological finding a provisional diagnosis of ectodermal dysplasia type - I or christen Siemen Taurine Syndrome was made.

Case – II

A 15 year old female patient reported to department with complaint of sharp teeth. Her medical history and family history was not contributory and she did not reveal any history of dental extractions. On examination she did not have other features except for conical teeth. Intraoral examination revealed

CASE I



Fig. 1



Fig. 2



Fig. 3

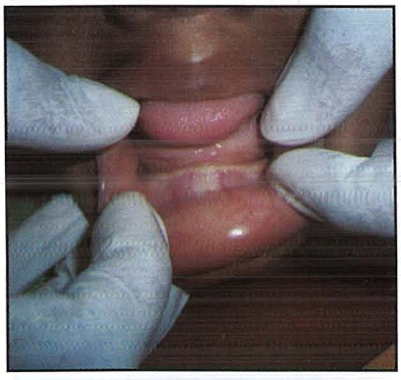


Fig. 4



Fig. 5

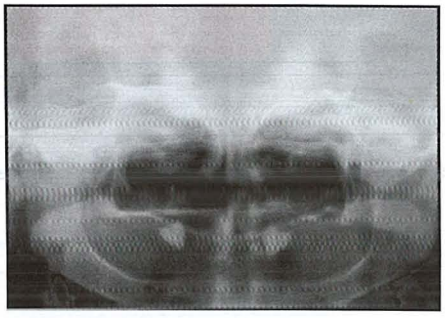


Fig. 6

CASE II



Fig. 1

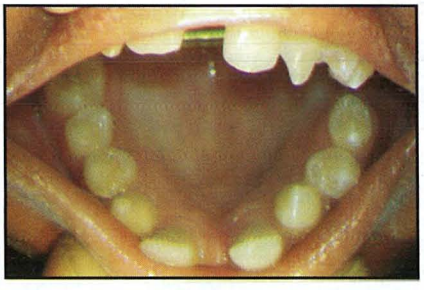


Fig. 2



Fig. 3

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missing 17, 12, 22, 26, 27, 37, 35, 44, 45 and retained deciduous 71, 81, 85. Her canine and laterals were conical in shape. Based on clinical and radiological findings we came to provisional diagnosis of ectodermal dysplasia type II (Clouston syndrome) was made.

Discussion

Charles Darwin from Wedderburn found this complex (ED) in complex in closely inbred Indian group in the year 1839 which was early by Danz in1792 according to Perobo. Anther early report was by Thurman in 1848. Christ in 1913 further defined it as a "congenital ectodermal defect" , and Weech in 1929, found the depression of sweat gland function, and coined the term anhidrotic ED Felcher in 1944 pointed out that that the skin is rarely, if ever, complete anhidrotic and suggested the adjective "hypohidrotic"[10]

ED is usually transmitted as x-liked recessive trait the gene being carried by females which is being manifested in males. In some other cases though the inheritance pattern is x linked recessive. The carrier mother exhibit minimal expression of the gene in the form of hypodontia and/or conical teeth. This is consistent with Lyon hypothesis for variability of expression.[10]

This is another form the autosomal dominant form of hydrotic ectodermal dysplasia which exists in family of French people who migrated to Canada, Scotland and northern United States.

This may be the same condition described by Papastratigakis, Tompkin and Nicolle and Halipre. Most of these patients, in contrast to those exhibiting the X linked recessive form, have (a) normal sweat and sebaceous gland function, (b) total alopecia, (c) severe dystrophy of the nails, (d) pigmentation of skin, especially over the joints, and (e) normal teeth. Other findings have included strabismus, mental deficiency, clubbing of fingers and tufting of distal phalanges, acrocyanosis, depigmentation of hair and palmer hyperkeratosis.

Another form of hydrotic ED has been described in which the teeth were conical in association were sensory nerve deafness, polydacty[1], and syndacty.[1]

The etiology of this disease is unknown; nevertheless genetic studies showed ectodermal dysplasia is due to a mutation of the gene "EDA" (ectodermal dysplasia anhydrotic). This gene is located in position q12 – q 13 of the chrosome. The EDA gene encodes a predicted founed transmembrane protein of 135 aminoacids found to be expressed in keratinocytes, hairfollicles, and sweat glands.[11]

The mutation responsible for ectodermal dysplasia has been thought to be attributed to a change in histidine /tyrosine in position 54 of the protein. Another mutation (A1270G) has also been revealed to be responsible for Tyr 343cys substitution in a patient with anhidrotic ED[12]

Differential diagnoses to be considered are Bloch Sulzbergers syndrome and the Ellisvan syndrome[13]

Treatment

The most common treatment in cases of ectodermal dysplasia with anodontia is complete dentures. Oral rehabilitation has a major psychological impact on the patients self esteem and facilitating social acceptance.[14]

Though oral rehabilitation is often difficult in these cases, patients must be attentively followed by a multi disciplinary team involving pediatric dentistry, orthodontics prosthodontics, and oral maxillofacial surgery. Serial set of dentures may be required as a child matures. Shaw et al describes the need for these to restore vertical dimension and prevent the undesirable protruding lips secondary to over closure, thereby improving the profile.[15]

Implant – supported prostheses have been associated with a high rate of success, as reported by Adell et al and Zarb and Symington. They also

provide a number of advantages, including increased retention and stability of prostheses[15].

CONCLUSION

Ectodermal dysplasia is a rare but serious disease that has to be diagnosed as soon as possible because of the important problems it can cause. Signs and symptoms occurring in the oral cavity can be very helpful in providing the clinician a provisional diagnosis. When indicated, appropriate care needs to be rendered throughout the child's growth cycle to maintain oral functions as well as to address the esthetic needs of the patient.

References

1. Dhanrajani PJ, Jiffy AO. Management of Ectodermal Dysplasia. A Literature Review. Dental Update 1998; 25:73-75.
2. Kupietzky K, Milton H. Hypohidrotic ectodermal dysplasia: Prosthetic managment of hypohidrotic ectodermal dysplasia. Quintessence Int 1995; 26:285-291.
3. Lorenzo Lo, Francesco , Claudio Scott. Prosthetic rehabilitation of a child affected from anhidrotic ectodermal dysplasia: a case report. The journal of contemporary dental practice,2005; 6 :2-6.
4. Pinheiro M, Freire-Maia N. Ectodermal dysplasias: A clinical classification and casual review. Am J Med Genet 1994;53: 153-162.
5. Richard AS, Karin V, Gerard K, Carles B, Kournjiarn J. Placement of an endosseous implant in a growing child with ectodermal dysplasia. Oral Surg, Oral Med, Oral Pathol 1993; 75:669-673.
6. Daniel E, McCurdy EA, Shashi V, et al. Ectodermal dysplasia: otolaryngologic manifestations and management. Laryngoscope 2002;112: 962-7.
7. Sitton JE, Reimund EL. Extramedullary hematopoiesis of the cranial dura and anhydrotic ectodermal dysplasia. Neuropediatrics 1992;23: 108-10.
8. Loth M, Waibel M, Albrecht G. Christ-Siemens-Touraine syndrome. Hautarzt 1998;49: 505-8.
9. Cankaya H, Kosem M, Kiris M, et al. Nasopharyngeal rhabdomyosarcoma in a patient with hypohydrotic ectodermal dysplasia syndrome. Auris Nasus Larynx 2002;29: 313-6.
10. Gorlin Rj, Pinborg JS, Cohen MM. Hypohidrotic ectodermal dysplasia. In: syndrome of head and neck. Newyork, McGraw Hill, pp 303-311
11. Hertz JM, Norgaard Hansen K, Juncker I, et al. A novel missense mutation (402C—>T) in exon 1 in the EDA gene in a family with X-linked hypohydrotic ectodermal dysplasia. Clin Gent 1998;53: 205-9.
12. Kobielak A, KobielaK K, Biedziak B, et al. A novel mutation A1270G of the EDA1 gene causing Tyr343Cys substitution in ectodysplasin-A in a family with anhydrotic ectodermal dysplasia Acta Biochimica Polonica 2003;50: 255-258.
13. Ekaterini Paschos, Karin Christine, Reinhard Hickel clinical management of hypohidrotic ectodermal dysplasia with anodondia: case report. J Clin Pediatr Dent 27(1): 5-8, 2002.
14. Shaw RM. Prosthetic management of hypohidrotic Ectodermal dysplasia with anodontia. Case report. Aust Dent J 1990, 35, 113-116.
15. Gerard Kearns, Brian Scmidt, Arun Sharma, et al Placement of endosseous implants in children and adolescents with herediatary ectodermal dysplasia. Oral Surg, Oral Med, Oral Pathol 1999; 88:5-10.

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