Volume : 3 | Issue : 4 | April 2014 • ISSN No 2277 - 8160

Research Paper | Medical Science

Ectodermal Dysplasia: A Rare Disease

Gupta Virender	Resident, Nims Medical College & Hospital, Jaipur, India.
Patel Hitalkumar	Resident, Nims Medical College & Hospital, Jaipur, India.
Kanodia S.	Associate Professor, Nims Medical College & Hospital, Jaipur, India.
Singh Kishor	Professor & Head, Nims Medical College & Hospital, Jaipur, India.

KEYWORDS :

Introduction

- Ectodermal dysplasia is an extremely rare genetic disorder characterized by faulty development of ectodermal structures.
- The clinical features include sparse hair, abnormal or missing teeth, and inability to sweat due to lack of sweat glands.

A case report of Ectodermal Dysplasia

- Ectodermal dysplasias are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures.
- It is an X linked recessive disorder.
- It is characterized by the triad of signs which comprises of
  – Sparse hair (atrichosis or hypotrichosis),
  – Abnormal or missing teeth (anodontia or hypodontia),
  – Inability to sweat due to lack of sweat glands (anhidrosis or hy pohidrosis).
- The etiology of ectodermal dysplasia appears to be genetic in nature

Case History

- A 15yr old male of consanguineous marriage presented to our institution with a history of sparse and brittle hair since childhood.
- His hair remained sparse despite various treatments.
- He also had complaint of delayed in the eruption of deciduous and permanent teeth and repeated dental caries. He also had abnormal permanent teeth.

On Examination:

- Intra oral examination revealed multiple missing teeth in the maxillary and mandibular arches.
- Patient had dental abnormalities like Hypodontia, oligodontia, carious teeth, upper fixed denture and high arched palate.
- Multiple missing teeth in the maxillary and mandibular arches.
- High arched palate
- Nail examination: no specific changes.
- Frontal bossing & slightly everted lips present.
- Sparse eyebrow and eyelashes
- sparse, fine and lusterless scalp hair
- pigeon shaped chest
- Hairs on the torso and extremities were absent
- Hypohidrosis without hyperthermia.
- Associated findings include : pigeon shaped chest without any respiratory problem.

Discussion:

- The Ectodermal dysplasias are a group of inherited disorder that share in common developmental defects involving at least two of the major structure classically hold to derive from the embryogenic ectoderms hair, teeth, nails, sweat glands [1]
- The incidence of ectodermal dysplasia in males is estimated at 1 in 100,000 births [2].
- More than 192 distinct disorders have been described till date . Freire Maia and Pinheiro published an exhaustive review and classification system for these disorders using a numeric system of 1 (hair), 2 (teeth), 3 (nail), 4 (sweat glands) for characterization [3].
- The major concern seen in these patients is the lack of teeth and the special appearance, as seen in our case [4].
- The most characteristic findings is the reduced number and abnormal shape of teeth. The delay in eruption of teeth is often the first step in the diagnosis.
- The men have an easily recognizable facies, also referred to as an old man facies.
- The extra oral features seen in this disorder are frontal bossing with the forehead appearing square in shape, prominent supra orbital ridge, depressed nasal bridge (saddle nose) however this feature was not seen in our case.
- The other features include mid face hypoplasia, pointed chin and pro-truberent and everted lips as seen in our case [5].
- Prenatal diagnosis of ectodermal dysplasia has occasionally been reported which has been diagnosed by foetal skin biopsy, obtained by fetoscopy by 20 weeks gestation.[6]
- The pointed conical teeth provide the most valuable indication and should suggest the need for sweat test and a skin biopsy.
- The treatment usually comprises of complete restoration of function and aesthetics normalise the vertical dimension and provide adequate support to the facial soft tissues.

Figure 1: ED4.tif

Figure 2: ED1.tif

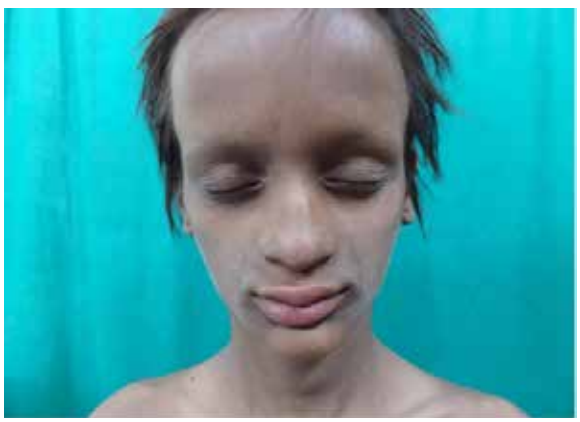

Figure 3: ED2.tif

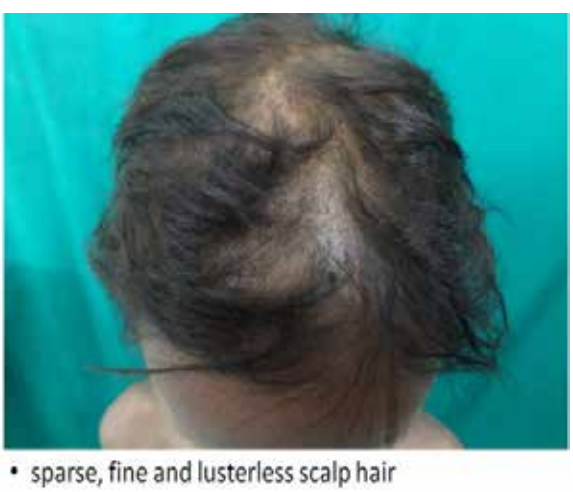

Figure 4: ED3.tif

REFERENCES

1. TM Anoop, S Simi, PN Mini, Manjula Ramachandran, PK Jabbar, PK Rajakumari, P Sujathan. Hypohydrotic Ectodermal Dysplasia.JAPI, 2008;56: 268-270. | 2. Stevenson AC, Kerr CB. On the distribution of the frequencies of mutation to genes determining harmful traits in man. Mutat Res 1967;4:339-52. | 3. Freire – Maia, Pinheiro M. Ectodermal Dysplasias. A clinical and genetic study. New York, liss. 1984. | 4. Mortier K, Wackens G. Ectodermal Dysplasia anhidrotic. Orphanet Encyclopedia. 2004:1-6. | 5. Johnson E.R., Roberts M.W., Guckes A.D., Bailey L.J., Phillips C.L., Wright J.T.: Analysis of craniofacial development in children with hypohidrotic ectodermal dysplasia. Am. J. Med.Genet. (2002) 112: 327-34. | 6. Bergendal B., Koch G., Kurol J., Wänndahl G.:Consensus conference on ectodermal dysplasia with special reference to dental treatment. The Institute for Postgraduate Dental Education, Jönköping, Sweden 1998. |