International Journal of Medicine Research
ISSN: 2455-7404; Impact Factor: RJIF 5.42
www.medicinesjournal.com
Volume 1; Issue 2; May 2016; Page No. 145-147

Ectodermal Dysplasia with Keratoconus and Other Corneal Manifestations

Pundareekaksha rao P

Assistant Professor, A. V. College & Hospital, 242-B, Trichy road, Sulur, Coimbatore, Tamil Nadu, India

Abstract

Keratoconus is a progressive degenerative condition which results from thinning of the cornea and causes vision distortion. Keratoconus is one the cause of blindness and usage of contact lens, corneal transplantation is the most commonly preferred treatment. Ectodermal dysplasia (ED) is a disorder manifest as abnormality of any two ectodermal derivatives namely skin, nails, hair, teeth, and sweat glands. ED with keratoconus is an extremely rare condition. The general aim of this article is to help health professionals make the best decisions about treatment or care for a particular condition. Herein, we present a male ED patient who developed keratoconus in right eye with corneal scar and opacity.

Keywords: Ectodermal dysplasia, Keratoconus, Corneal scar, Corneal opacity

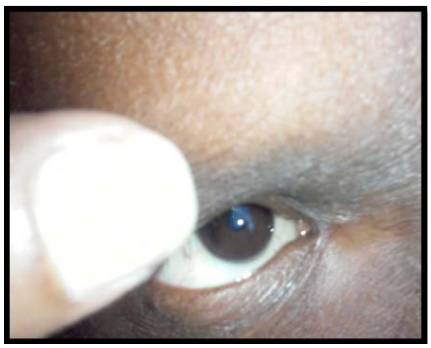
1. Introduction

A 17 year old male patient presented with hypohidrosis, hypotrichosis and, prominent forehead, saddle nose. He is the first of two children, and he had been born after a normal pregnancy. The antenatal, birth h/o and post natal h/o were normal. His younger brother is being normal. His parents also were normal and consanguineous. During 4 years of age, he had experienced bronchitis. He was admitted in one hospital and was diagnosed as hypohidrotic ectodermal dysplasia. Both eyes had watered from birth; He did not remember exactly when this corneal opacity had begun.

Dermatological examination revealed skin is dry, dark pigmented, senile facial appearance and atrophic. Hairs on the scalp were scanty, coarse, silky, very thin, brown and with patchy alopecia. He also had a bilateral conductive hearing loss. The shape of the nails of the fingers and toes seemed to be normal. Oral examination revealed anodontia. The alveolar ridges were rather atrophic. The color of oral mucosa was normal. Lip and palate are normal, no evidence of cleft. There were no findings of ectrodactyly, syndactyly of hands and feet.

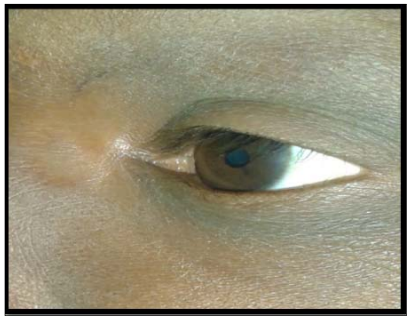
Eyebrow and eyelashes hairs were scanty. His lids were thickened, cracked and scaling. There was a moderate watery discharge from each eye. Lacrimal puncta were narrow in both eyes. Conjunctiva of both eyes was dry and muddy in appearance. Sclera of both eyes was normal. The rest of the ocular examination gave normal findings. Additionally, on Slit-lamp examination, we found both eyes upper and lower lids were having scanty, thin eye lashes on anterior border, some trichiasis of the center of the right lower lid and slight retro placement of the lower puncta behind the lid margin. Posterior borders having normal mebomian openings. Left eye cornea appeared normal, and the irises were brown. The right cornea was cone in shape with a scaring in centre of the visual axis and it extended to left. Peripheral superficial stromal vascularisation and infiltrates present. On Ophthamoscopy left eye, media is clear. Lens, blood vessels, optic disc, macula was normal. Right eye, media is not clear, a dark area present. Rest lens, blood vessels, optic disc, macula normal. Based on these findings, the patient was diagnosed as keratoconus wit ED.

Keratoconus and Hydropes in right eye

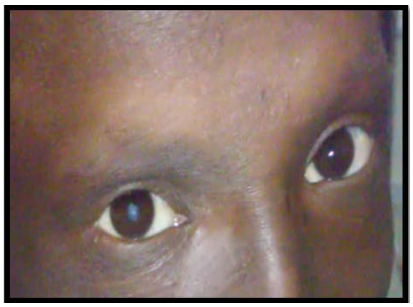


Right eye with corneal Scar, Scanty eye lashes

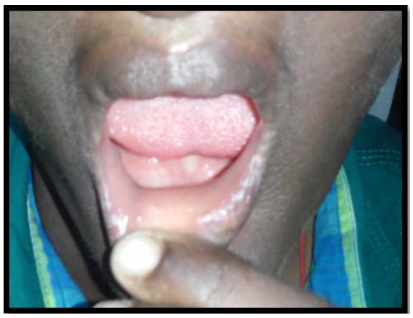
ARK appearance of right eye



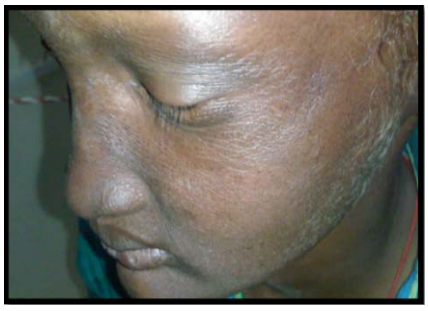
Left eye with scanty eye brow, eye lashes



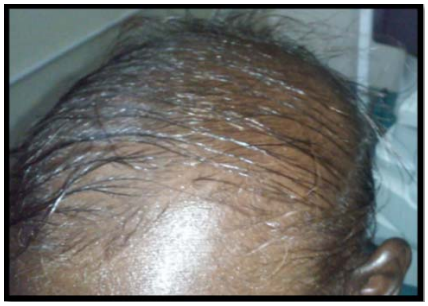
Bilateral loss of eye brow and eye lashes



Dry lips and anodontia, Normal nails



Dry and cracked skin, abnormal nose



Scanty hair on scalp

2. Discussion

ED manifest variable defects in the morphogenesis of hair follicles, nails, skin, and eccrine glands, such as sweat glands, sebaceous glands, mammary glands, and mucous producing glands in the upper aerodigestive tract and in the endothelium in the lungs [1, 2, 3]. EDs form a large and complex nosologic group with over 200 different clinical conditions [4]. It has been proposed to classify all these forms according to clinical findings [5], molecular genetic data and corresponding clinical features [6] or according to the function of the protein encoded by the mutated gene [7]. The etiology of ectodermal dysplasia lies in genetic anomalies, which can be inherited through either parents or manifested via gene mutations [8].

The patients with Hypohidrotic ectodermal dysplasia (HED) are represented as hypotrichosis, hypohidrosis and cranial abnormalities. The face is smaller because of frontal bossing, saddle nose, and absence of sweat glands resulting in smooth, dry skin and/or hyperkeratosis of hands and feet, as well as immunodeficiency. Oral characteristics may present with anodontia, hypodontia and conical teeth [9, 10]. Ocular anomalies are less frequently observed.

Alterations of the eyebrows and lashes are mentioned in combination with several ectodermal dysplasia syndromes [11, 12]. Others have reported infantile bilateral glaucoma in a child with ectodermal dysplasia. Alan A McNab *et al.* (1989) reported that two of our patients out of six developed spontaneous corneal perforations seems to be more than coincidence. There are many reasons for patients such as these to develop such a complication: the cornea is partially derived from ectoderm, there are tear film and lid abnormalities, and there may be trichiasis and an obstructed lacrimal drainage apparatus harboring potentially pathogenic organisms. The lacrimal apparatus is also derived from surface ectoderm and commonly affected [13].

Keratoconus is most commonly an isolated disorder, although clinical reports describe an association with ectodermal dysplasia. Keratoconus is a progressive and non-inflammatory thinning of the cornea, which may result in severe visual impairment due to irregular curvature and scarring. The corneal epithelium renewed every 7-14 days. Tissue at the edge of the cornea continually forms the corneal epithelium from stem cells. If these stem cells are affected, this leads to an unhealthy cornea which can result in defects in the corneal epithelium, ulcers, and scarring. It is a progressive disorder ultimately affecting both eyes, although only one eye may be affected initially [14, 15].

Despite intensive clinical and laboratory investigation, the etiology of keratoconus remains unclear. Clinical studies provide strong indications of a major role for genes in its etiology. It most likely represents a multigenic disease with a complex mode of inheritance and environmental factors contributing to the disease manifestation [16]. The central corneal thickness (CCT) is a highly heritable endophenotype of keratoconus, and it is estimated that up to 95% of its phenotypic variance is due to genetics. It is characterized by thinning and weakening of the cornea, and its symptoms range from mild astigmatism and myopia to severe vision distortion [17].

Early in the disease there may be no symptoms, in advanced disease there is a significant distortion of vision accompanied by profound visual loss. Other accompanying signs might include epithelial nebulae, anterior stromal scars, enlarged corneal nerves, and increased intensity of the corneal endothelial reflex and subepithelial fibrillary lines [18, 19]. In more advanced cases

of keratoconus which have been studied, acute hydrops will develop. In hydrops, marked stromal oedema occurs as a result of the endothelium and Descemet's membrane rupturing, allowing aqueous humour to enter the stroma. The oedema usually resolves over a period of time and eventually results in scarring. If this scarring is in the area of the visual axis then the visual acuity can decrease.

Keratoconus cones can classified by shape and position. This may be helpful when selecting the design of contact lens to fit the cornea. The suggested classes are:

• Nipple - small diameter (5mm); cone lies in the lower nasal quadrant within a few millimetres of the visual axis

• Oval - larger (>5mm); lies more commonly in the infero-temporal quadrant.

• Globus - largest diameter (>6mm); 75% of the cornea is effected.

3. Diagnosis

The differential diagnosis of keratoconus includes keratoglobus, pellucid marginal degeneration and Terrien's marginal degeneration. In these early cases, where the cornea appears normal but keratoconus is suspected, measuring the anterior topography of the central and paracentral cornea is also extremely useful to confirm the diagnosis [18]. Ultrasonic pachymetry (US) is currently accepted as the 'gold standard' when assessing the thickness of the cornea. Histopathologic features of keratoconus include corneal stromal thinning, iron deposition in the epithelial basement membrane, and breaks in Bowman's layer.

Placido Disk Studies, Keratometry, Photokeratoscopy, Computer-Assisted Video Keratoscopy, Videokeratography Studies of Keratoconus are commonly used for diagnosis of keratoconus. Videokeratography is playing an increasing role in defining the genetics of keratoconus, since early forms of the disease can be more accurately detected and potentially quantified in a reproducible manner [20].

4. Treatment

Mild keratoconus can be corrected with spectacles. When a keratoconic patient is no longer able to obtain good visual acuity as a result of increasing levels of irregular astigmatism and higher-order aberrations, rigid contact lenses will be required, effectively to provide a new anterior surface to the eye. In very advanced cases, where contact lenses fail to improve vision, corneal transplant is the best and most successful surgical option. Perforation of cornea required penetrating keratoplasty to be carried out on an urgent basis. Corneal collagen cross-linking (CXL or C3R) can stop keratoconus getting worse with stiffen the cornea. Ultraviolet (UV) cross-linking has proved effective when performed in early stages of keratoconus. In our case conservative treatment for corneal ulcers are started with antibiotics, tear supplements, patching. The case is referred to higher institutions for corneal transplantation. The follow up was limited in this case, so the evaluation of the treatment result could not be mentioned.

5. Conclusion

Keratoconus is a progressive and non-inflammatory thinning of the cornea. Keratoconus with Ectodermal dysplasia (ED) is manifest as abnormality of any two ectodermal derivatives namely skin, nails, hair, teeth with cone shape of cornea and other corneal changes. The differential diagnosis of keratoconus includes keratoglobus, pellucid marginal degeneration and Terrien's marginal degeneration. Keratoconus can be corrected with spectacles in early stages, contact lens, corneal transplantation, CXL are other measures. Counseling is needed for ED patients to move in society.

6. References

1. Irvine AD. Towards a unified classification of the ectodermal dysplasias: opportunities outweigh challenges. Am J Med Genet A. 2009; 149A:1970-1972.
2. Mikkola ML, Thesleff I. Ectodysplasin signaling in development. Cytokine Growth Factor Rev. 2003; 14:211-224.
3. Irvine A. Ectodermal dysplasias. In: Harper J, Oranje O, Prose N, eds. Pediatric dermatology. 2nd ed. Oxford: Blackwell Scientific. 2005, 1412-1466.
4. Itin PH, Fistarol SK. ectodermal dysplasias. A MJ Med. Genet 2004; 131C:45-51.
5. Pinheiro M, freire-Mara N. ectodermal dysplasias: Aclinical classification and a causal review. A MJ Med. Genet 1994; 53:153-62.
6. Priolo M, silengo M, Lerone M, Ravazzolo R. Ectodermal dysplasias: not only skin deep. Clin Gent 2000; 58:415-30.
7. Lamartine J. Towards a new classification of ectodermal dysplasis. Clin experiment dermatol 2003; 28:351-5.
8. Hickey AJ, Vergo Jr TJ. Prosthetic treatments for patients with ectodermal dysplasia. J Prosthet Dent. 2001; 86:364-368.
9. Callea M, Faletra F, Maestro A, Verzegnassi F, Rabusin M, Vinciguerra A, *et al.* Dental Phenotype in a Patient With Hypohidrotic Ectodermal Dysplasia And Severe Immunodeficiency. J Int Dent Med Res. 2011; 4:17-20.
10. Akgun MO, Sabuncuoglu F, Altun C, Guven G, Basak F. Multidisciplinary Treatment Approach of Patient With Ectodermal Dysplasia. J Int Dent Med Res. 2010; 3:141-145.
11. Kaercher T. Ocular symptoms and signs in patients with ectodermal dysplasia syndromes. Graefes Arch Clin Exp Ophthalmol 2004; 242:495-500.
12. Yavuz I, Baskan Z, Ulku R, Dulgergil TC, Dari O, Ece A, *et al.* Ectodermal dysplasia: Retrospective study of fifteen cases. Arch Med Res 2006; 37:403-409.
13. Alan A McNab *et al.* The EEC syndrome and its ocular manifestations, British Journal of Ophthalmology, 1989; 73:261-264.
14. Rabinowitz YS, Nesburn AB, McDonnell PJ. Videokeratography of the fellow eye in unilateral keratoconus. Ophthalmology. 1993; 100:181-186.
15. Lee LR, Hirst LW, Readshaw G. Clinical detection of unilateral keratoconus. Aust N Z J Ophthalmol 1995; 23:129-133.
16. Kathryn P Burdon, Andrea Vincent L. Insights into keratoconus from a genetic perspective, Clin Exp Optom 2013; 96:146-154
17. Bikbov MM, Bikbova GM, Khabibullin AF. Corneal collagen cross-linking in keratoconus management. Vestn. Oftalm, 2011; 127:21-25.
18. Krachmer JH, Feder RS, Belin MW. Keratoconus and related noninflammatory corneal thinning disorders. Surv Ophthalmol. 1984; 28:293-322.
19. Maguire LJ, Meyer RF. Ectatic corneal degenerations, in Kaufman H (ed): The Cornea. 1988, 485-510.
20. Yaron Rabinowitz s, Keratoconus. Survey of ophthalmology 1998; 42(4):297-319.