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Case Report

A rare case report of AEC syndrome, Ankyloblepharon, ectodermal dysplasia and cleft LIP/cleft palate

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ABSTRACT

The AEC syndrome or Hay wells syndrome is an unusual autosomal dominant disorder characterised by Ankyloblepharon, Ectodermal dysplasia and Cleft palate and/or Cleft lip. This syndrome occurs as a result of missense mutation in TP63 affecting P63 SAM of the gene, which is a protein-protein interaction domain. It is associated with some irregularities like Cleft palate /Cleft lip, severe scalp erosions and abnormalities of epidermal appendages including hypotrichosis, hypodontia, absent or dystrophic nails & mild hypohydrosis. We, here by report a case of full term baby born to third degree consanguineous parents with features of cleft lip and cleft palate, multiple erosions present over back, alopecia over left side of scalp along with few erosions over right side of scalp, sparse eye brows & eye lashes, Ankyloblepharon, Microphallus, Dystrophic finger and toe nails due to its rarity. Diagnosis is made commonly by clinical examination and Genetic analysis.

1. Introduction

AEC SYNDROME (Ankyloblepharon, Ectodermal dysplasia and Cleft palate/Cleft lip) or Hay wells Syndrome is a rare autosomal dominant disorder.[1,2] It is due to the missense mutation in TP63 affecting P63 SAM of the gene which is a protein-protein interaction domain.[3] The syndrome is characterised by cleft palate /cleft lip, severe scalp erosions & abnormalities of appendages including hypotrichosis, hypodontia, absent or dystrophic nails and mild hypohydrosis.[4] One rubricating distinctive feature of this syndrome is ankyloblepharon filiforme adnatum-partial thickness fusion of eyelid margins.

2. Case Report

A full term baby was born to third degree consanguineous parents with Ankyloblepharon, cleft lip & cleft palate, multiple erosions over back, alopecia over left side of scalp with few erosions, sparse eyebrows and eyelashes, Dystrophic finger & toe nails and microphallus. Based

Fig. 1: Newborn showing cleft lip

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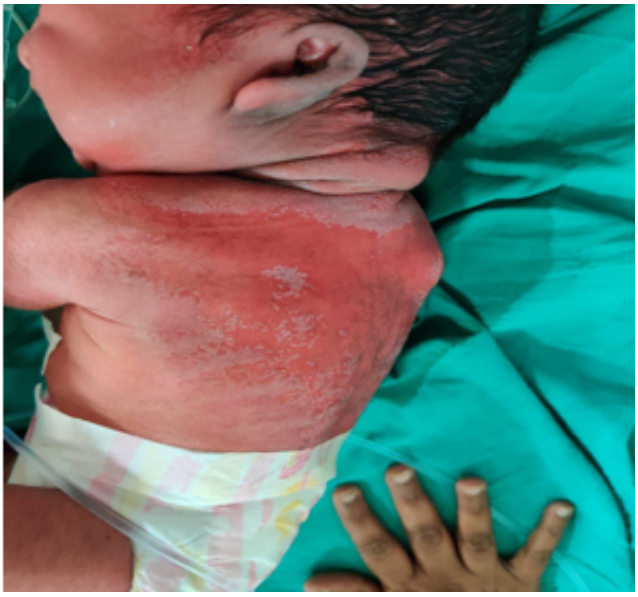

Fig. 2: Erosions over back

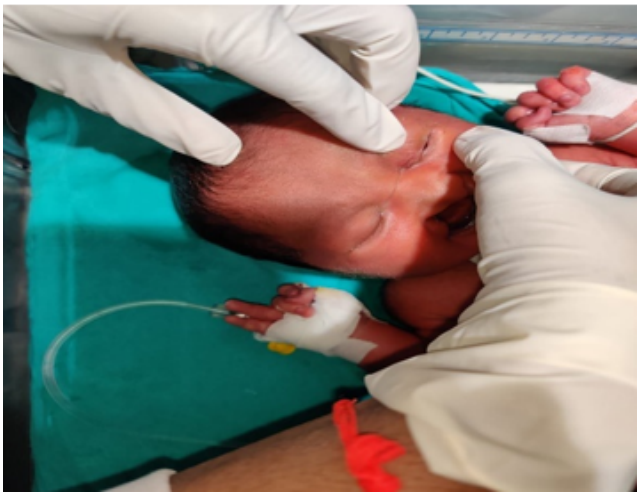

Fig. 3: Thickening of eyelids

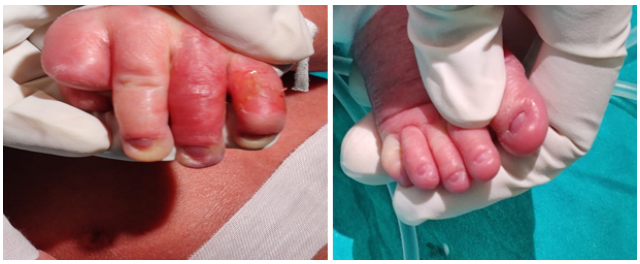

Fig. 4: Dysplastic nails

on clinical features, baby was diagnosed to have Ankyloblepharon, Ectodermal dysplasia, Cleft palate and/or cleft lip syndrome. Genetic analysis is not done owing to the lack of adequate facilities. Emollients and topical antibiotics are advised. Saline compresses are advised over erosions. Neonates must be handled with extreme care owing to their skin fragility. Abnormalities such as cleft palate and cleft lip may be surgically corrected.

3. Discussion

AEC Syndrome as discussed earlier is an uncommon and rare autosomal dominant disorder characterised by Ankyloblepharon, Ectodermal dysplasia, Cleft palate and/or cleft lip with irregularities of epidermal appendages.[1] It manifests at birth with eroded scalp dermatitis and colloidon membrane. Patients continue to suffer from scalp erosions and recurrent scalp infections.[4] Both sexes are equally affected as it is an autosomal dominant pattern. It results due to missense mutation in TP63 affecting the P63 SAM of the gene which is a protein-protein interaction domain.[3] The disorder is today considered to be identical with Rapp–Hodgkin syndrome.[5] This syndrome mainly presents with the features of Cranio facial anamolies, mid facial hypoplasia with high forehead, small mouth and narrow nose, short philtrum & short vermilion border of upperlip cleft lip or palate, hypoplasia of uvula, poor dentition and weak hair growth, dystrophic nails, hypohydrosis.[4] Other features include erythroderma and scaling skin at birth, atretic ear canal, hypoplastic maxilla, hypoplastic maxilla, bilateral punctal atresia, under development of lacrimal ducts and corneal scarring.

4. Conclusion

Neonates with AEC syndrome have extreme skin fragility and should be handled with extreme attention. Treatment includes emollients, topical antibiotics and oral antibiotics. Scalp erosions and chronic scalp infections may be severe enough to warrant surgical intervention with skin grafting.[6,7] Cleft lip/Cleft palate, hypospadiasis and maxillary hypoplasia may be surgically corrected. This case is being presented because it is an uncustomary and rare autosomal dominant syndrome.

5. Source of Funding

None.

6. Conflicts of Interest

There is no conflict of interest.

References

1. Mancini AJ, Paller AS. What syndrome is this? Ankyloblepharon-ectodermal defects-cleft lip and palate (Hay-Wells) syndrome. *Pediatr Dermatol*. 1997;14(5):403–5. doi:10.1111/j.1525-1470.1997.tb00994.x.
2. Itin P. Ectodermal dysplasias. In: Rooks Textbook of Dermatology. John wiley & sons; 2016. p. 1785.
3. Serra G, Antona V, Giuffré M, Pomi FL, Scalzo LL, Piro E, et al. Novel missense mutation of the TP63 gene in a newborn with Hay-Wells/Ankyloblepharon-Ectodermal defects-Cleft lip/palate (AEC) syndrome: clinical report and follow-up. *Ital J Pediatr*. 2021;47(1):196. doi:10.1186/s13052-021-01152-y.
4. Ramya BG, Gondane S, Kumar S, Dangi S. Ankyloblepharon-ectodermal dysplasia-clefting syndrome. *Indian J Dermatol Venereol Leprol*. 2015;81:629–30. doi:10.4103/0378-6323.168333.
5. Cambiaghi S, Tadini G, Barbareschi M, Menni S, Caputo R. Rapp-Hodgkin syndrome and AEC syndrome: are they the same entity? *Br J Dermatol*. 1994;130(1):97–101. doi:10.1111/j.1365-2133.1994.tb06891.x.
6. Rachisan AL, Cainap S, Andreica M, Miu N. Ectodermal Dysplasia Showing the Clinical Overlap between Hay Wells Syndrome and Bowen Armstrong Syndrome. *Iran J Pediatr*. 2011;21(1):121–5.
7. Cole P, Hatef DA, Kaufman Y, Magruder A, Bree A, Friedman E, et al. Facial clefting and oroauditory pathway manifestations in ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome. *Am J Med Genet*. 2009;149A(9):1910–5. doi:10.1002/ajmg.a.32836.

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