# ADULT (acro–dermato–ungual–lacrimal–tooth) Syndrome: A Case Report from India

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## Case Report

A 15-year-old girl presented with baldness of scalp over frontoparietal region. History revealed sparse hair growth over the scalp since childhood. There was no history of local trauma, drug intake, or any stress prior to the illness. She was born of a nonconsanguineous marriage and had an uneventful antenatal and postnatal period. She didn't complain of photosensitivity, heat intolerance, or frequent infections. She had an average scholastic performance. Treatment history revealed probing for frequent epiphora and medical records showed chronic right dacryocystitis with lacrimal fistula and dacryocele at the age of 4 years. No significant family history was noted and no anomaly was present in her three siblings.

Mucocutaneous examination revealed alopecia over frontoparietal scalp with normal hairline comprised of sparse lightly pigmented hairs [Figure 1a]. Bald scalp also showed increased freckling and xerosis [Figure 1a: arrowheads]. Freckling was also noted in photo distribution. A puckered scar was noted just below the right medial canthus as a sequela of a previous lacrimal fistula [Figure 1b: arrowhead] along with crusts on eye lashes, possibly due to dried up rheum which got accumulated as a result of nonpatent nasolacrimal duct. Dysplastic nails with central splitting were noted [Figure 2a]. Even though digits were of normal anatomy and number, fine exfoliation was noted at the tips of fingers [Figure 2a]. Examination of oral cavity revealed decayed permanent teeth especially incisors [Figure 2b]. No cleft lip or palate was demonstrated. Hypoplastic breasts were noted. Hair pull test was negative. Hair mount showed normal telogen hairs with a few normal anagen hairs, without any shaft defects. Based on these features, a clinical diagnosis of acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome was made.

We tried topical minoxidil (2%) for her alopecia. There was no improvement in the regrowth of hair even after 4 months of application. Hence, we advised artificial wigs for her. Our prosthodontist advised dental implant for her dental defects. Photoprotective measures were advised to prevent excessive freckling. Parents didn't give consent for genetic analysis.

ADULT syndrome, a rare ectodermal dysplasia (OMIM103285), is an autosomal dominant disorder with variable expression, caused by a heterozygous mutation in the transformation-related protein 63 (TP63) gene on chromosome 3q27. Affected individuals are characterized by limb anomalies (acro-), excessive freckling, sparse hair involving the frontal scalp (-dermato-), onychodysplasia (-ungual-), lacrimal duct stenosis or atresia (-lacrimal-), hypodontia or early loss of permanent teeth (-tooth), athelia or hypoplastic nipples, and breast hypoplasia.[1] The ADULT syndrome was first reported by Propping and Zerres[2] in a German family. The manifestations of ADULT syndrome are typically present at birth, although they may become more prominent with age.

Our patient did not have limb ray defects of ADULT syndrome. Whittington *et al.*[3] have recently reported a family of ADULT syndrome without limb ray defects, but with fingertip exfoliations as in our patient. Exfoliative dermatitis of the fingers and toes (also referred to as "neurodermitic sign") has been considered by some authors as an expression of digital anomalies of ADULT syndrome.[3,4]

The TP63-related disorders with overlapping features include ectrodactyly-ectodermal

**How to cite this article:** Razmi MT, Narang T, Handa S. ADULT (acro–dermato–ungual–lacrimal–tooth) syndrome: A case report from India. Indian Dermatol Online J 2018;9:194-6.

**Received:** July 2017. **Accepted:** October 2017.



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**Access this article online**

**Website:** www.idoj.in

**DOI:** 10.4103/idoj.IDOJ_195_17

**Quick Response Code:**

dysplasia-cleft lip/palate syndrome, limb-mammary syndrome, ankyloblepharon, ectrodactyly, cleft lip/palate syndrome, Rapp-Hodgkin syndrome, split-hand/foot malformation-4, and isolated cleft lip/cleft palate.[1] Sutton *et al*. had summarized the salient features of different TP63-related disorders [Table 1].[1] Excessive freckling and absence of cleft lip/palate distinguishes ADULT syndrome from these allelic variants.

Ectodermal dysplasia in ADULT syndrome may not always be demonstrable on skin biopsy.[5] Owing to the rarity of the condition, the specific genotype-phenotype correlation is hard to delineate. The classic clinical presentation coupled with a positive test for a TP63-related mutation helps to make a collaborative diagnosis of ADULT syndrome.[6] A multidisciplinary approach should be followed for the evaluation and the management of different manifestations of the syndrome.

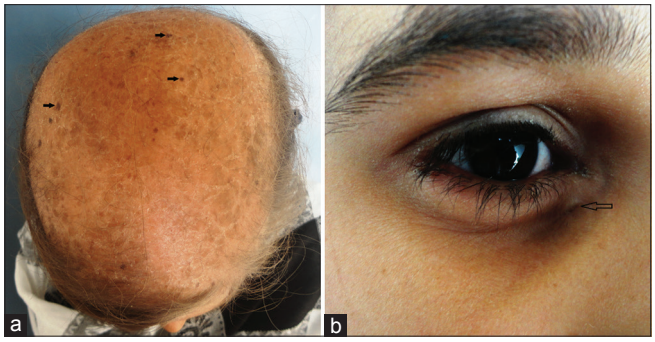

**Figure 1: Alopecia involving frontoparietal scalp with preserved hairline. Dry scaly skin noted along with excessive freckling (arrowheads), (a). Puckered scar of previous lacrimal fistula noted just below the medial canthus of right eye (arrowhead) and dry fine crusts over eyelid margin as a sequel of nasolacrimal duct obstruction, (b)**

On literature search in "MEDLINE" and "EMBASE" databases (1993–March 16, 2017), we couldn't find any previous report of ADULT syndrome from India. Only less than 20 reports of ADULT syndrome have been described in English literature. In this case, a diagnosis of ectodermal dysplasia was delayed till her teenage, even though the patient had clinical manifestations of lacrimal duct stenosis, sparse hair, and dystrophic nails since her infancy. Early diagnosis could have relieved undue stress on the parents regarding unusual manifestations in the girl. By explaining the varied presentation in the girl as a clinical expression of a genetic defect, unnecessary medical expenditure on the parents could have been avoided.

Dermatologists should be sensitized about this rare ectodermal dysplasia to avoid delay in diagnosis and to reassure the parents about a normal life span without any serious comorbidities in these patients. Educating the public on the need of genetic evaluation in these disorders cannot be over emphasized given the repeated refusal for genetic testing by the parents of the index case.

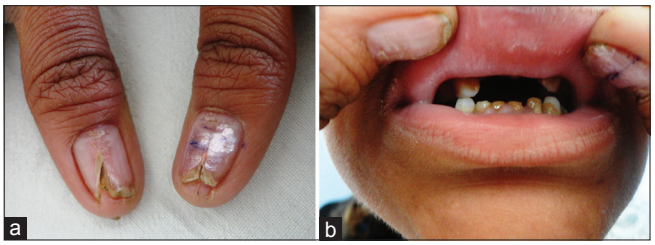

**Figure 2: Dysplastic ridged nails with central splitting; fine exfoliations at the finger tips, (a). Decayed incisors with normal lips, (b)**

**Table 1: Clinical features of transformation-related protein 63 (TP63) related disorders***

| Feature | TP63-related disorder | | | | | |
|---|---|---|---|---|---|---|
| | AEC | ADULT | EEC3 | Limb-Mammary | SHFM4 | Isolated CL/P |
| Ankyloblepharon filiforme adnatum | X | | | | | |
| Ectodermal dysplasia | X | X | X | | Rare | |
| Hypohidrosis (mostly subjective) | X | X | | X | | |
| Nail dysplasia | X | X | Mild | X | | |
| Sparse hair | X | X | X | | | |
| Tooth abnormalities | X | X | X | X | | |
| Cleft lip/palate | X | | X | X | | X |
| Split-hand/foot malformation/syndactyly | X | X | X | X | X | |
| Lacrimal duct obstruction | X | X | X | X | | |
| Dermal erosions | X | | | | | |
| Hypopigmentation | X | X | X | | | |
| Hypospadias | X | | X | | | |
| Trismus | X | | | | | |
| Excessive freckling | | X | | | | |
| Hypoplastic breasts | | X | | X | | |
| Hypoplastic nipples | | X | | X | | |

AEC = Ankyloblepharon-ectodermal defects-cleft lip/palate; ADULT = Acro-dermato-ungual-lacrimal-tooth; EEC = Ectrodactyly (split-hand/foot malformation), ectodermal dysplasia, clefting; SHFM4 = Split-hand/foot malformation type 4; CL/P = Cleft lip/cleft palate. * Source: http://www.genereviews.org/; © 1993-2017 University of Washington. (https://www.ncbi.nlm.nih.gov/books/NBK43797/; [Last Accessed on 2017 June 26]

## *Declaration of patient consent*

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that name and initial will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

## *Financial support and sponsorship*

Nil.

## *Conflicts of interest*

There are no conflicts of interest.

## References

1. Sutton VR, van Bokhoven H. TP63-Related Disorders. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, *et al*., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.
2. Propping P, Zerres K. ADULT-syndrome: an autosomal-dominant disorder with pigment anomalies, ectrodactyly, nail dysplasia, and hypodontia. Am J Med Genet. 1993;45:642-8.
3. Whittington A, Stein S, Kenner-Bell B. Acro-Dermato-Ungual-Lacrimal-Tooth Syndrome: An Uncommon Member of the Ectodermal Dysplasias. Pediatr Dermatol. 2016;33:e322-6.
4. Slavotinek AM, Tanaka J, Winder A, Vargervik K, Haggstrom A, Bamshad M. Acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome: report of a child with phenotypic overlap with ulnar-mammary syndrome and a new mutation in TP63. Am J Med Genet A. 2005;138a:146-9.
5. de Almeida HL, Jr., Caspary P, Duquia RP, Meijer R, van Steensel M. Adermatoglyphia, previously unrecognized manifestation in ADULT syndrome. Am J Med Genet A. 2010;152a:2656-7.
6. Kiritsi D, Valari M, Mileounis K, Bruckner-Tuderman L, Has C. 'Double trouble': diagnostic challenges in genetic skin disorders. Br J Dermatol. 2015;172:276-8.