



## Case Report

# A rare cause of irrevocable childhood alopecia feigning alopecia universalis: Atrichia congenita with papular lesions

Sai Sreenivasulu Nookala<sup>1,\*</sup>, UVS Akhila Reddy<sup>1</sup>

<sup>1</sup>Dept. of Dermatology Venereology and leprosy ( D V L), GGH , Kurnool Medical College, Kurnool, Andhra Pradesh, India



### ARTICLE INFO

#### Article history:

Received 04-07-2022

Accepted 29-08-2022

Available online 29-09-2022

#### Keywords:

Atrichia Congenita

Congenital Hairloss

Genodermatoses

### ABSTRACT

Atrichia congenita with papular lesions is rare, autosomal recessive condition that leads to complete hairloss which is irreversible and with keratin filled papular lesions. An insertion mutation in the exon 2 of hairless gene is responsible for the hairloss. Middle and lower portions of hair follicle are replaced by keratinizing cysts with absence of hair shafts. We here by reporting a case of 8-year-old girl with complete hairloss over scalp, eyebrows, eyelashes, and body which started at birth and complete hairloss was gradual by 9 months age on view of its rarity and misdiagnosis.

This is an Open Access (OA) journal, and articles are distributed under the terms of the [Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License](https://creativecommons.org/licenses/by-nc-sa/4.0/), which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: [reprint@ipinnovative.com](mailto:reprint@ipinnovative.com)

## 1. Introduction

Congenital alopecia has wide range of differential diagnosis and invoke various diagnostic and therapeutic challenges. Atrichia congenital with papular lesions (APL) represents a complex and heterogenous group of genodermatoses with irreversible hair loss at or immediately after birth associated with keratin filled cysts over the body.<sup>1,2</sup>

## 2. Case Report

A 8-year-old –female child presented with complete hair loss and multiple asymptomatic lesions over the body since childhood. Patient had scanty distribution of hair at birth with complete loss of hair gradually in a span of nine months. At 5 years of age, the parents noticed skin-coloured raised lesions over the body which progressively increased in number. The child attained normal milestones. There was no history of decreased sweating, blurring of vision, seizures, atopy or decreased hearing.no history of consanguinity in the parents. No history of similar

complaints in the family. On examination there was complete absence of hair over body and scalp. Multiple skin-coloured keratotic papules of size 0.5cm distributed over anterior and the posterior trunk with relative grouping in the midline, extensor and flexor aspects of both upper and lower limbs, nape of neck, skin over supraorbital ridges and forehead. Palms and soles were normal. No mucosal lesions were noted. No bony abnormalities or dysmorphic features was present. Systemic involvement was absent. Routine investigations were normal. Punch biopsy from papules histopathologically showed keratin filled cysts in the dermis and no terminal hair follicles. Parents and the child were counselled regarding the condition and its unresponsive nature to the treatment.

## 3. Discussion

APL is a rare autosomal recessive disorder with irreversible hair loss and keratin filled cysts.<sup>3</sup> Mutations in the human hairless gene located on chromosome 8 encoding putative zinc finger transcription factor have been studied.<sup>4,5</sup> Middle and lower portions of hair follicle are replaced by keratinizing cysts with absence of hair shafts.<sup>6</sup> The

\* Corresponding author.

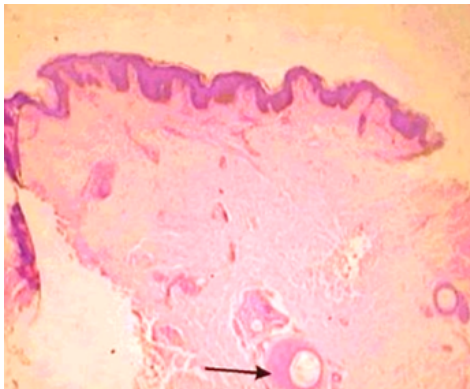
E-mail address: [seenuookala@gmail.com](mailto:seenuookala@gmail.com) (S. S. Nookala).



**Fig. 1:** Child with complete hair loss of scalp, eyebrows, and body.



**Fig. 2:** Papular lesions on the body.



**Fig. 3:** Histopathology showing keratin filled cysts and empty follicles.

main pathomechanism underlying is towards the end of

anagen phase of hair bulb.<sup>7,8</sup> Proximal inner and outer root sheath undergo premature apoptosis and disintegrate into cell clusters losing contact with dermal papilla.<sup>9,10</sup>

#### 4. Conclusion

Congenital alopecia has wide range of differential diagnosis and invoke various diagnostic and therapeutic challenges. We are reporting this case because of its rarity and is wrongly misdiagnosed as alopecia universalis and other masquerades of alopecia with unnecessary treatment burdens.

#### 5. Source of Funding

There was no funding for this work.


#### 6. Conflict of Interest

None.

#### References

1. Verma R, Vasudevan B, Pragasam V, Badad A, Mitra D, Neema S, et al. Congenital atrichia with papular lesions. *Indian J Paediatr Dermatol.* 2014;15(2):94–5.
2. Zlotogorski A, Panteleyev AA, Arita VM, Christiano AM. Clinical and molecular diagnostic criteria of congenital atrichia with papular lesions. *J Invest Dermatol.* 2001;117(6):1662–5.
3. Indelman M, Bergman R, Lestringant GG, Peer G, Sprecher E. Compound heterozygosity for mutations in the hairless gene causes atrichia with papular lesions. *Br J Dermatol.* 2003;148(3):553–7. doi:10.1046/j.1365-2133.2003.05177.x.
4. Ahmad W, Panteleyev AA, Christiano AM. The molecular basis of congenital atrichia in humans and mice: Mutations in the hairless gene (Symposium Proceedings/the Society for Investigative Dermatology). *J Invest Dermatol Symp Proc.* 1999;4(3):240–3. doi:10.1038/sj.jidsp.5640220.
5. Miller J, Djabali K, Chen T, Liu Y, Ioffreda M, Lyle S, et al. Atrichia caused by mutations in the vitamin D receptor gene is a phenocopy of generalized atrichia caused by mutations in the hairless gene. *J Invest Dermatol.* 2001;117(3):612–7. doi:10.1046/j.0022-202x.2001.01438.x.
6. Bansal M, Manchanda K, Lamba S, Pandey S. Atrichia with papular lesions. *Int J Trichology.* 2011;3(2):112–4. doi:10.4103/0974-7753.90827.
7. Thomas M, Daniel S. Atrichia congenita with papular lesions. *Indian J Dermatol Venereol Leprol.* 2011;77(1):70–2. doi:10.4103/0378-6323.74994.
8. Lin HC, Lee JY, Lin JH. Atrichia with papular lesions in a Taiwanese patient without hairless (HR) gene mutation. *Dermatol Sin.* 2010;28(1):40–3.
9. Robati RM, Posh FB, Sakoei S. Unusual presentation of atrichia with papular lesions. *Indian J Dermatol.* 2015;60(1):107. doi:10.4103/0019-5154.147889.
10. Xu L, Liu KX, Senna MM. A practical approach to the diagnosis and management of hair loss in children and adolescents. *Front Med.* 2017;4:112. doi:10.3389/fmed.2017.00112.

**Author biography**

**Sai Sreenivasulu Nookala**, Post Graduate 3rd Year  
 <https://orcid.org/0000-0002-2405-8712>

**UVS Akhila Reddy**, Senior Resident  <https://orcid.org/0000-0002-2405-8712>

**Cite this article:** Nookala SS, Reddy UVSA. A rare cause of irrevocable childhood alopecia feigning alopecia universalis: *Trichia congenita* with papular lesions. *IP Indian J Clin Exp Dermatol* 2022;8(3):202-204.