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Case Report

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

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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.

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.[1,2]

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.[3] Mutations in the human hairless gene located on chromosome 8 encoding putative zinc finger transcription factor have been studied.[4,5] Middle and lower portions of hair follicle are replaced by keratinizing cysts with absence of hair shafts.[6] The

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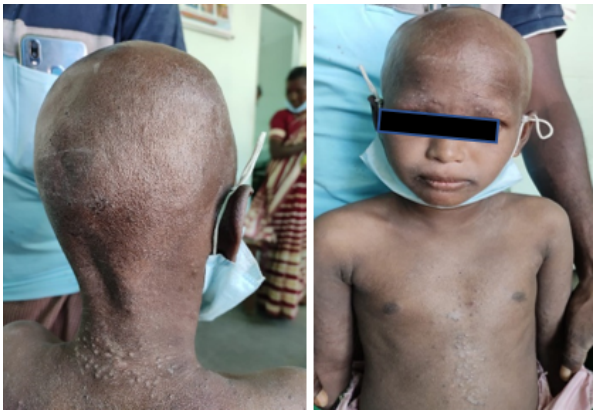

Fig. 1: Child with complete hair loss of scalp, eyebrows, and body.

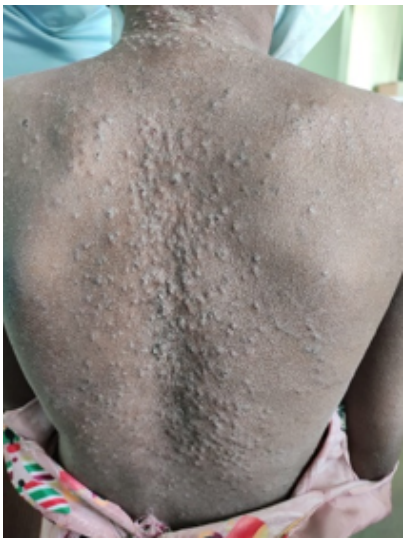

Fig. 2: Papular lesions on the body.

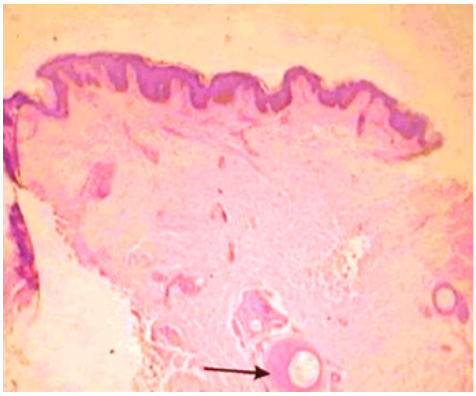

Fig. 3: Histopathology showing keratin filled cysts and empty follicles.

main pathomechanism underlying is towards the end of anagen phase of hair bulb.[7,8] Proximal inner and outer root sheath undergo premature apoptosis and disintegrate into cell clusters losing contact with dermal papilla.[9,10]

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.

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6. Conflict of Interest

None.

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