

CASE REPORT

ATRICHIA WITH PAPULAR LESIONS – A CASE REPORT

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ABSTRACT: Atrichia with papular lesion is a rare disorder of hair development characterized by almost complete loss of scalp and body hair few weeks after birth and is not replaced by terminal hair. Here we report a case of papular atrichia in a 4 year old female child presented with loss of hair and multiple follicular keratotic papules.

CASE REPORT: 4 Year old female child, first born to 3rd degree consanguineous parent, immunized for her age, presented with paucity of scalp hair since childhood. Her younger sibling is absolutely normal. She had H/O scalp hair during birth & was shed within few weeks of life and is not replaced by terminal hair (figure.1). Since then she has been almost devoid of scalp hair, eyebrows and eye lashes (figure.2). Follicular papules on the scalp started appearing in the 1st year. No growth or developmental delay. Local examination revealed few fuzzy hairs on the scalp with multiple follicular keratotic papules (figure.3). There were multiple skin colored papules over the neck, shoulder and extremities (figure.4). Teeth, nail, available mucosa and sweating were normal. Other systems were normal.

Hematological and biochemical parameters were within normal limit. Hair shaft under light microscopy is normal which ruled out monilithrix (figure.5). Scraping and KOH preparation showed no fungal element under microscope. Histopathology revealed normal epidermis, remnants of hair follicle along with perifollicular lymphocytic infiltrate and fibrosis in dermis (7). Patient was treated with topical tretinoin 0.05% cream and tacrolimus 0.03% ointment along with biotin supplementation. Both parents and child were counseled for artificial hair. Patient is followed up regularly with no improvement.

DISCUSSION: APL is an autosomal recessive form of total alopecia, characterized by hair loss soon after birth and the development of follicular papules with absence of terminal hair growth.

In 1954, Damste and prakken⁽¹⁾ followed by lowenthal and Prakken in 1961^(2, 4) used the terminology for patients with atrichia with multiple follicular keratinous cysts. Ahammed et al 1998, who referred this disease as congenital atrichia reported the identification of missense mutation in zinc finger domain of hairless gene that maps on chromosome 8p12 and is implicated in the pathogenesis of APL⁽⁵⁾. Dr. Angela Christino found similar type of mutation occurring in hairless mouse. Hence APL is considered to be human analog of hairless mouse⁽⁶⁾

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.All affected individual were considered as homozygous for A-G transition at nucleotide position 2909 (exon14) leading to amino acid change of asparagines to serine on codon 970 .

Clinical and molecular diagnostic criteria of APL are as follows. (8)

I. FAMILY HISTORY

*Family history, pattern of inheritance established as autosomal recessive, possible history of consanguinity.

*Patients are sometimes born without hair and none ever grows. More typically, patients are born with normal hair that is shed after several months and never regrows.

*Papules that start to appear during the first year of life, particularly under the midline of the eye, on the face, and on the extremities.

II. EXAMINATION

*Complete lack or almost complete lack of scalp hair.

*Sparse eyebrows and eyelashes.

*Lack of secondary axillary, pubic, or body hair.

*Papules – few to many distributed over some or all of the following areas: scalp, cheeks, arms, elbows, thighs and knees.

*Normal nails and teeth, normal sweating, and no growth or developmental problems.

*Whitish hypopigmented streaks on the scalp.

III. LABORATORY TESTS

*Lack of response to any treatment modality.

*Biopsy - absence of mature hair follicle structures, cysts filled with cornified material.

*Mutation in the hr gene.

Based on the above criteria ,we arrived at the diagnosis of APL. Papules and cysts around the elbow, pitted scars over the healed lesions, atrophoderma vermiculatum and hypo pigmented streaks over the scalp can be associated with APL (7). In our patient, keratotic papules over shoulder, elbow and knee were present..Differential diagnosis to be considered were monilithrix, alopecia universalis and vitamin D- dependant rickets type 2A. Molecular gene analysis for pathogenetic mutation will help in confirmation and differentiation of these disorders.

CONCLUSION: According to present knowledge no treatment option is available to stimulate hair growth in papular atrichia (3). We report this case because of its rarity, accurate diagnosis, and discrimination from other disorders should prevent unnecessary treatment of patients with APL.

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