

Case Report

Atrichia with Papules - A Rare Condition

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Abstract

Observations: A 6 years old girl presented with total alopecia with papular lesions involving scalp and eyebrows. She was born with sparse hair and started losing hairs shortly after birth. Hairs did not grow thereafter. Papular lesions appeared later, some 2 years after alopecia. Based on classical history, diagnosis of atrichia with papules (AP) was made. Histopathological findings supported the diagnosis.

Introduction

Atrichia with papules (AP) is characterized by failure to re-grow nearly all their hair after shedding their initial growth of hair shortly after birth and development of cystic papules later in life. It is an autosomal recessive condition and is caused by mutation of hairless (HR) gene. Our case, a 6 years old girl, presented with classical features of AP. Histopathological findings were consistent with the diagnosis. The case is being presented for rarity.

Case Report

A 6 years old girl presented with almost total loss of hair over scalp and eyebrow since birth. On enquiry it was found that she was born with sparse scalp hair but soon started "losing" hair and hairs did not grow thereafter. She was having total alopecia by 1 year of life. She started developing asymptomatic minute papules over scalp and eyebrow region after 2 years of developing alopecia and soon her scalp and eyebrows were full of such

lesions. She was a child born of consanguineous marriage and was born by normal vaginal delivery. Rest of the history including developmental milestones and immunization is unremarkable. None of her siblings or any other family member is having a similar disease. However her mother was having a congenital melanocytic nevus over face (incidental finding).

On examination scalp was having few hairs of 6-7 cm in length over occipital region. Numerous discrete minute skin colored papules were noted all over scalp (**Figure 1**). Similar changes were noticed over eyebrows too. However eyelashes were spared (**Figure 2**). She had not developed axillary or pubic hair as yet but no papular lesions were noted. Rest of the mucocutaneous examination was unremarkable.

Based on classical history and clinical findings, diagnosis of atrichia with papules was made. Histopathological examination of a papule from occipital region of scalp was performed and findings were consistent with the diagnosis. Both functional and non functional hair follicles were found. Presence of some functional hair follicle corroborates with



Figure 1. A) Clinical photograph (side profile) showing almost total loss of hair. Numerous papules on scalp can be appreciated. B) Clinical photograph (top view) showing few hairs over occipital region.



Figure 2. Clinical photograph showing loss of eyebrow with multiple skin colored papules. Note sparing of eyelashes.

few hairs seen clinically. Inflammatory cells were absent (**Figure 3**). Keratinous cyst with both epidermoid and trichilemmal differentiation was noted which is almost pathognomonic for this condition (**Figure 4**).

Discussion

Atrichia with papules (AP) is a rare autosomal recessive condition and is characterized by loss of hair beginning shortly after birth and development of cystic papules [1]. Most of the cases have been documented in children born of consanguineous marriage [2].

This condition is caused by mutation in hairless (HR) gene on chromosome 8 p12 [2]. This gene encodes a zinc finger transcription factor expressed in skin and brain [2]. In normal

hair cycle, during catagen lower segment of hair follicle undergoes well controlled apoptosis while dermal papilla is protected from apoptotic process. The perifollicular sheath forms a fibrous streamer comprised of fibroblasts, small blood vessels, and collagen and dermal papilla condenses and migrates towards epidermis to come to rest below the bulge at the lower portion of the isthmus. This upward migration of dermal papilla is crucial for continuation of hair cycle [3]. In AP, there is failure of proper involution of lower segment of hair follicle and of upward migration of dermal papilla. As a result no new anagen follicles ever form, presumably because the stem cells in bulge region cannot interact with dermal papilla which appears to be absolutely necessary. Over the time

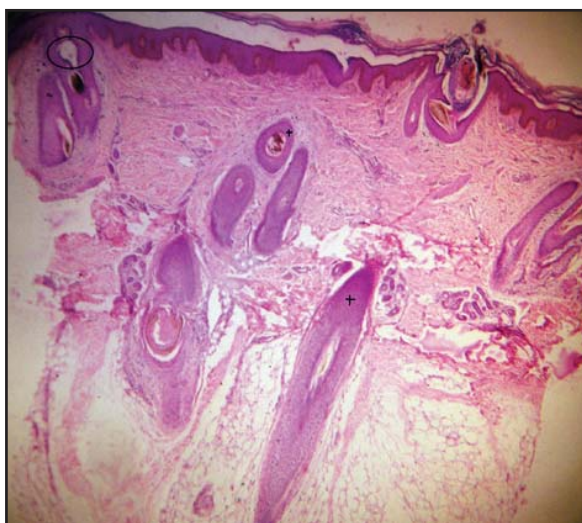


Figure 3. Histopathological photograph showing both functional (some of them have been marked with +) and non functional hair follicle. Non functional (encircled) hair follicle shows keratinous cyst (H&E stain x 40)



Figure 4. Histopathological photograph showing keratinous cyst with both epidermoid and trichilemmal differentiation (H&E stain x400)

follicular epithelium undergoes cystic changes which accounts for development of papules [3].

Similar phenotypic changes have been found in vitamin D-resistant rickets in which mutation in gene for vitamin D receptor on chromosome 12q12-q14 is noted [3, 4]. It has been found that both the hairless (HR) gene and the vitamin D receptor gene produce zinc finger proteins and may be in the same genetic pathway that controls postnatal cycling of the hair follicle [4].

The history and presentation is characteristic and are usually sufficient for making the diagnosis. The children are born with hairs, though sparse. Shortly after birth, they start losing hair, never to be replaced again [2]. Universal alopecia is noted by 1 year of life [5]. Papular lesions appear later, usually by 2 years [2]. However appearance of papular lesions may be as late as up to 18 years [5]. Scalp and body are universally involved. Eyebrows may be involved too, however eyelashes may be spared [2].

Differential diagnoses include alopecia universalis and conditions where there is widespread follicular keratosis and alopecia. Some of these include monilethrix, KID syndrome, hereditary mucoepithelial dysplasia (HMD) and IFAP syndrome. Monilethrix can be differentiated by beaded appearance of hairs. KID syndrome (**K**eratitis, **I**chthyosis, **D**eafness) may be differentiated by presence of vascularizing keratitis and a profound sensorineural hearing loss. HMD may be differentiated by presence of fiery-red palatal and gingival mucosae, photophobia, keratitis and corneal vascularization, and psoriasiform lesions in the perineal area and on the extensor limbs. IFAP syndrome (Ichthyosis follicularis, alopecia/atrichia, photophobia) may be differentiated by involvement of eyelashes, severe photophobia and corneal vascularization [6]. Alopecia universalis can be differentiated by absence of cystic papules.

Histopathological findings are quite characteristic. There is absence of developed hair

follicle. Follicular infundibula are present without lower segment of hair follicle. Intact sebaceous glands may be seen. Biopsy from the papule shows keratinous cysts with differentiation towards infundibulum and isthmus. Sometimes the cyst may rupture, leading to foreign body granuloma [2].

As such there is no treatment for this condition. But identification of this condition is important for many reasons. First, such patients should be evaluated for vitamin D deficiency and treated if required. Second, differentiation from alopecia universalis is required for prognostic purpose. Alopecia universalis may improve with therapy but AP will not improve. And lastly, various associated symptoms as described in differential diagnosis should be assessed in alopecia associated with follicular keratosis.

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