A Rare Case of Pili Annulati

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Abstract

Pili annulati is a rare hair disorder characterized by the presence of bright and dark bands on the hair shaft when viewed by reflected light. The appearance of light bands is due to clusters of air-filled cavities within the hair shaft and reduplicated lamina densa in the region of the root bulb. The condition is inherited in an autosomal-dominant fashion and requires no treatment. We report a case of an 18-year-old female diagnosed with pili annulati with no other associated dermatologic conditions.

Introduction

Pili annulati, also known as ringed hair or spangled hair, is a rare, autosomal-dominant hair disorder characterized by shiny and speckled hair with a pattern of alternating light and dark bands. Pili annulati was first described in 1866 by Landois et al., and there have been fewer than 50 reports describing it in the literature.1,2 This condition most commonly occurs in scalp hair, but can also occur in other regions of the body. It has a variable age of onset. The disease expression can vary along each individual hair shaft and between hairs in the same individual. Hairs can also present with other surface abnormalities such as trichorrhexis nodosa; however, the tensile strength and growth are not affected.2

Case Report

An 18-year-old female presented to our outpatient dermatology clinic for evaluation of moles on her trunk with no complaints of her hair. Her skin examination was normal with the exception of her hair findings. On physical exam, her hair demonstrated a blonde speckled appearance affecting the length of an entire hair strand, involving every hair strand of her scalp (Figures 1, 2). On closer inspection, each hair strand had alternating blonde and light brown bands. Her scalp exam was unremarkable with no evidence of scaling, flaking, erythema, or scarring. The patient denied having any hair products in her hair. She denied any itch or discomfort of her scalp and denied fragility of her hair. She stated that her hair developed these features in childhood. She admitted to her maternal grandmother possessing the same hair quality and texture. Microscopic evaluation of a single hair strand showed alternating light and dark bands.

Discussion

Clinical presentation and pathogenesis

The clinical presentation of pili annulati is fairly typical between patients. There are alternating light and dark bands on the hair shaft when viewed by reflected light. Clinically, bands that appear light to the unaided eye correspond to dark bands by light microscopy. They are seen as air-filled cavities within the cortex of the hair shaft by electron microscopy. It is this periodic occurrence of air-filled cavities along the hair cortex that scatter and reflect the light while preventing it from being transmitted. Those cavities have a keratin-lined inner surface and are believed to occur as a result of failure in the assembly of cytokeratins or other structural proteins, resulting in an insufficient formation of microfibrils.3 Other proposed mechanisms include a matrix formation defect and a defective regulatory protein involved in the extracellular matrix formation.2

Histology

There are no histological abnormalities noted on scalp biopsies, and the cytokeratin distribution noted with immunohistochemical studies of the scalp of affected individuals demonstrates the same pattern as the unaffected scalp. However, a wavy basement membrane zone has been reported in affected cases. Electron microscopy studies of the scalp of patients with pili annulati have demonstrated the root bulb exhibiting a reduplicated lamina densa, in contrast to the single thin electron-dense band seen in control specimens.4

Genetics

Pili annulati has been reported to have an autosomal-dominant mode of inheritance. The gene locus 12q24.32-24.33, with a critical interval of 3.9 Mb, has been mapped to pili annulati. However, no specific mutations responsible for this region have been identified in the coding or promoter sequences. It is currently believed that regulatory gene elements may be implied in the pathogenesis of pili annulati. A possible phenotypical expression of nonsense Wnt mutations (highly conserved signaling molecules) affecting the ligand or receptor regions has also been suggested.2

Associations with other diseases

Alopecia areata has been reported in association with pili annulati in some cases. However, there have been extensive studies, including family pedigree analyses, that have failed to identify a strong correlation between the two conditions,
as they seemed to have occurred independently. Linkage studies and nucleotide polymorphism tests on regions responsible for alopecia areata have not identified the pretense of the loci known to be responsible for pili annulati. In addition, the locus for pili annulati contains the gene Frizzled 10, which is involved in organogenesis and development. The regions associated with alopecia areata are associated with genes expressed in the hair follicle and involved in T cell-mediated immunity.2

Pili annulati has also been described in association with alopecia areata, blue nevi, leukonychia, melanoderma, syndactyly, and polydactylyism.5 Osorio et al. described a case of an African American female with central centrifugal cicatricial alopecia whose trichoscopy evaluation revealed the spangled light and dark pattern of the hair shafts characteristic of pili annulati.6 It has been proposed that individuals with autoimmune disease could be predisposed to have antigenic alterations in the hair root that could lower the threshold for developing conditions such as pili annulati. Thus, genes from different locations could result in a synergistic combination of autoimmunity and molecular changes to the hair bulb that result in frequent occurrence of both alopecia areata and pili annulati.2

Differential Diagnoses
When a pili annulati diagnosis is suspected, the clinician should also consider pseudopili annulati and monilethrix. Pseudopili annulati is a twisted-hair phenotype that has a normal hair shaft under light microscopy but clinically appears as a banding pattern. Monilethrix, an autosomal-dominant condition, appears as alternating nodes and constrictions under light microscopy; it also presents with fragile hair.3 Other differentials include trichorrhexis nodosa, trichorrhexis invaginata, and pili torti, which can be differentiated by close inspection and fragility of the hair.

Treatment
There is no current treatment for pili annulati. Patients rarely have any complications with the condition. Nevertheless, patients may benefit from minoxidil; it stimulates normal hair matrix production, which may help with hair structure abnormalities.7

Conclusion
Pili annulati is a hair disorder not commonly encountered in practice. Our case represents a typical clinical presentation of the rare condition. The patient did admit to her maternal grandmother having the same pattern that can be traced to the autosomal-dominant mode of inheritance. Our patient had no associated hair conditions or history of autoimmune conditions. She had no complaints about her hair and did not seek treatment. Often, patients present with rare but benign findings in dermatology practice. It is important to recognize and reassure our patients of the benign nature of their condition and be mindful of possible associated findings and differential diagnoses. In the case of our patient, after close inspection and microscopic evaluation, she left our clinic educated about pili annulati.

References