

Pili Annulati: A Report of 2 American Families

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Practice Points

- Pili annulati is characterized by a banded or speckled appearance due to alternating light and dark bands, mostly on scalp hair.
- Inheritance of pili annulati may be autosomal dominant or sporadic.
- Pili annulati should be distinguished from conditions such as pseudopili annulati, bubble hair, pili torti, and monilethrix.

Pili annulati (PA) is a rare, benign, autosomal-dominant or sporadic hair shaft disorder characterized by scalp hair with a banded or speckled appearance. We present 2 American families with PA and highlight the characteristic clinical and microscopic features. Among the 2 families, a total of 11 individuals with PA were identified, spanning 3 generations in family 1 and 4 generations in family 2.

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Pili annulati (PA), or ringed hair, is a rare benign hair shaft disorder that was first described by Anton Karsch in his 1846 dissertation and was first cited in the English language literature by Landois¹ in 1866. Pili annulati is characterized by a banded or speckled appearance due to alternating light and dark bands, mostly on scalp hair. Clinically,

the light bands correlate with dark air-containing spaces on light microscopy (LM), which can be verified as air-filled cavities within the cortex on electron microscopy (EM).^{2,3} Inheritance of PA may be autosomal dominant or sporadic. The usual age of onset is 2 years or older with a lifelong duration and normal hair growth. The banding pattern is more apparent with fair-colored hair or age-related graying, but individuals who present at a younger age tend to have more severe phenotypes.³ Banding may be subtle or only detected on LM. A range of 20% to 80% of scalp hairs may be affected. Involvement of beard, axillary, or pubic hair is not common.⁴ Banding occurs at irregular intervals along the hair shaft⁵ and becomes less frequent distally along the hair shaft.³ We present 2 American families with PA and highlight the characteristic clinical and microscopic features. The PA in the proband of family 1 was incidentally discovered when she presented with telogen effluvium.

Case Reports

Family 1—A 10-year-old white American girl presented with mild diffuse hair loss from the roots of 2 months' duration. Hair growth was normal without increased fragility. She had a history of Graves disease that was treated with radioablation 9 months earlier, and she was taking levothyroxine sodium for the last 3 months for hypothyroidism. The family on her father's side were of German descent. Family history was notable for Graves disease in her paternal grandmother, hypothyroidism in her maternal grandmother, alopecia areata in her maternal cousin,

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and androgenetic alopecia on her paternal side. On physical examination, her scalp hair was blonde-brown with a speckled appearance (Figure 1A). Hair from all sections of her scalp was clinically affected.

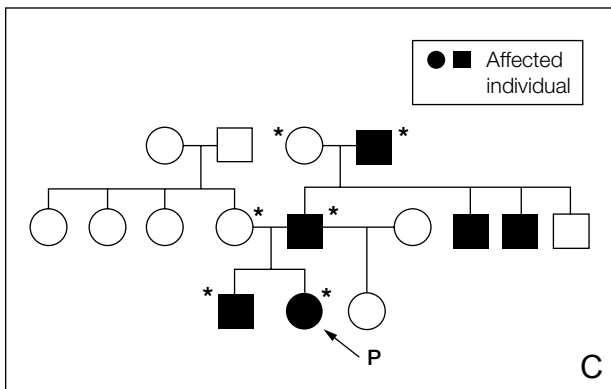


Figure 1. Blonde-brown scalp hair with a speckled appearance on physical examination of a 10-year-old girl (the proband)(A). Light microscopy revealed a banded appearance on her scalp hair with dark air-containing spaces in the cortex (B). The proband (arrow) had a family history of pili annulati; asterisk indicates the individual was clinically examined (C).

No clinical banding could be found on other body hair, but she was prepubertal. Light microscopy examination revealed a banded appearance on her scalp hair with dark air-containing spaces in the cortex (Figure 1B). Further evaluation of her medical history revealed that the banded appearance was initially noticed at 2 years of age. Her brother, father, 2 paternal uncles, and paternal grandfather were similarly affected (Figure 1C); however, the affected hair was only present on the scalp without concerns of hair fragility or hair loss, except for androgenetic alopecia. Medical evaluation also revealed that she was diagnosed with telogen effluvium related to her thyroid disease. Her hair loss resolved within 1 month after her evaluation without changes in her PA. Clinical examination and LM of the hair of her brother and grandfather confirmed familial PA. The proband and her brother appeared to demonstrate strong phenotypes, as the entire scalp and all scalp hair tested were affected. The phenotypic severity of her grandfather's disease was difficult to assess due to his concomitant androgenetic alopecia and short hair. Hair brought in from her father was tested using LM

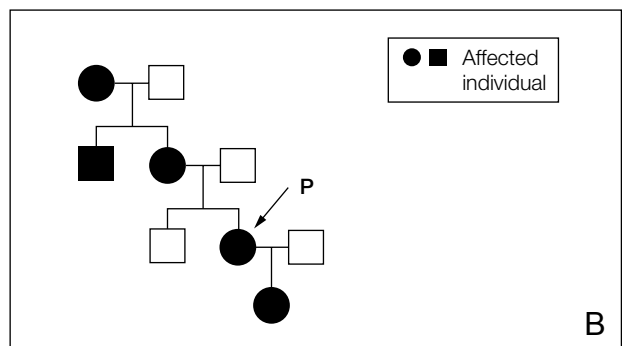


Figure 2. Scalp hair with alternating light and dark bands along individual hair shafts that created a shiny appearance in the light in a 29-year-old woman (the proband)(A). The proband (arrow) had a family history of pili annulati (B).

and no clinical examination was performed. Family history also included narcolepsy in her father and 2 paternal uncles.

Family 2—A 29-year-old white American woman contacted one of the authors (K.A.G.) with a concern of beaded hair that she had observed since 13 years of age. The patient described her hair as strange and observed alternating light and dark bands along individual hair shafts that created a shiny appearance in the light (Figure 2A). Her hair did not grow longer than shoulder length and was always straight. Her aunt worked in a forensics laboratory with access to the Internet and medical literature, and she offered a diagnosis of PA. The patient contacted us with questions about possible associated symptoms. Her family was of French and German descent. Her mother, uncle, maternal grandmother, and daughter also were diagnosed with PA (Figure 2B), which was confirmed by LM. Her family reported no other medical conditions and no concerns of hair loss or pigment changes in their hair.

Comment

In PA, light bands to the naked eye correspond to dark air-containing bands on LM, which can be verified by EM.^{2,5,6} The air-filled cavities reflect scattered light, thus appearing darker on LM.² Hair shaft caliber is normal. Suspected PA bands must be closely distinguished from an interrupted pigmented medulla.⁷ Electron microscopy also may reveal longitudinal gaps between cortical macrofibers.^{3,8} Transmission EM demonstrates a reduplicated lamina densa in the hair root bulb, and the basement membrane zone appears unusually wavy with immunohistochemical stains using antibodies to its structural components.⁹ These results suggest that the primary defect in PA may involve proteins that are important in the assembly of the basement membrane zone. The severity of PA can be quantified by features of LM including the percentage of scalp hairs affected, the percentage of affected hairs with banding throughout the full hair shaft length, and the percentage of affected hairs with full-width bands (>80% of the width of the hair).¹⁰ Amino acid analysis reveals elevated lysine and decreased cysteine in PA hair shafts.^{3,5}

Some cases of alopecia areata have been reported in patients with PA.¹⁰⁻¹² Case reports of PA with woolly hair, blue nevi,⁵ leukonychia, melanoderma, syndactyly, and polydactyly also have been described. We hypothesize that neither alopecia areata nor telogen effluvium of the proband (family 1) were associated with PA. Instead, we believe the development of alopecia areata or telogen effluvium often leads patients to visit the dermatologist and enables the incidental diagnosis of PA.

Pili annulati seems to be associated with mild variable hair fragility, even though patients affected with PA do not report increased hair fragility and reports in textbooks classically have stated that PA is not associated with increased hair fragility.^{3,13} None of the individuals in our report had noticed increased hair fragility. In particular, the nodes or places of air-filled cavities in PA may be weak, which predisposes the hair to hair shaft breakage with trauma.^{3,5,13} There was no difference in tensile strength in a study of 1 family, but breakage always occurred at the nodes.⁵ Features of weathering have been observed on EM, including regular indentations or longitudinal curtainlike folds along the hair shaft with intact cuticular cells.^{3,13} Rarely, cases have demonstrated severe abnormalities at the nodes with damaged cuticle and cortex and trichorrhexis nodosa–like breaks.¹³ Atomic force microscopy has demonstrated that dark bands are stiffer to the naked eye than light bands, whereas light bands are comparable in stiffness to control hair.¹⁴

The differential diagnosis of PA includes pseudopili annulati (PPA),^{15,16} bubble hair, pili torti, and monilethrix. The affected hair in PPA demonstrates a banded appearance similar to PA. Pseudopili annulati likely is a variant of normal with no reports of familial cases or comorbid conditions. Banding in PPA is caused by optical effects related to twisted hair with unusual geometry. On LM, PPA does not demonstrate the dark air-containing bands that are characteristic of PA.^{15,16} The fibers have elliptical cross-sections and partial twisting. Pseudopili annulati otherwise is normal on EM. In PPA, the banded appearance only is seen if light strikes at a 90° angle to the hair's long axis (transverse illumination). In contrast, the banding in PA is apparent with light from all angles but is best seen with longitudinal illumination.^{15,16} Bubble hair is caused by the use of hot blow-dryers and curlers and may be associated with acquired alopecia with a localized or diffuse distribution.^{17,18} In bubble hair, large irregular vacuoles form within the hair shafts, resulting in brittle broken hairs. Light microscopy of bubble hair does not reveal the dark air-containing bands characteristic of PA. On EM there is a loss of cortical cells and medulla, resulting in a Swiss cheese–like appearance.^{17,18}

No treatment is necessary for PA because it is benign and rarely bothersome. Intralesional triamcinolone did not improve PA in 1 report.³ Occurrence of PA may be sporadic or with autosomal-dominant inheritance, similar to our 2 families, as well as with variable expression of the PA phenotype.^{3,7,19} The pathogenesis is unknown. Alterations in the basement membrane zone have been described.⁹ No abnormalities in the expression of a number of cytokeratins were detected in 1 study.²⁰ Genetic mapping

has narrowed the locus to a 2.9-Mb region at the telomeric end of chromosome 12 and excluded mutations in the coding regions of all 36 possible candidate genes by sequence analysis.¹⁹ Determining the genetic defect that causes PA should provide insight into hair shaft biology and development.

In summary, PA is an uncommon benign hair shaft disorder. Pili annulati may be familial but rarely is bothersome, and most patients clinically do not report increased hair fragility. Pili annulati should be distinguished from conditions such as PPA, bubble hair, pili torti, and monilethrix.

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