



Coexisting trichorrhexis nodosa and pili annulati: a case report of hair shaft abnormalities in a Syrian family

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Introduction: Pili annulati (PA) and trichorrhexis nodosa (TN) are rare genetic hair disorders. PA presents with sparkly, spotted hair due to air-filled cavities in the cortex, mainly affecting the scalp. TN causes brittle, breakable hair shafts, often due to physical or chemical damage.

Case Presentation: A 23-year-old Syrian woman presented with glistening, brittle hair and whitish nodules since puberty, reminiscent of TN. Trichoscopy confirmed TN alongside PA, displaying characteristic dark-light bands. Interestingly, TN breakage occurred within the PA bands. Her aunt displayed similar TN patterns. Microscopy revealed ring-like structures and a 'thrust paint brush' appearance, indicative of TN, within PA-affected regions. Management included avoidance of hair treatments and prescription of vitamins and minoxidil to enhance hair strength and density.

Discussion: PA is usually congenital and autosomal dominant, often without causing hair fragility. However, some cases report fragility, particularly in light bands. While PA is typically benign, its combination with TN underscores the complexity of hair shaft disorders. Only five such cases exist. Proposed causes for PA and TN include genetic factors and hair shaft abnormalities. PA's fragility may stem from band stiffness differences and lower cysteine content. Treatment is typically unnecessary, but avoiding excessive heat styling is advised.

Conclusion: This case highlights the rare co-occurrence of PA and TN, emphasizing the need for further research into their relationship and potential systemic associations. Understanding the underlying mechanisms is crucial for informing treatment and counseling patients effectively, especially considering the rarity of this association.

Keywords: case report, dermoscopy, hair abnormalities, pili annulati, trichorrhexis nodosa, trichoscopy

Introduction

Pili annulati (PA) is a rare genetic disorder with an autosomal dominant pattern that affects the hair shaft^[1]. Visually, individuals with PA present with sparkly, spotted hair characterized by alternating black and bright bands along the hair shafts^[1]. This appearance arises from a defect in the hair structure, resulting in the alternating formation of air-filled cavities within the cortex^[1]. PA predominantly impacts the scalp region; occurrences in other

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HIGHLIGHTS

- Pili annulati (PA) is a rare genetic disorder affecting hair shafts, characterized by sparkly, spotted hair with alternating black and bright bands.
- Despite its distinctive visual characteristics, PA is typically benign, not affecting hair strength, and individuals usually do not experience fragility symptoms.
- Trichorrhexis nodosa (TN) is a hair disorder characterized by brittle, dry hair shafts prone to breakage and often results from physical, heat, or chemical damage to the hair.
- This case study involves a 23-year-old woman with both PA and TN, along with a family history of these disorders.
- Investigating the co-occurrence of PA and TN provides insights into their clinical management, genetic basis, and potential shared pathophysiological mechanisms.

areas, such as beard hair, pubic hair, and armpit hair, are less common. Hence, it is worth noting its potential occurrence in other areas, albeit infrequently^[2].

The onset of PA ranges from infancy to adulthood, reflecting the varied age of presentation and highlighting the need for vigilance in diagnosis across different age groups^[3]. Despite its distinctive visual characteristics, PA is often benign, not affecting hair strength, and individuals typically do not experience fragility symptoms^[3].

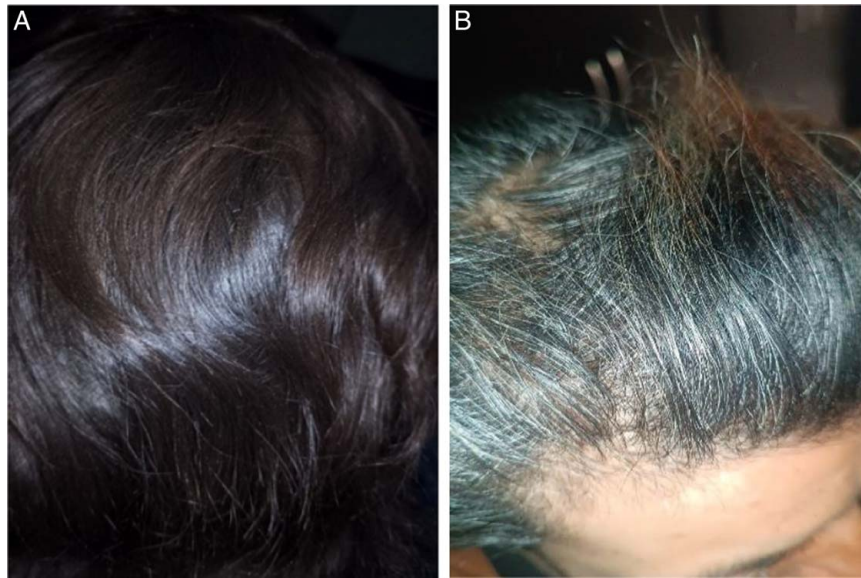


Figure 1. a Clinical finding of our patient: brittle hair and numerous white dots consistent with TN. b Clinical finding of patient's aunt: brittle hair with several tiny whitish nodules (TN).

Trichorrhexis nodosa (TN) is another hair disorder, characterized by hair shaft abnormalities that result in brittleness, dryness, and susceptibility to breakage^[4]. This condition often arises from physical damage to the hair, such as vigorous combing or rubbing, exposure to heat during styling, and chemical damage caused by hair-straightening agents^[4]. It is more prevalent in acquired cases than congenital ones and is notably observed in individuals of African descent due to the unique characteristics of their hair^[5]. Despite primarily affecting scalp hair, recent reports have highlighted TN extending to facial hair, broadening the understanding of its potential manifestations^[5].

This paper reports a unique case study involving a 23-year-old woman admitted to Aleppo University Hospital with both PA and TN, alongside a family history. Investigating the co-occurrence of these two rare hair disorders offers valuable insights into their clinical management, genetic underpinnings, and potential shared pathophysiological mechanisms.

Case presentation

A 23-year-old Syrian woman with no medical history presented to the hospital with complaints of an unusually glistening texture in her hair since puberty. She reported that her hair never grew longer and had brittle strands, along with several tiny whitish nodules and broken ends in the temporal, frontal, and occipital areas (Fig. 1a). There was no consanguinity between her parents. Her aunt also had short and brittle hair since puberty (Fig. 1b).

During the clinical examination, her scalp hair exhibited a banded appearance with alternating dark and light colors, increased fragility, and numerous white dots consistent with TN in the temporal and frontal areas and mild TN in the occipital area. No other abnormalities were observed on the scalp, and her axillary and pubic hairs appeared normal.

Trichoscopy revealed multiple white bands indicative of PA and broken hair shafts in the affected regions with TN (Fig. 2a).

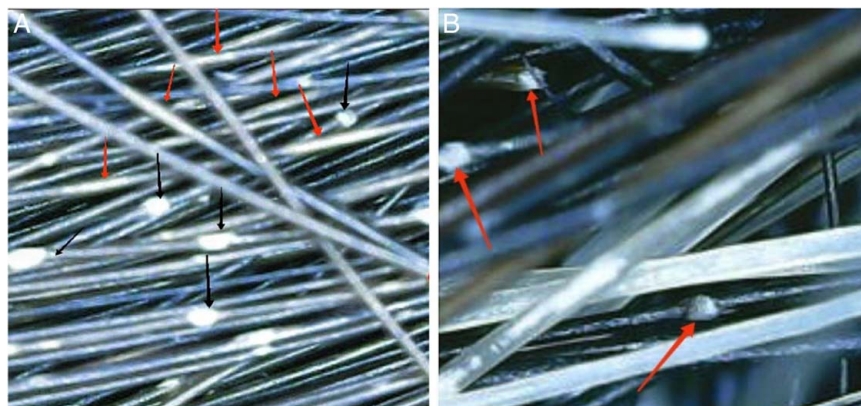


Figure 2. a Trichoscopy shows multiple white bands indicative of Pili Annulati (PA) 'red arrows' and regions of TN breakage within the hair shafts 'black arrows'. b Patient's aunt: Typical TN affecting hairs from the temporal area with 'thrust paint brush' appearance.

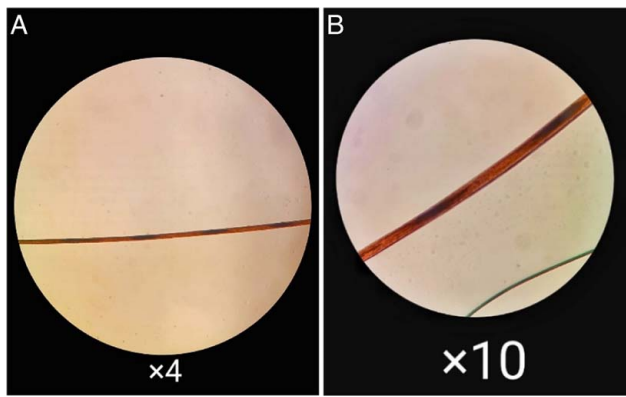


Figure 3. a Light microscopy display hair shaft with periodic dark and light bands under optical microscopy (4 × magnification). b Light microscopy display hair shaft with periodic dark and light bands under optical microscopy (10 × magnification).

Notably, the regions of TN breakage were found within the PA white bands. Additionally, a trichoscopy examination of her aunt's hair showed severe TN in the temporal and frontal areas (Fig. 2b) and mild TN in the occipital area, similar to our patient.

Under light microscopy, hair shafts displayed periodic dark and light bands resembling rings (Fig. 3a) under optical microscopy (4 × magnification) and (Fig. 3b) under optical microscopy (10 × magnification). Also, it showed a 'thrust paint brush' appearance of the hair shaft, which pointed to TN (Fig. 4a) under optical microscopy (10 × magnification). There were regions affected by PA and TN in the same microscopic field (Fig. 4b).

As part of the management plan, we recommended the patient avoid heat and chemical treatments on her hair. We also prescribed vitamins, biotin, and minoxidil to support the strength and density of her hair.

Discussion

PA is an uncommon benign disorder, nearly 50 cases of PA have been documented in the literature^[6]. It is characterized by alternating dark and light hair bands, most commonly found in scalp hair but may also occur in pubic, axillary, and beard hair. These

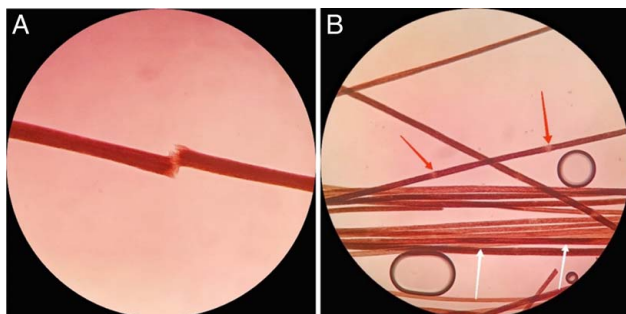


Figure 4. a Light microscopy shows a 'thrust paint brush' appearance of the hair shaft. b The red arrows indicate white dots consistent with TN. The white arrows indicate the periodic dark and light bands consistent with PA.

bands are caused by air-filled cavities in the hair shafts that alter the scattering and transmission of light, resulting in decreased light transmission and increased light reflection compared to normal hairs^[6].

PA is classified as a hair shaft disorder that typically does not cause hair fragility. However, some cases in the literature report an increase in fragility associated with pili annulate, making the hair more susceptible to weathering, particularly in light bands^[3,2]. While some cases are sporadic, PA is usually a congenital disease with an autosomal dominant pattern^[6].

This report describes a patient with PA and TN, which is a rare association. TN is another hair disorder characterized by the formation of small nodules and broken ends in the hair strands, it is often attributed to chemical and cosmetic use on hair or physical trauma but can also be inherited or acquired^[2,4].

To our knowledge, there are only five cases that describe PA associated with TN in the literature^[3]. The exact pathogenesis of PA and TN remains unclear, but underlying genetic factors and structural abnormalities in the hair shaft have been proposed as potential mechanisms^[3].

An explanation for the hair breakage in the PA suggests that the difference in stiffness between the light and dark bands, as observed through atomic force microscopy, may play a role. However, a case study involving a patient with both PA and acquired TN found that TN affected hairs unaffected by PA and areas outside the dark bands, indicating that cuticular damage, rather than band stiffness, may be a key factor in hair breakage. The increased fragility observed in PA hair could also be linked to a lower cysteine content, as analyses have shown that PA hair has less cysteine and more lysine compared to normal hair^[3].

Although PA typically does not require treatment, physicians should advise patients to avoid excessive use of heat styling tools, as they can further damage fragile hair strands^[6].

In conclusion, this case report sheds light on the association between PA and TN, highlighting the need for further research to understand their relationship and potential associations with other systemic conditions.

Ethical approval

This retrospective review of patient data did not require ethical approval in accordance with local/national guidelines.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Author contribution

S.I.: supervised and helped in writing the manuscript; N.A.R.: diagnosed and treated the patient; B.R.J., F.B., J.Z., M.A., and

B.M.: wrote the manuscript; B.R.J.: critically revised the manuscript. All authors read and approved the final manuscript.

Conflicts of interest disclosure

The authors declare that there is no conflict of interests in this study.

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Not applicable.

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Data availability statement

All data generated or analysed during this study are included in this published article and its supplementary information files are available upon reasonable request.

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