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REVIEW AND NEW CASE REPORTS ON SCANNING ELECTRON MICROSCOPY OF PILI ANNULATI, MONILETHRIX AND TRICHOTHIODYSTROPHY

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Abstract

Pili annulati, Monilethrix and Trichothiodystrophy are uncommon conditions in which the hair shaft has a distinct appearance as seen by optical microscopy and scanning electron microscopy (SEM). We report several new cases and review the characteristic ultrastructural abnormalities investigated by SEM.

Pili annulati: abnormal areas which show a longitudinal, "curtain-like" folding of the cuticular cells, alternating with normal areas. The latter present regularly non-systematized, superficial depressions. Hair specimens of the patient's mother show the same surface irregularities.

Monilethrix: most hair shafts show variations in thickness giving a typical nodal appearance. In the isthmus area we noticed longitudinal ridging and cuticular scales that are extended lengthwise in a fish-scale-like pattern. In the nodus area a smooth surface due to a complete loss of cuticular cells, was observed.

Trichothiodystrophy: the hair morphology observed by SEM is characterized by severe cuticular and secondary cortical degeneration along the entire length of the flattened hair shaft, with longitudinal ridging, cuticle loss, trichorrhexis nodosa formation and trichoschisis.

SEM observations show morphological abnormalities which are characteristic for each pathological condition described. This method may provide data that add some clarity in the surface changes of the different hair shaft anomalies.

Key Words: Dermatology, Pili annulati, Monilethrix, Trichothiodystrophy, light microscopy, scanning electron microscopy.

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Introduction

Scanning electron microscopy (SEM) has facilitated the three-dimensional study of the fine architecture of hair shaft surface of both normal and several pathological conditions of the hair shaft. The morphological study by the usual optical microscopical observations of three hair shaft anomalies: pili annulati, monilethrix, and trichothiodystrophy was complemented by a SEM study. The purpose of this study is to review all morphological pathognomic features of these inherited hair anomalies, most of which have been previously reported.

Materials and Methods

Hairs were mounted without any prior treatment such as dehydration with alcohol or critical point drying on aluminium supporting disks using double-sided tape. They were coated with a 10 to 20 nm thick layer of gold and observed in a JEOL JSM-820 SEM at 10 and 12 kV and at 10^{-6} A beam current.

Hairs from normal subjects and from patients with **pili annulati** (2 patients: a girl and her mother), **monilethrix** (2 patients), and **trichothiodystrophy** (3 members of the same family) were studied by this method.

Results

The cross-section of the normal hair shaft showed little variation in diameter along its length. In the SEM, we noted that one layer of cuticular scales was imbricated with its free edge directed towards the distal end of the hair.

Pili annulati (ringed hair)

A 12-year old girl was presented in January 1990 for a diffuse alopecia which started one year ago. Her hair had been permanent-waved at that time. The parents are cousins.

Clinical examination revealed a scintillating ringed appearance of the patient's hair. Also her mother's hair showed, at ocular inspection, the characteristically bright and dark bands but there was no clinical

problem with hair growth.

Light microscopic (LM) examination showed abnormal, opaque areas alternating with normal areas all along the hair shaft in every hair, which confirmed the diagnosis of pili annulati or ringed hair (Fig. 1).

SEM examination of the patient's hair shaft showed no gross reduction of hair diameter and the presence of abnormal areas alternating with normal areas. The abnormal areas were characterized by longitudinal flutes and ridges or "curtain-like" folding and punctiform depressions (Fig. 2). The so-called normal areas regularly presented longitudinal, superficial depressions. In both areas we noticed a pronounced fragility of the cuticular cells along the hair shaft, translated by transversal cuticular fissures and occasionally absence of scales (Fig. 3). At places we observed cortical fibers dissociated by the air filled cavities.

Hair specimens from the patient's mother were then processed for SEM. Abnormal areas showed the same distinctive surface irregularity and are alternating with normal areas.

Monilethrix

Hair was obtained from two patients, a 11-year-old girl and a 40-year-old woman with monilethrix since infancy. Clinically occipital baldness was prominent. The hair was sparse and short with a beaded appearance and emerged from follicular hyperkeratotic papules.

When examined by LM and SEM, most hair shafts showed more or less regular variation in shaft thickness, giving a nodal appearance (Fig. 4). This abnormality was different in degree among individual hairs, depending on the localization. Hair of the occipital area showed a periodical reduction of the hair diameter (Fig. 4), while a hair specimen from the anterior part of the scalp presented no or only slight constriction.

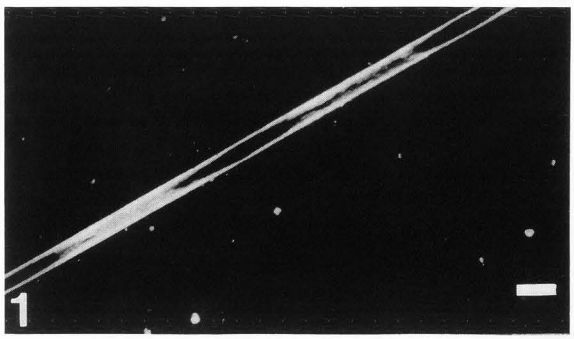
In the isthmus area of the highly developed beaded hair, we noticed longitudinal ridging and cuticular scales that were extended lengthwise in a fish-scale-like pattern. Cuticular scales became gradually thinner and in the nodus area a smooth surface, due to a complete loss of cuticular cells, was observed (Fig. 5).

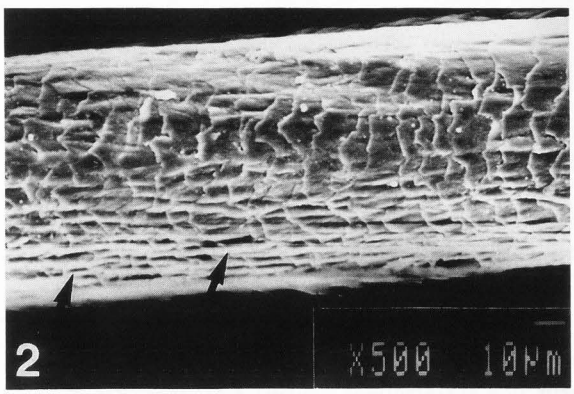
The monilethrix hair with no or only slight constriction also showed an abnormal cuticle pattern. The cuticular scales had irregular jagged borders in a saw-tooth-like pattern and showed indentations in a S-form (Fig. 6).

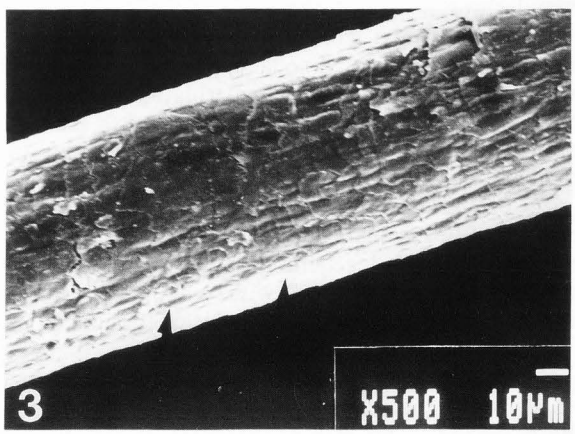
Trichothiodystrophy

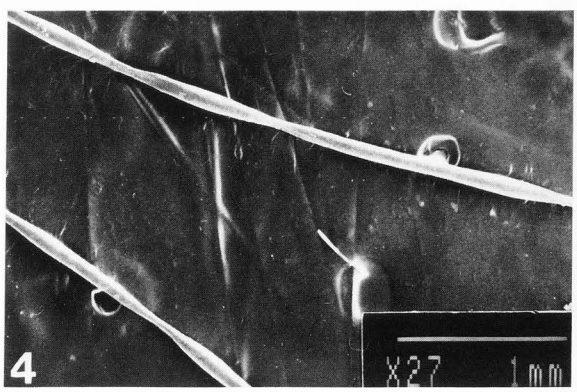
We had the opportunity to observe a family where 3 children had trichothiodystrophy. The parents are cousins but showed no clinical signs of this anomaly. Clinically, the hair from the three patients was sparse, fragile and failed to attain a normal length.

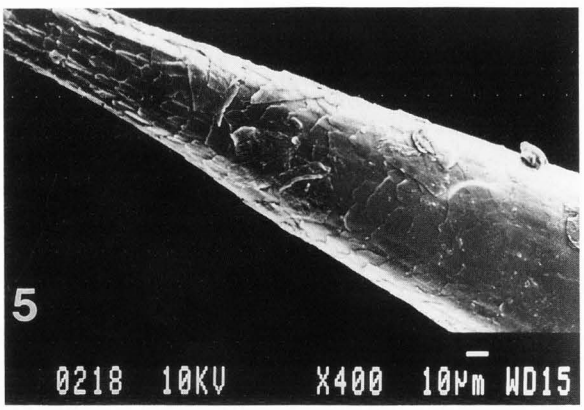
On LM examination, the hair showed a characteristic ribbon-like flattening. Two types of fractures were observed: trichoschisis and a type resembling trichorrhexis nodosa. Under plane polarizing microscopy we noted alternating bright and dark regions that gave a handed appearance to the hair shaft when the hair axis

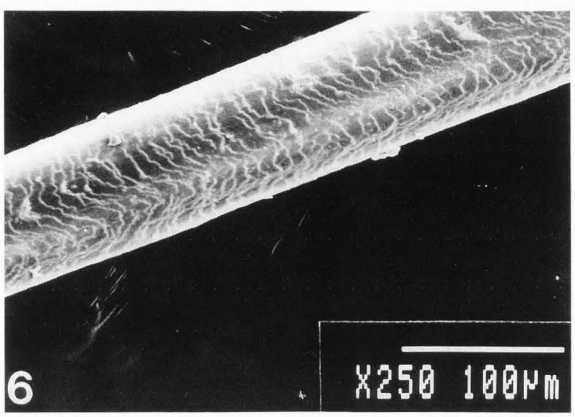












Figures 1-3. Pili annulati. Figure 1. Opaque areas alternating with normal areas are seen when examined on polarized light. (Bar = 100 μm). **Figure 2.** "Curtain-like" folding in the abnormal area (arrow). **Figure 3.** Longitudinal, superficial depressions and transversal cuticular fissures along the hair shaft (arrow).

Figures 4-6. Monilethrix. Figure 4. Nodal appearance of monilethrix hair. **Figure 5.** Longitudinal ridging at the isthmus area and loss of cuticular scales at the nodus area. **Figure 6.** Hair with no or slight constriction, showing indentations of the cuticular scale in a S-form.

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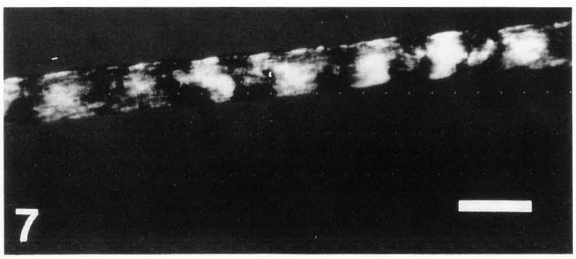
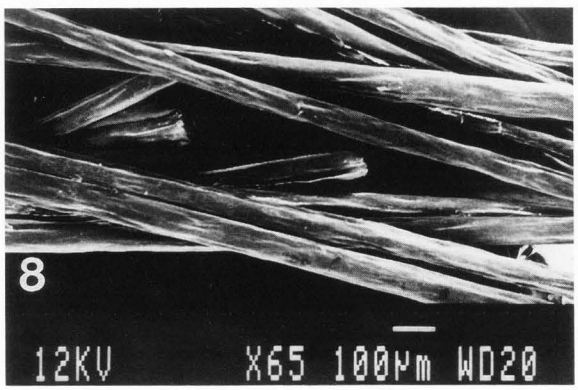
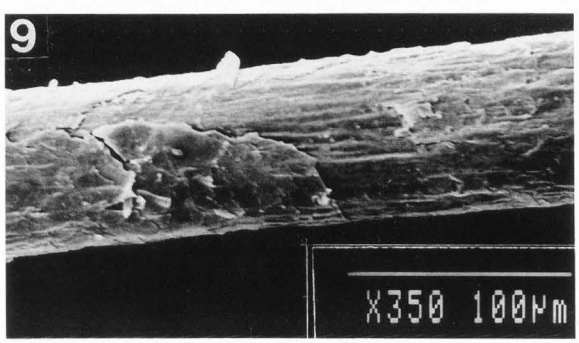
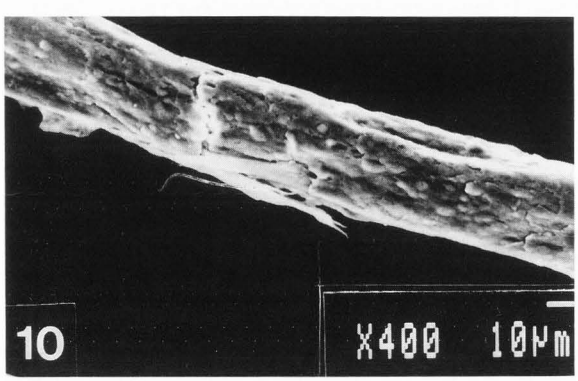
was approximately aligned with one of the polarizer directions (Fig. 7).

SEM examination showed the hair shafts to be flattened in cross-section and longitudinally ridged (Fig. 8). The fragility of the hair was demonstrated by an abraded or lost cuticular layer and transverse splits in the cuticle cells (Fig. 9). Partial breaks of the trichorrhexis nodosa type, as well as complete brush breaks, were seen (Fig. 10).









Figures 7-10. Trichothiodystrophy (TTD). Figure 7. Alternating bright and dark regions that give a banded appearance to the TTD hair when examined on polarized light. Bar = 100 μm. **Figure 8.** Flattened and longitudinally ridged hair shafts. **Figure 9.** Abraded cuticular layer and transverse splits. Bar = 100 μm. **Figure 10.** Partial breaks are seen. Bar = 10 μm.

Discussion

Pili annulati

Ringed hair is a rare inherited anomaly or may occur sporadically (Price *et al.*, 1968). Most of the cases may show abundant scalp hair but some cases may show a fragility of hair and develop alopecia. From our results we conclude that the mother is probably one of the carriers without clinical symptoms.

The presence of abnormal air-filled cavities in pili annulati has been clearly demonstrated. Transmission electron microscope (TEM) and SEM studies have confirmed the presence of large hair spaces within the cortex of abnormal bands of the hair shaft (Dini *et al.*, 1988; Ito *et al.*, 1988).

TEM observations have also shown similar cortical cavities attributed to a lack of cortical cell proteins to occupy the space available (Price *et al.*, 1968). Gummer and Dawber (1981) demonstrated the presence of electron opaque, cystine positive material in the intermacrofibrillar spaces. They also found important undulations of cellular membranes of the cuticle in the abnormal areas touching the exocuticle and the endocuticle. This could be responsible for the folding aspect of the hair surface in pili annulati. The relation to the cortical defect however is not clear. Punctiform depressions in the abnormal areas could represent invaginations of already undulated cuticular cell membranes, while in the normal areas superficial, longitudinal depressions may correspond to "moderated but distinctive" undulations (Gummer and Dawber, 1981).

Further TEM studies have shown the holes through the cortex to be located both intracellularly and intercellularly (Dini *et al.*, 1988). Recent investigations suggest that pili annulati may be a disorder of protein metabolism involving a partial dysfunction of cytoplasmic ribosomes, resulting in a lack of cortical keratin formation (Ito *et al.*, 1988).

Monilethrix

Monilethrix is usually inherited as an autosomal dominant trait. The characteristic beading of the hair shaft, resulting from the alternation of the wider nodes and the narrower internodes, is responsible for the extreme brittleness of the monilethrix hair, especially in the occipital area.

The regular beading may be due to a periodic malfunction of the germinative cells of the cortex, producing a periodic decrease in number of cortical cells and reduction in shaft diameter with complementary restriction of the cuticle diameter (Gummer *et al.*, 1981). This results in folding of the cuticle membrane in the isthmus area of the moniliform hairs.

In the nodus area, we have the impression that the cuticle is being virtually stretched, so that the hair surface appears to be smooth. This corresponds to the findings made by Lubach *et al.* (1982). These authors examined the members of one family with monilethrix. The hair with no or only slight constriction of the clinically affected children had cuticular scales with jagged borders and irregular margins. Such surface structures could also be observed in the case of the clinically healthy sister and father.

A computer stereographic study showed that severe periodic reduction of diameters occurs in the cortex, moderate reduction in the inner root sheet, and the reduction seldom occurs in the outer root sheet (Ito *et al.*, 1990). Severe thinning of the cortex is now known to be caused by a degenerative change of the matrix cells of the cortex which may be genetically determined. The abnormal reduction in diameter of the cortex is already detectable histologically in the heterogenous zone and becomes visible after complete keratinization of the cortex as an internode. The factors that may cause the cell degeneration are still unknown (Ito *et al.*, 1990).

Trichothiodystrophy

In patients with trichothiodystrophy, the hair is typical]y very short, brittle and flattened. Trichothiodystrophy, a rare inherited neuroectodermal complex, is characterized by specific biochemical abnormalities of the hair shaft. A general reduction in the cystine and sulphur content of both the cuticle and the cortex has been observed (Price *et al.*, 1980; Gummer *et al.*, 1984). The reduction occurs in the ultra-high sulphur proteins of the cortex (Gillespie and Marshall, 1983; Venning *et al.*, 1986). It is reasonable to postulate that the lack of disulphide cross-links in the non helical matrix proteins is responsible for the "floppiness" and fragility of the hair shaft in Trichothiodystrophy (Venning *et al.*, 1986).

A lack of high-sulphur proteins in the cuticle, particularly in the A-layer and the exocuticle is in part responsible for the degenerative changes seen in trichothiodystrophy.

Affected hairs, observed by SEM, show an advanced degree of weathering in abrasion and loss of cuticle cells, brush breaks, pitting and splitting of the exposed cortex, and fractures of the trichorrhexis nodosa and trichoschisis types, both in the proximal and distal part of the hair shaft (Van Neste and Bore, 1983; Venning *et al.*, 1986).

Gummer and Dawber (1985) demonstrated gross distortion of all the component cell layers of the hair follicle below and through the zone of keratinization. The effects on the formation of the hair shaft could be due to an alteration of a regulatory gene which suppresses the synthesis of some high-sulphur proteins and enhances the synthesis of a new group of proteins, which may be intermediate protein units in the production of the final keratin molecule. This would explain why keratin production is started but is not completed (Gillespie and Marshall, 1983).

In conclusion, SEM investigations show structural abnormalities which are characteristic for each of the three pathological conditions we described. Moreover, our study underlines the fact that the fine analysis made

of the hair surface using SEM makes it possible to recognize clinically silent carriers of the gene, responsible for the hair shaft anomaly.

Acknowledgments

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Discussion with Reviewers

E. B. Rest: Was the father's hair of your case of Pili annulati examined? Although Pili annulati has been reported to occur with an autosomal dominant inheritance it does not seem appropriate to comment on the inheritance pattern without discussing both parents.
Authors: We did not have the opportunity to examine the father's hair, therefore we cannot comment on the inheritance pattern.

E. B. Rest: The presence of longitudinal depressions along the hair shaft is seen in a number of hair shaft anomalies and in normal people. Are the authors proposing this as a specific defect of pili annulati? Is this a final common pathway? A normal variant?
Authors: Alternating normal and abnormal zones which show longitudinal depressions along the hair shaft is known as a specific defect for pili annulati.

E. B. Rest: Fragility of cuticular scales is mentioned along with transverse cuticular fissures and absence of scales in pili annulati. Pili annulati is not generally associated with increased fragility of hair. Do these changes express themselves clinically as increased hair fragility?
Authors: In our case, the patient was presented with diffuse alopecia. We believe that increased hair fragility in pili annulati is related to the ultrastructural anomalies reported.

E. B. Rest: There are many overlapping abnormalities seen in the structural hair shaft anomalies of trichothiodystrophy. Which of these findings do the authors feel were pathognomonic or was it a combination of the findings?
Authors: The authors agree that the combination of several findings in trichothiodystrophy is pathognomonic.