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Light and scanning electron microscopic examination of late changes in hair with hereditary trichodysplasia (Marie Unna hypotrichosis)

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ABSTRACT

Objective: Our aim was to investigate the microscopic surface structural alteration in hair with hereditary trichodysplasia. This article presents the results of light and scanning electron microscopy (SEM) examination of cases having hereditary trichodysplasia.

Methods: The biopsy specimens were obtained from 2 girls of ages 3 and 5-years, Department of Pediatrics, Faculty of Medicine, Hacettepe University in 2001. A large number of hair specimens were obtained from these 2 cases having hereditary trichodysplasia. Routine light microscopic and SEM procedure was performed on the tissue specimen, and then they were examined by light microscopy and SEM.

Results: Hair specimens taken from both patients had great similarities. Our results reveal that the atypical looking hair were flattened, twisted and partly scattered

at the end. Moreover, these hairs had sheath structures with abnormal proliferation and these structures were damaged, the cuticles had fractures and were degenerative.

Conclusion: There is only a small number of SEM studies in literature reporting the ultrastructural changes of hereditary trichodysplasia. Scanning electron microscopy is a 3 dimensional examination technique revealing easily comparable images and it is indispensable for diagnosis in various tissues which permit considerable magnification. As it is used in the hereditary trichodysplasia syndrome its routine usage in many dermatologic and hair diseases will result in valuable contributions to scientific literature.

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Hereditary trichodysplasia (Marie Unna hypotrichosis) is a rare autosomal dominant disorder manifested by almost complete congenital absence of scalp hair, eyebrows, and eyelashes; decreased body hair; and widespread facial milia.^{1,2} In early childhood, hair growth may occur but the hair soon becomes coarse, flattened, or twisted. During puberty the hair becomes very sparse,

particularly on the vertex and scalp margins, resulting in a high frontal and nuchal hairline.² Scattered follicular horny plugs may be associated with this disorder, and histologic examination of cutaneous biopsy specimens of involved areas reveals an abnormal proliferation of the internal root sheath in many of the follicles. When examined under the dissecting microscope, abnormal hairs are

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seen as flat, twisted, and ribbon-like. Electron microscopic examination of hair may reveal peeling of the cuticle, increased interfibrillar cortical matrix, and intracellular fractures of the cuticular cells, cortical cell fibrils, and medullary cells.¹⁻³ There is only a single SEM studies in literature reporting the ultrastructural changes of hereditary trichodysplasia.

Methods. The biopsy specimens were obtained from 2 girls of ages 3 and 5-years. Both of the girls had almost no hair at birth. The sparse hair which grew with very slowly till the age of one was light colored but there was no graying. They did not take any medication. They washed their hair with regular hair shampoo and normal tap water. We couldn't ask for all factors that influence the hair. The patients taking part in this study had some, or all of the classical manifestations of hereditary trichodysplasia: loss of almost all the hair, eyebrows and eyelashes, decreased body hair and wide spread facial milia. We examined 16 hair specimen from the patients. Routine light microscopic procedure was performed to the tissue specimen. Also, the tissue samples were fixed in 2.5% glutaraldehyde for 24-hours, washed in phosphate buffer (pH: 7.4), post-fixed in 1% osmium tetroxide for one hour, washed in phosphate buffer (pH: 7.4), dehydrated in increasing concentrations of acetone, critical point dried and mounted on metal stubs with a double sided adhesive band.⁴ Then, the samples were sputtered with a 100 Angstrom thick layer of gold in a BIO-RAD sputter apparatus. Their photographs were taken with a Jeol SEM-ASID 10 scanning electron microscope. We evaluated the details of surface structure: hair surface, cuticulae pattern, hair's filamentous-keratinized structures, hair degeneration.

Results. In light microscopic examination, we found abnormal proliferation of the cuticular cells of the shaft of hair with Hereditary Trichodysplasia (**Figure 1**) observed obviously when compared with normal hair (**Figure 2**). With SEM, we evaluated hair according to 6 criteria: hair surface structure, cuticulae pattern, hair's filamentous-keratinized structures, color, hair degeneration and factors that influence hair. We found all hair's surface structure deformed. Sheath structures were showing abnormal proliferation (**Figure 3**). There was fractured hair sheaths (**Figure 4**). Distal ends of the hair showed deformed cone shaped structure (**Figure 5**). Proximal ends of hair shaft was showing scattered pattern (**Figure 6**). We detected areas where hair nodes were partially absent (**Figure 7**). Generally, cuticulae irregularities were together with degeneration in the scalp hair. We detected degeneration of the cuticular cells and irregular cuticular pattern in all specimens. Regularity of the cuticulae was altered in these

areas. The hair's filamentous, keratinized structure was also altered. We noted flattened hair structures in some segments (**Figure 8**).

Discussion. It is known that hereditary trichodysplasia causes alterations on hair. In literature, histologic examination of cutaneous biopsy specimens of involved areas reveals an abnormal proliferation of the internal root sheath in many of the follicles. When examined under the dissecting microscope, abnormal hair are seen as flat, twisted, and ribbon-like. Electron microscopic examination of hair reveals peeling of the cuticle, increased interfibrillar cortical matrix, and intracellular fractures of the cuticular cells, cortical cell fibrils, and medullary cells.¹⁻³ Since the protein structure of the hair has been found normal in Hereditary Trichodysplasia, the studies about this disease are focused on the hair surface. Surface of hair^{5,6} are studied for different purposes: Pavlov studied cuticulae patterns of scalp hair of 5 subjects from different countries and found that cuticulae could show variation according to racial factors.⁷ Selvaag et al¹⁰ studied hair of patients that showed different structural abnormalities like twisted hair, longitudinal grooves, trichorrhexis nodosa as well as variations in the hair caliber. Nanko et al⁹ found that hair discoloration was mainly due to cuticulae damage in swimmers. We detected alterations on hair with Hereditary trichodysplasia in light and scanning electron microscopy. There is only a single study with SEM in literature reporting the ultrastructural changes in hair with hereditary trichodysplasia. Wirth et al¹⁰ in 1985 found that hereditary trichodysplasia is not an anomaly in the protein structure of the hair but describes changes only in the hair morphology.

In this article, these morphologic changes are described but our study gives a much detailed description of these and some additional morphologic changes of the hair of these patients. Wirth et al¹⁰ described peeling of the cuticle and longitudinal ridding. We also observed the peeling in the cuticle but longitudinal ridding was absent. In addition, we observed flattened segments, proliferation, fractured cuticle and scattered endings. Our results are similar for both of the our patients. The atypical looking hair of the patients was flattened and had twisted and had partly scattered appearance. Moreover; these hair had sheath structures with abnormal proliferation and these structures were damaged, the cuticles had fractures and were degenerative. Scanning electron microscopy is a 3 dimensional examination technique revealing easily comparable images and it is indispensable for examination in various tissues which permit considerable magnification. As it is used in the Chediak-Higashi syndrome¹¹ its routine

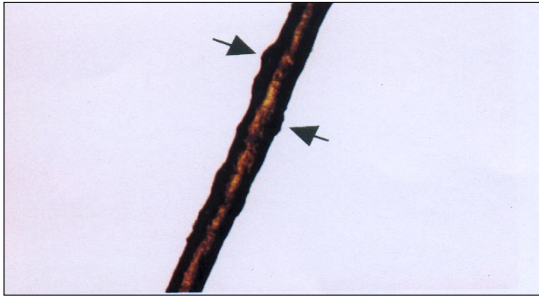


Figure 1 - Light microscopic photograph showing a case with hereditary trichodysplasia (HT) (abnormal proliferation).

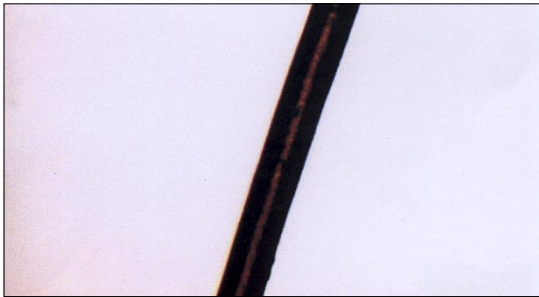


Figure 2 - Light microscopic photograph showing normal hair structure.

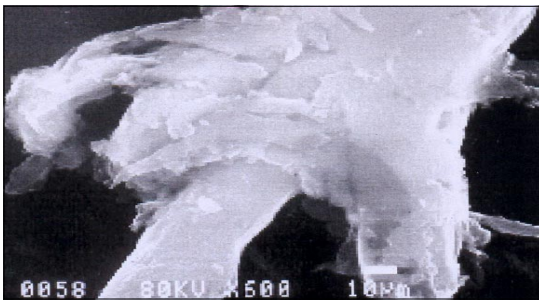


Figure 3 - Fractured hair sheaths (HT).

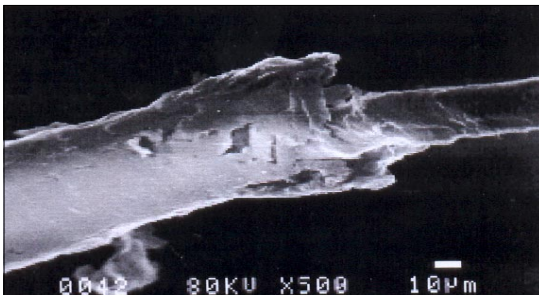


Figure 4 - Fractured hair sheaths (HT).



Figure 5 - Distal end of the hair (HT) (scattered distal ending).

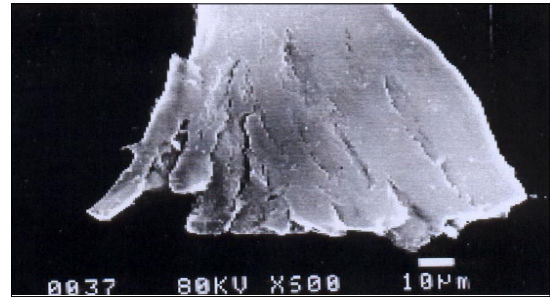


Figure 6 - Proximal end of the hair (HT) (an abnormal ending).



Figure 7 - Areas where cuticle structures having partial nodes were absent (HT).

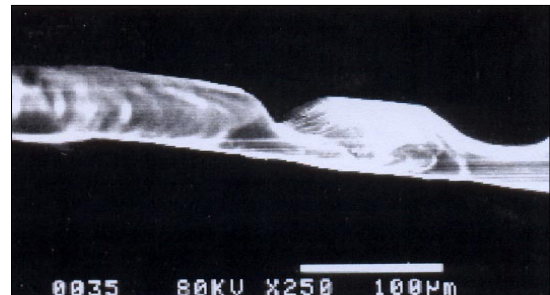


Figure 8 - Flattened hair structures in some segments (HT).

usage in many dermatologic and hair diseases with surface alterations will result in valuable contribution to scientific literature.

Our study demonstrates the structural changes of the hair with hereditary trichodysplasia in detail. Light microscope can be used for routine diagnoses of hereditary trichodysplasia whereas SEM can be used to differentiate hair with this disease from the other diseases effecting hair surface morphology. So, our results can be presented as an ultrastructure demonstration of this disease. In many dermatologic and hair diseases our findings will be useful in clinical application.

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