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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.
SHORT COMMUNICATION

Structural hair shaft abnormalities in hypomelanosis of Ito and other ectodermal dysplasias

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Hair samples from patients with different ectodermal dysplasias; hypohidrotic ectodermal dysplasia, pachyonychia congenita, tricho-dento-osseous syndrome, tricho-rhino-phalangeal syndrome, and hypomelanosis of Ito were investigated using a scanning electron microscope. The hairs of the patients showed different structural abnormalities; twisted hairs, longitudinal grooves, trichorrhexis nodosa as well as variations in the hair caliber. Hair shaft abnormalities, as in our patients with tricho-dento-osseous syndrome, and hypomelanosis of Ito have so far not been described.

Key words: Ectodermal dysplasia, hair shaft abnormalities, hypomelanosis of Ito, pachyonychia congenita, tricho-dento-osseous syndrome, tricho-rhino-phalangeal syndrome, scanning electron microscopy, X-linked hypohidrotic ectodermal dysplasia

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Ectodermal dysplasias comprise a large group of inherited disorders involving tissues and organs of ectodermal origin, and are classified clinically into two major groups; group A consists of conditions with defects in at least two of the structures hair, teeth, nails and sweat glands with and without malformations/defects in other tissue; group B includes conditions with defects in only one of the above-mentioned structures plus at least one other ectodermal defect. In all, more than 150 different conditions have been classified according to the scheme developed by Pinheiro and Freire-Maia (1).

Different hair shaft abnormalities in ectodermal dysplasia have been reported, and the aim of this study

Fig. 1. A. Scanning electron microscopical investigations of hair from a girl with hypomelanosis of Ito showing longitudinal grooves. B. Scanning electron microscopical investigations of hair from a girl with hypomelanosis of Ito showing twisted hair.

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Hair shaft abnormalities in hypomelanosis of Ito

was to systematically investigate for structural hair shaft defects using a scanning electron microscope.

Hair samples from patients with X-linked hypohidrotic ectodermal dysplasia, pachyonychia congenita, tricho-dento-osseous syndrome, tricho-rhino-phalangeal syndrome, and hypomelanosis of Ito were collected and fixed onto probes with double-sided scotch tape, coated with 30 nm layer of gold/palladium alloy in a Polaron E 5100 Sputter Coater (Polaron Equipment Ltd., Watford, UK); the specimens were examined and photographed in a Philips SEM 515 microscope (Philips, Eindhoven, The Netherlands).

The girl with hypomelanosis of Ito had developed depigmented streaks in whorls on the trunk and extremities consistent with Blaschko’s lines. Her hair showed curly growth in stripes across the scalp and on the temporal regions, the alterations of normal and curly hair following the lines of Blaschko on the scalp. Using a scanning electron microscope, hair from the temporal region showed longitudinal grooves (Fig. 1A) and occasionally twisting (Fig. 1B); findings that explain the impaired hair growth in this region, owing to increased fragility of these hairs.

The hair of the patient with tricho-dento-osseous syndrome showed pronounced curling; odontological findings included small teeth, enamel defects and taurodontia of all molar teeth. Nails and skin were normal. Ultrastructurally, his hair showed longitudinal grooves (Fig. 2A).

The boy with anhidrotic ectodermal dysplasia had agenesis of all teeth in the lower jaw, and familial history and phenotype consistent with X-linked inheritance. In addition, he suffered from diffuse alopecia, absence of eyebrows, inability to sweat, and he had a supranumerous mamilla on the left thoracic side. The nails were normal. In scanning electron microscopy, his hairs showed longitudinal grooves (Fig. 3A), sometimes with twisting (Fig. 3B). The hairs from the patients with pachyonychia congenita and tricho-rhino-phalangeal syndrome did not show pathological findings.

To our knowledge, structural hair defects have not been reported in hypomelanosis of Ito, tricho-dento-osseous syndrome or pachyonychia congenita. Previously, flattening of hair with an elliptoid transverse section has been reported in patients with tricho-rhino-phalangeal syndrome (2, 3), findings not found in our patient.

The spectrum of primary hair shaft abnormalities reported in ectodermal dysplasia ranges from flattened hair (1–3), twisted hair (4), longitudinal impressions (5, 6), to a combination of both twisting and impressions of the hair shaft in pili torti et canaliculi (7) and corkscrew hairs (8). The different shaft abnormalities may be detected in various ectodermal dysplasias, sometimes in the same patient. Whereas longitudinal
grooves are a frequent finding (6, 9), pili torti et canaliculi appears to be a characteristic finding in ectodermal dysplasias combined with clefing of lip/palate (10). There thus seems to be a crescendo in structural abnormalities, with increasing shaft defects associated with the degree of clinical dysmorphology (6, 7), with corkscrew hairs as an exaggeration of pili torti et canaliculi (11, 12).

Secondary, unspecfic changes, such as trichorrhexis nodosa (13) and loss of cuticle (14), are due to the increased fragility often in seen in these hair shaft abnormalities. Differences in the calibre of the hair (15) and cuticular irregularities (7) are other findings previously reported. In our patients though, the cuticular pattern was regular, and no other signs of weathering of the hair shafts were seen.

In conclusion, we demonstrate structural hair shaft abnormalities in different ectodermal dysplasia; hair shaft abnormalities have not so far been described in hypomelanosis of Ito nor in tricho-dento-osseous syndrome.

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