

Uncombable hair and atopic dermatitis in a case of tricho-dento-osseous syndrome

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Summary

The tricho-dento-osseous syndrome is a rare genetic disorder due to a mutation in the DLX3 gene on chromosome 17q21. It can affect hair, teeth, bone and nails, causing phenotypic variability. We report on an 8-year-old girl with tricho-dento-osseous syndrome, who presented in our allergy center with severe atopic dermatitis. Additional clinical findings included light, kinky hair reminiscent of pili trianguli et canaliculi (uncombable hair), enamel hypoplasia and enlarged pulp chambers of the molar teeth (taurodontism). A genetic investigation revealed a de-novo mutation in the DLX3 gene on chromosome 17q21. Electron microscopic examination of the curly hair showed a flattened hair shaft with longitudinal grooves.

Keywords

tricho-dento-osseous syndrome – atopic dermatitis – pili trianguli et canaliculi – uncombable hair

Introduction

Tricho-dento-osseous (TDO) syndrome is a rare autosomal dominant disorder belonging to the ectodermal dysplasias. It is caused by a mutation of the DLX3 gene on chromosome 17q21 that, among others, is expressed in bones, hair and nails [1].

The most frequent clinical findings are uncombable hair, enamel hypoplasia,

enlarged pulp chambers of the molar teeth (taurodontism) as well as increased mineral density of the bones [2]. These signs and symptoms are expressed in variable clinical intensity. The diagnosis is made on a clinical basis and can be confirmed by genetic investigation.

Case report

An 8-year-old girl presented in our allergy center due to atopic dermatitis. She had severe atopic dermatitis on the eyelids, the arms and legs. Additionally, blond curly tousled hairs without increased fragility were noted. The nails as well as sweat gland function were normal. Inspection of the teeth revealed enamel defects and pointed conical canine teeth of the upper jaw. Otherwise, there were no other clinical abnormalities (Figure 1).

History

Atopic dermatitis had appeared during early childhood. Tangled hair had also been present since this time. Combing the hair led to knotting and only worsened its appearance. The girl also had repeated dental problems, most recently a gum abscess.

A panorama x-ray of dentition revealed enlarged pulp chambers (taurodontism) as well as enamel hypoplasia.

Allergy findings

The prick test showed moderately positive reactions to house dust mite,

dog epithelia and grass pollen. RAST towards house dust mite, dog epithelia, birch and grass pollen as well as a food mix (egg, milk, fish, wheat, peanut, soy) was negative; total IgE was in the normal range with 63 kU/l. Histamine was slightly elevated with 0.4 ng/ml; diamine oxidase as well as vitamin B6 were in the normal range.

Electron microscopy

A longitudinal groove in the hair shafts was already visible in light microscopy at 10-fold magnification. Subsequent electron microscopy of the hair revealed zones with flattened hair shaft and short longitudinal impressions at irregular intervals (Figure 2, 3).

Diagnosis

The above findings made tricho-dento-osseous syndrome the suspected diagnosis. An externally performed genetic investigation revealed a c.524 T>C mutation of the affected DLX3 gene causing replacement of isoleucine by serine at position 175 of the protein chain. Inheritance is autosomal dominant. As there were no indications of a similar mutation in the family history and the parents of the girl were not carriers of the mutation, our patient had a de-novo mutation.

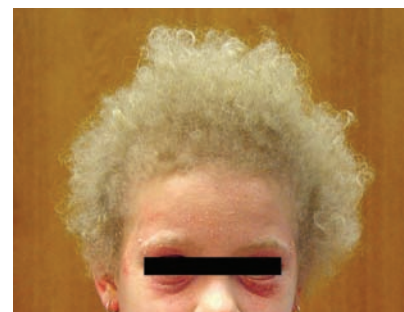


Figure 1: Tousled, uncombable hair and severe atopic eczema in a patient with TDO syndrome.

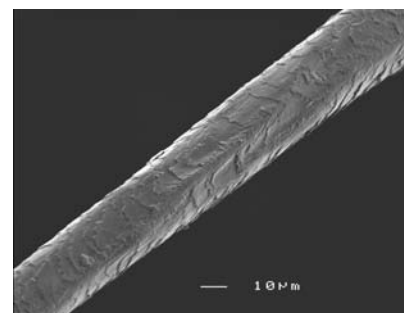


Figure 2: Electron microscopic investigation showing a flattened hair shaft with longitudinal grooves.

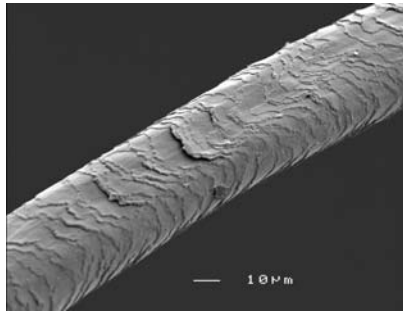


Figure 3: Electron microscopy showing impressions of the hair shaft.

Course

The atopic dermatitis was treated successfully. Due to the elevated histamine level a histamine-free diet was recommended; at a return visit two weeks later the histamine level had normalized. The girl remains under dental care.

Discussion

Ectodermal dysplasias constitute a large, heterogeneous group of non-progressive genetic diseases. Tricho-dento-osseous syndrome is considered the ectodermal dysplasia with a high penetrance even if the individual signs and symptoms can be present in variable intensities. Despite the variable clinical expression dental anomalies such as enamel hypoplasia and taurodontism as well as curly hair are the most common and constant manifestations [2] followed by thickened skull bones, flat and fragile nails as well as missing pneumatization of the mastoid [3].

The girl we present primarily attracted attention due to curly, unruly hair and enamel abnormalities. An anomaly of the canine teeth with a pointed conical form as sometimes observed in hidrotic ectodermal dysplasias [4] was also present in our patient. These findings together with the panorama x-ray of the upper and lower jaws revealing enlarged pulp chambers made the diagnosis of TDO syndrome appear likely.

Uncombable hair is characterized by shaggy, curly hair that cannot be combed. It can exist as an isolated anom-

aly of the hair shaft or may be associated with other anomalies in the framework of ectodermal dysplasias. Electron microscopy differentiates between pili canaliculi, pili trianguli et canaliculi as well as pili torti. Pili canaliculi have a longitudinal groove; pili trianguli et canaliculi also have a triangular or kidney-shaped cross-section. Pili torti have a hair shaft twisted by 180°. Occasionally several anomalies are present in the same patient. In TDO syndrome, macroscopically curly hair is present which could correspond macroscopically to pili canaliculi [5], but microscopy does not always reveal abnormalities. In our patient light microscopy already demonstrated zones with longitudinal impressions in some hair that could be confirmed by electron microscopy. In contrast to typical pili trianguli et canaliculi the alterations were not present over the entire length of the hair and were less marked.

The simultaneous occurrence of ectodermal dysplasias and atopic dermatitis has been reported by several authors [4, 6–8]. Our patient was also referred primarily because of severe atopic dermatitis. However, it is generally assumed that atopic dermatitis is an independent disease. Some cases of uncombable hair responded to biotin [9], in others therapy was unsuccessful. Spontaneous improvement of uncombable hair during puberty and adolescence has been reported. <<<

Conflicts of interest

None.

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