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CASE REPORT

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Uncombable hair improved by biotin

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Abstract

Uncombable hair syndrome (UHS) is a rare disorder of hair shaft morphology. UHS presents in early childhood with blonde, dry and frizzy scalp hair. The rate of hair growth may be normal or slow. The UHS hair shaft is characterized by a triangular shape with longitudinal grooves along the hair shaft axis. At writing, no curative treatment is available for UHS. However, research suggests that oral biotin supplementation results in a clinical improvement in growth rate and texture in some cases. Here, we present the case of a girl with UHS who was successfully treated with oral biotin supplementation. Our case highlights the diagnostic pathway in UHS, and demonstrates that two cycles of treatment with biotin, each separated by a period of 3 years, led to a significant improvement in combability and growth rate in a patient with UHS. We propose that biotin is a safe treatment option for UHS when both the health care provider and the family are aware of the possible interference with laboratory test results.

KEYWORDS

genodermatosis and biotin, hair shaft anomaly, PADI3, pili trianguli et canaliculi, uncombable hair syndrome

INTRODUCTION

Uncombable hair syndrome (UHS) is a rare disorder of hair shaft morphology. UHS presents in early childhood with blonde, dry and frizzy scalp hair. The rate of hair growth may be normal or slow. The UHS hair shaft is characterized by a triangular shape with longitudinal grooves along the hair shaft axis (Figure 1). As a result, the scalp hairs point in differing directions, which renders styling almost impossible.[1] While most cases of UHS are sporadic, familial cases also occur. Recently, our group elucidated the molecular genetic background of UHS, and demonstrated that in most of the patients, the condition is inherited in an autosomal recessive manner via pathogenic variants in the genes *PADI3*, *TCHH* and *TGM3*, which encode for proteins involved in hair shaft formation.[2] In some cases, an association with brachydactyly, ectodermal dysplasia, juvenile cataract and oligodontia has been reported. Here, we present the case of a girl with UHS who was successfully treated with oral biotin supplementation.

CASE REPORT

A 3-year-old girl presented to the dermatology department with a history of sparse and slow-growing hair since birth. The hair was difficult to style due to its frizzy and unruly texture. Light microscopy revealed triangular hair shafts with longitudinal canals, and molecular

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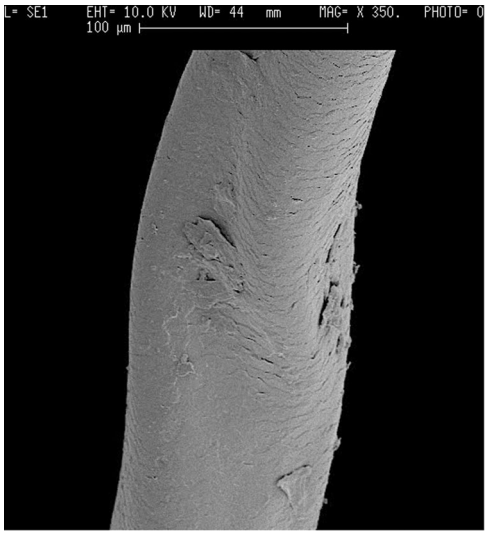

FIGURE 1 Scanning electron microscopy of the hair shaft from a patient affected by uncombable hair syndrome. Triangular aspects of the hair shaft can be observed.

genetic analysis showed the presence of the homozygous pathogenic variant c.881C>T; p.Ala294Val in exon 8 of the gene *PADI3*.[2] A diagnosis of UHS was therefore assigned, and treatment with oral biotin 5 mg once daily was initiated. At 6-month follow-up, the parents reported an improvement in combability. Clinical examination revealed a smoothening of scalp hair texture. Treatment was then discontinued, as the condition was expected to improve with age.

Three years later, the patient was rereferred with slow hair growth (Figure 2a,b). The earlier improvements in combability and smoothness of the hair had not been maintained after cessation with biotin. Treatment with oral biotin 5 mg once daily was resumed. At 6-month follow-up, the patient and her parents reported an improvement in both combability and growth. They also considered her hair to be glossier in appearance. Clinical examination revealed a smoothening of the scalp hair, and an average increase in length of 10 cm (Figure 3a,b). The patient also reported an increase in self-esteem secondary to the improvement in the condition of her hair. No treatment side effects were reported.

DISCUSSION

At writing, no curative treatment is available for UHS. However, research suggests that oral biotin supplementation results in a clinical improvement in growth rate and texture in some cases.[3,4] Biotin is a water-soluble vitamin otherwise known as vitamin B7, which serves as a cofactor in several carboxylase reactions, thus rendering it an

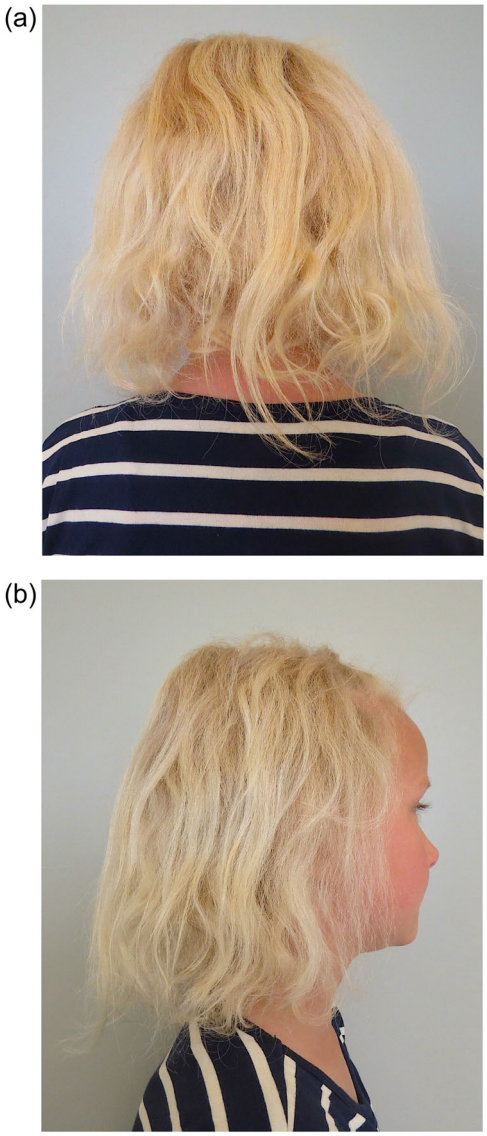

FIGURE 2 (a, b) Blonde, dry and frizzy hair in a patient with uncombable hair syndrome before treatment with oral biotin supplement.

essential component in multiple metabolic pathways.[5] When prescribing biotin, an important consideration for health care providers is the potential for interference with laboratory test results, since a high serum concentration of biotin can compromise immunoassays that are based on biotin-streptavidin technology.[5,6]

To conclude, our case highlights the diagnostic pathway in UHS, and demonstrates that two cycles of treatment with biotin, each separated by a period of 3 years, led to a significant improvement in combability and growth rate in a patient with UHS. We propose that biotin is a safe treatment option for UHS when both the health care provider and the family are aware of the possible interference with laboratory test results. In general, the prognosis of UHS is good, with improvement

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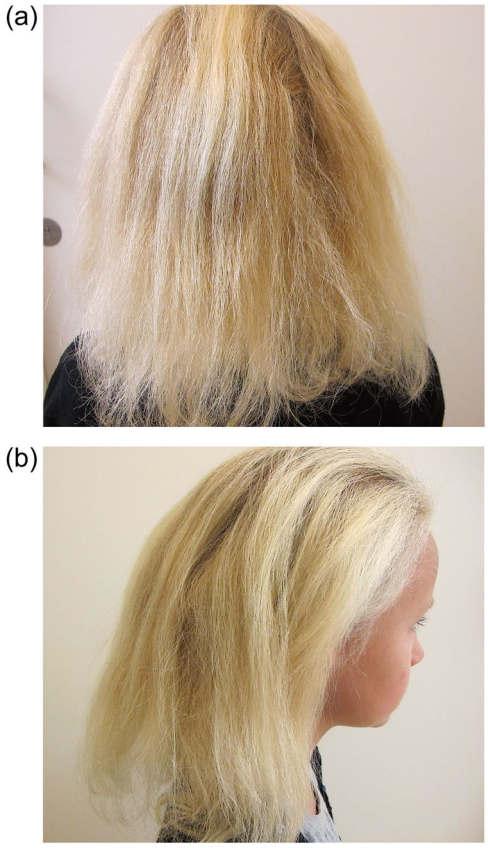

FIGURE 3 (a, b) At 6-month follow-up, the hair was longer and smoother following treatment with oral biotin supplementation.

occurring during puberty in a substantial proportion of cases.

AUTHOR CONTRIBUTIONS

Jakob L. Drivenes wrote the first draft and edited the final draft of the manuscript. Ramon Grimalt commented on the final draft of the manuscript. Regina C. Betz performed the molecular genetic analyses and edited the final draft of the manuscript. Anette Bygum was responsible for the clinical management of the patient, and cowrote and edited the first and final drafts of the manuscript.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

DATA AVAILABILITY STATEMENT

Research data are not shared.

ETHICS STATEMENT

The study did not need ethical approval. *Written informed consent for publication was obtained from the patient's parents.*

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