

Single Case

A Young Boy with Brittle Hair

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Keywords

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Abstract

Trichothiodystrophy (TTD) is a rare multisystem disorder with an autosomal recessive mode of inheritance. TTD presentations vary from only hair abnormalities like brittle, fragile hair to physical and mental retardation. Mutations of DNA repair genes have been identified as responsible for the disease. A 5-year-old boy presented with sparse, short, and brittle hair to our clinic. He was born to consanguineous parents. Trichoscopy and light microscopy revealed broken hairs with no specific shaft defect. Due to the inaccessibility of the polarized microscopy, a bedside technique was employed. We used a polarized dermatoscope and a mirror in order of achieving transillumination of the hair shafts, which revealed striking bright and dark bands. These bands are referred to as “tiger tail,” which is the pathognomonic sign of TTD. Subsequent polarizing microscopy also confirmed the clinical diagnosis. This highlighted a feasible method for observing the tiger tail, which expanded the known clinical diagnostic tools of TTD.

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Introduction

Trichothiodystrophy (TTD) is a rare, autosomal recessive disease resulting from mutations of DNA repair genes such as ERCC3 (XPB), ERCC2 (XPD), or GTF2H5 (TTDA) in photosensitive phenotypes; nonphotosensitive patients may have mutations in GTF2E2

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Fig. 1. **a** Sparse and brittle hair of a young boy. **b** Short broken hairs at trichoscopy ($\times 20$ magnification). **c** Light and dark bands of hairs at polarized transilluminating dermoscopy ($\times 10$ magnification). **d** Tiger tail pattern under polarizing microscopy ($\times 10$ magnification).

or C7orf11 and the ring finger protein RNF113A [1–5]. TTD can be associated with a broad range of clinical manifestations, from only hair abnormalities characterized by sulfur-deficient coarse fragile hair to severe neuroectodermal symptoms. These may include cutaneous photosensitivity, microcephaly, intellectual and growth retardation, impaired motor control, recurrent infections, ichthyosis, nail abnormalities, as well as decreased fertility. Congenital cataract, short stature, and skeletal abnormalities may also be seen. As a result, several acronyms have been used to distinguish the different clinical features of TTD patients, including PIBIDS, BI(D)S, and IBI(D)S, which are as follows: photosensitivity, ichthyosis, brittle hair, intellectual impairment, decreased fertility, and short stature [6, 7]. The clinical diagnosis can be made employing polarizing light microscopy and confirmed with genetic analysis and whole exome sequencing. Due to limited access to polarizing light microscopy, this process could be costly and time consuming. Herein, we report our experience using a simple bedside alternative method for the clinical diagnosis.

Case Report

A 5-year-old boy presented with sparse, short, and brittle hair (Fig. 1a). He was born to a consanguineous marriage with no relevant family history. His physical examination was otherwise normal, and he had no signs of ichthyosis, neurologic decline, photosensitivity, or ocular problems. Trichoscopy showed broken hairs and only nonspecific shaft changes (Fig. 1b). Hair shaft examination with light microscopy revealed no more findings. Hairs with surface irregularities were clipped and placed on a glass slide with a few drops of gel. The slide was put on a mirror, and the hair shafts were observed with the polarized mode of Fotofinder Medicam 1000 (Fig. 1c).

Discussion

Most hair shaft defects with brittle hair can be easily seen by either light microscopy or trichoscopy [8–10]. TTD is a disease characterized by brittle hair due to low sulfur and cysteine content of the hair shafts showing nonspecific changes on light microscopy [6]. The presence of hair brittleness without any specific shaft defect encouraged us to use a bedside test to screen TTD. The hallmark of TTD is observing the specific “tiger tail” pattern by polarized microscopy. Unfortunately, polarized light is not widely available in most laboratories; therefore, we opted a bedside technique proposed by Swanson and coworkers [11]. Accordingly, we used a polarized dermatoscope and a mirror to achieve transillumination of the hair shafts [11]. By this simple method, the banding of the birefringent hairs characteristic of TTD could be seen easily at bedside without sending the specimen to a laboratory equipped with a sophisticated polarized microscope.

We detected alternate light and dark bands by this quick method (Fig. 1c), and the tentative diagnosis of TTD was made. Polarized light microscopy confirmed our findings (Fig. 1d). Unfortunately, genetic study was not performed.

Statement of Ethics

The study conforms to the guidelines established by the Declaration of Helsinki. This retrospective review of patient data did not require ethical approval in accordance with local guidelines of the Institutional Review Board at Kawasaki Medical School. Written informed consent was obtained from the patient and his legal guardian for publication of this case report and any accompanying images.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contribution

Nassim Tootoonchi, Vahideh Azhari, and Zahra Razavi were responsible for managing the case and participated in the data collection. Shadab Seraji and Nika Kianfar participated in drafting of the case report and critical revisions. Hamidreza Mahmoudi and Maryam Daneshpazooch collaborated to conceive the study.

Data Availability Statement

All data generated or analyzed during this study are included in this article. Further inquiries can be directed to the corresponding author.

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