Case Report

Trichothiodystrophy without Associated Neuroectodermal Features in Two Siblings

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ABSTRACT

Trichothiodystrophy (TTD) is characterized by the common feature of sulfur-deficient brittle hair associated with a constellation of neuroectodermal symptoms. There is a wide phenotypic variation in the severity; ranging from isolated hair defect to multiple neuroectodermal symptoms such as photosensitivity, ichthyosis, intellectual impairment, decreased fertility, and short stature. This case report describes TTD in two sisters with only hair fragility and no other associated feature. This case highlights the variable clinical presentation of TTD and the need for regular follow-up in such patients for an early detection of any neurological, physical, and sexual impairment.

Key words: Brittle hair, neuroectodermal syndrome, trichothiodystrophy

INTRODUCTION

Trichothiodystrophy (TTD) is a rare autosomal recessive disorder characterized by the common feature of sulfur-deficient brittle hair associated with a constellation of neuroectodermal symptoms. The hair fragility unique to this disorder gives it its name, which is derived from Greek words – tricho, thio, dys, and trophe meaning hair, sulfur, faulty, and nourishment, respectively. Apart from the constant feature of hair shaft defect, there is a wide phenotypic variation in the severity; ranging from isolated hair defect to multiple neuroectodermal symptoms such as photosensitivity, ichthyosis, intellectual impairment, decreased fertility, and short stature. Here, we describe TTD in two sisters with only hair fragility and no other associated feature.

CASE REPORT

Two sisters, aged 3 years and 9 months, presented with sparse hair on scalp since birth. There was no history of having normal hair at any time after birth. Their parents were unaffected and had a nonconsanguineous marriage. Antenatal history was uneventful and both were born at term. There was no history of similar complaints in the family. On examination, sparse, thin, short rough hair was seen all over the scalp with relatively longer length hair in vertex and frontal areas of the scalp. Both had ciliary and supraciliary madarosis also. Multiple 1–3-mm follicular keratotic papules were present on the scalp, eyebrows, extensor aspects of extremities, and sides of the trunk. Hair, teeth, nails, and sweating were normal. There was no photosensitivity and physical or psychomotor impairment. Dry trichoscopy of scalp and eyebrows hair with cross-polarized dermatoscope (Cosderma DS100) showed short broken hair and the papular lesions revealed a broken hair underneath the skin indicating hair breakage inside the infundibulum. Hair shaft analysis on light microscopy revealed clean transverse term. There was no history of similar complaints in the family. On examination, sparse, thin, short rough hair was seen all over the scalp with relatively longer length hair in vertex and frontal areas of the scalp. Both had ciliary and supraciliary madarosis also [Figure 1a-c]. Multiple 1–3-mm follicular keratotic papules were present on the scalp, eyebrows, extensor aspects of extremities, and sides of the trunk. Hair, teeth, nails, and sweating were normal. There was no photosensitivity and physical or psychomotor impairment. Dry trichoscopy of scalp and eyebrows hair with cross-polarized dermatoscope (Cosderma DS100) showed short broken hair and the papular lesions revealed a broken hair underneath the skin indicating hair breakage inside the infundibulum [Figure 1d]. Hair shaft analysis on light microscopy revealed clean transverse
fractures (trichoschisis), trichorrhexis nodosa, irregular cortical outline, and distal brush breaks [Figure 2a-c]. On polarized microscopy, characteristic tiger-tail banding was observed [Figure 2d]. Based on the characteristic light and polarized microscopy of the hair shaft and in the absence of other neurological and cutaneous symptoms, a final diagnosis of TTD without associated neuroectodermal syndrome was made.

**DISCUSSION**

The clinical spectrum of TTD and its associated syndromes is complex and is further expanding with the increasing understanding of the cellular, genetic, and molecular mechanisms. Recent gene studies have pointed toward the nucleotide excision repair defect in TTD patients with photosensitivity, putting it in the same group as xeroderma pigmentosum (XP) and Cockayne syndrome. For the patients without photosensitivity, no gene has been isolated as yet.\[1\] Brittle hair and lack of predisposition to cancers is a feature which separates TTD from other DNA repair defect diseases like XP.

Brittle hair is the hallmark of the disease and is the only mandatory clinical feature for the diagnosis of TTD. It has been concluded in a previous study with a series of TTD patients and controls that it can be reliably diagnosed on the basis of clinical examination and careful light microscopy showing hair shafts with irregular contour and two distinctive features of trichoschisis and trichorrhexis nodosa-like defects and tiger-tail banding on polarized microscopy.\[1\] Amino acid analysis can be used for confirmation. Our patients exhibited the short brittle hair with the characteristic light and polarized microscopy findings. Amino acid analysis could not be done in our patients due to its unavailability.

To the best of our knowledge, only five such cases of “mild” TTD without associated neuroectodermal features have been reported in the literature.\[5-8\] Although not exhibiting any other features at this stage, a diagnosis of TTD in such patients warrants regular follow-up for early detection of any neurological, physical, and sexual impairment.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest

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