Trichothiodystrophy with gonadal hypoplasia and delayed development: A case report

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ABSTRACT:

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Trichothiodystrophy (TTD) encompasses the group of autosomal recessive disorders characterized by sulfur-deficient brittle hair and other neuroectodermal disorders which are highly variable in expression and severity. The hallmark of hair finding is alternating light and dark bands (tiger tail hair) under polarizing microscopy. We report a case of a 5-year-old Thai boy with TTD. He presented with short, brittle and coarse scalp hair since birth. The physical examination revealed micropenis and global delayed development.

Key words: Delayed development, gonadal hypoplasia, sulfur-deficient brittle hair, tiger-tail hair, trichothiodystrophy

บทคัดย่อ:

อลิสา ทั่งตระกูล, กอบกุล อุณหโชค รายงานผู้ป่วย TRICHOTHIODYSTROPHY ที่มีระบบอวัยวะสืบพันธุ์เจริญผิดปกติและพัฒนาการช้า วารสารโรคผิวหนัง 2557; 30: 141-145.

แผนกผิวหนัง กองอายุรกรรม โรงพยาบาลพระมงกุฎเกล้า

Trichothiodystrophy เป็นกลุ่มโรคทางพันธุกรรมที่มีลักษณะสำคัญได้แก่ ผมเปราะแตกง่ายจากการขาดซัลเฟอร์ ร่วมกับมีความผิดปกติของ neuroectoderm ทั้งนี้การแสดงออกและความรุนแรงของโรคจะแปรผันได้มากในผู้ป่วยแต่ละราย ลักษณะเด่นของเส้นผมที่พบได้จากการตรวจโดยกล้องจุลทรรศน์ชนิดโพลาไรซ์ ได้แก่ แถบสว่างสลับแถบมืดลักษณะคล้ายหางเสือ ในรายงานนี้ได้นำเสนอ เด็กชายไทยอายุ 5 ปี ได้รับการวินิจฉัยเป็นโรค trichothiodystrophy ผู้ป่วยมีผมเส้นสั้นหยาบ เปราะ ทั่วศีรษะ ตั้งแต่กำเนิด ร่วมกับการตรวจพบองคชาติขนาดเล็ก และพัฒนาการช้า

คำสำคัญ: Trichothiodystrophy, Tiger tail hair, ผมเปราะแตกจากการขาดซัลเฟอร์, อวัยวะสืบพันธุ์เจริญผิดปกติ, พัฒนาการช้า

Introduction

Trichothiodystrophy (TTD) is a heterogeneous group of autosomal recessive disorders with highly variable in expression, severity and phenotypes that range from isolated hair defect, severe neuroectodermal disorder to xeroderma lpigmentosum (XP)-like change.[1,2] The hallmark of hair finding is alternating light and dark bands (tiger tail hair) under polarizing microscopy. The majority of patients have XPD gene mutations and a few have mutations in XPB or TTDN1. These genes play roles in nucleotide-excision repair (NER) and transcription.[1,2] In contrast to XP, TTD patients with these mutations have not been reported to have increased risk of skin cancers.[1-3]

Case report

A 5-year-old boy presented with short, brittle and coarse scalp hair since he was born. His hair had grown to be only a short length and then fell off (Figure 1 a-b). The hair shedding was increased after the episodes of upper respiratory tract infection. His mother denied history of hair pulling or any other skin lesion. The physical examination revealed diffusely sparse and short scalp hair, varied in length up to 2 centimeters with positive hair pull test. Other systems were normal. The polarizing microscopy of the hair demonstrated alternating light and dark bands (Figure 2). Scalp biopsy showed presence of hair follicles with sparse lymphocytic infiltration. In addition, he had global delayed development and small penis which were in an investigation process. Although the hair amino acid analysis was not performed, the polarizing microscopic finding of hair and neuroectodermal abnormalities in this patient were consistent with the diagnosis of TTD. His hair problem was treated with topical 3% minoxidil

solution and family counseling. He had longer hair shaft but his mother observed increased hair shedding following the episodes of any illness.



Figure 1 (a) and (b) Clinical presentation with diffusely short, brittle and coarse scalp hair of the patient



Figure 2 Polarizing microscopy showed of alternating light and dark band (tiger tail hair)

Discussion

TTD, a classic differential diagnosis of congenital alopecia, is characterized by dry, coarse and sparse hair which tends to break after minor trauma. Intermittent hair loss is also observed during infection.[1,4] Hair abnormalities are found in all reported TTD cases. Nearly all have hair shaft defects and tiger-tail banding on polarizing microscopy which was found in 73% of patients.[4,5] Other hair findings are sparse hair (48%) and alopecia (39%).[4] The most common cutaneous finding was ichthyosis (65%) and nearly half of these patients had collodion membrane at birth. The second most frequent finding reported was photosensitivity (50%), mostly found in case of XPD gene mutation.[4] Other skin findings include dry skin (21%), eczema (8%) and freckles (7%).[4]

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Clinical spectrum of associated signs and symptoms in TTD is extensive. Besides those mentioned above, nervous system, ocular system, cardiopulmonary system, hematologic system, musculoskeletal system, genitourinary system, and hypogonadism have also been described.[1,4]

The diagnosis of TTD includes evaluation of hair features especially under polarizing microscopy that shows striking alternating bright and dark bands along entire hair shaft referred to as "tiger tail bands" which is highly characteristic.[6,7] In addition, trichoschisis, trichorrhexis nodosa-like fracture and ribboning can also be found.[6,8] Confirmation of the diagnosis by amino acids analysis of hair shaft shows low cysteine and low sulfur content.[6,7] The mechanism by which the low hair sulfur content causes hair abnormalities is unclear. A prior study from 15 TTD patients revealed the increased hair sulfur content and the decreasing frequency of hair defects detected.[6]

Our patient presented with diffusely sparse hair and failure to attain normal hair growth since birth. His hair shaft showed tiger-tail banding pattern on polarizing microscopy which is the characteristic feature of trichothiodystrophy. In addition, he also has micropenis. There are few reports of urologic malformation and gonadal dysfunction in TTD.[5-7] This may be explained by a defect in neuroectoderm similar to the report of TTD associated with photosensitivity, gonadal failure and striking osteosclerosis.[9]

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