Skinning the Surface of Bone Abnormalities in Trichothiodystrophy

Rina Allawah, Michael Xiong, Deborah Tamura, RN, John J. DiGiovanna, MD, Michael Collins, MD, Kenneth H. Kraemer, MD, Suvimil Hill, MD

National Cancer Institute
National Institutes of Health

INTRODUCTION

Trichothiodystrophy (TTD) is a rare autosomal recessive disease characterized by multisystem abnormalities: low birth weight, short stature, developmental delay, microcephaly, absent myelin in the brain, ataxia and altered reflexes, congenital cataracts, photosensitivity, ichthyosis and sulfur-deficient, brittle hair (Figure 1).

Bone abnormalities, which include central osteосlerosis, peripheral osteосlerosis, and hip abnormalities, such as coxa valga (Figure 1), subluxation and avascular necrosis, significantly impact the quality of life of TTD patients.

Patients with TTD exhibit a diagnostic “tiger-tail” banding pattern in hairs examined utilizing polarized microscopy (Figure 2). The underlying cause of TTD involves mutations in the genes, XPB, XPD, TTD3A, which collectively play a role in DNA repair and basal transcription (1). TTD1 gene, whose function is unknown, is also associated with TTD (1).