Brittle hair, developmental delay, neurologic abnormalities, and photosensitivity in a 4-year-old girl

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CASE SUMMARY

History

A 4-year-old Caucasian girl presented to the National Institutes of Health (NIH) for evaluation of sparse brittle hair, developmental delay, poor growth, recurrent infections, and photosensitivity (Fig 1 and Table I). The patient was born preterm at 35 weeks after a pregnancy complicated by elevated maternal alpha-fetoprotein at 16 weeks and pregnancy-induced hypertension beginning at 26 weeks. At birth, her skin showed generalized erythroderma and she was described as having a collodion membrane (Fig 1, *A*) that resolved over 2.5 weeks. By 1 month of age, the patient was reported to have congenital ichthyosis and mild scaling on her scalp, trunk, and lower extremities. By 23 months, she was reported to sunburn with minimal sun exposure and sweat very little in hot environments. Short brittle hair, nail spooning, head tremor (titubation), and asymmetric horizontal nystagmus were also noted. The patient did not sit unassisted until 12 months and did not crawl until 22 months.

Abbreviations used:

MRI: magnetic resonance imaging
NIH: National Institutes of Health
TTD: trichothiodystrophy
XP: xeroderma pigmentosum

A magnetic resonance imaging (MRI) scan of her brain at 19 months was read as normal. By age 3 years she had ataxia and ambulated only with a reverse walker. The patient experienced multiple ear infections and pneumonias beginning at 6 months of age that did not require hospitalization, were not cultured, and responded to empiric antibiotic treatment. Her parents reported that when she experienced severe respiratory infections the patient had temporary developmental regression that improved when the infection resolved. Complement and immunoglobulin levels were within normal limits at 16 months.

There was no family history of individuals with similar features. The parents denied consanguinity. The patient had an 8-year-old unaffected brother who was delivered after an uneventful pregnancy.

Physical examination

At the NIH, the patient was a cheerful, engaging girl with weight and height in the third centile and head circumference in the 10th centile. Examination revealed short brittle hair on her scalp and eyebrows but long eyelashes (Fig 1, *B*). She had mild micrognathia and a high-arched palate. The patient also had hyperlinear palms and soles and thin, peeling, spoon-shaped toenails (Fig 1, *C*). Mild hyperkeratosis was present on the side of the heels of both feet and fine scales were observed on the side of the trunk and ankles. The patient walked with an uneven stiff gait. Ophthalmologic examination revealed astigmatism in both eyes with no signs of cataracts or nystagmus. A summary of the clinical features is listed in Table I.

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Conflicts of interest: None declared.

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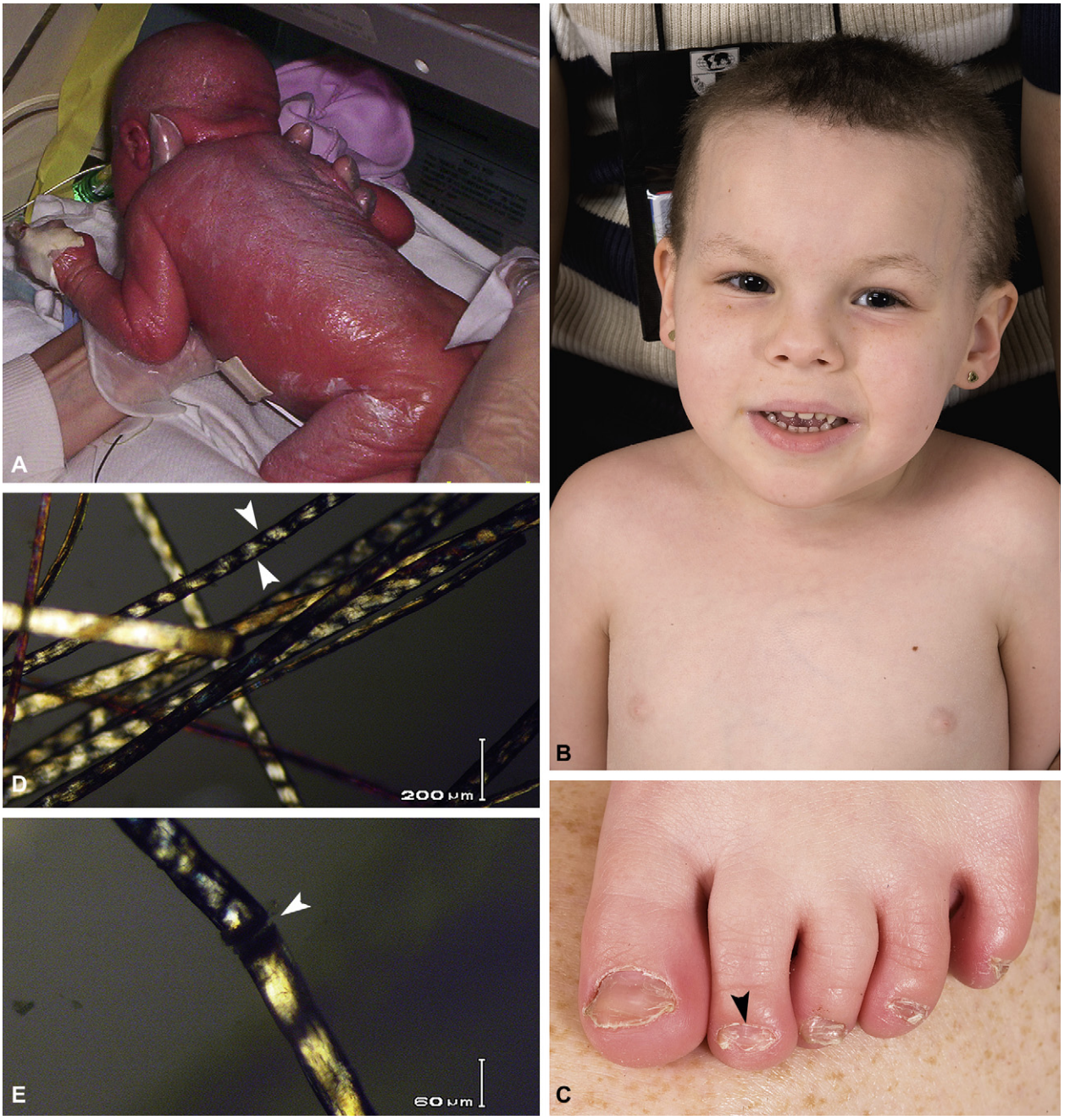

Fig 1. Clinical features of patient TTD421BE with trichothiodystrophy. **A,** Shiny, collodion membrane appearance at birth. **B,** Facial appearance showing short, sparse, brittle hair on scalp and eyebrows but long eyelashes. **C,** Thin, peeling, spoon-shaped nails (koilonychia) (*arrowhead*). **D,** Tiger-tail banding of cut hair under polarized microscopy (×4) (*arrowheads* point to alternating dark and light bands on hair shafts). **E,** Trichoschisis (×10) (*arrowhead*).

Diagnostic studies

Clinical laboratory tests at the NIH were significant for leukopenia (white blood cell count, 4780/μL [reference: 4860-13,180]), neutropenia (absolute neutrophil count, 1100/μL [reference: 1600-8290]), elevated hemoglobin A_2, 4.3% (reference: 2.2-3.2), IgM deficiency, less than 21 mg/dL (reference: 24-210), and elevated creatine kinase, 245 U/L (reference: 0-143). She had a normal mean corpuscular volume, 73.3 fL (reference: 72.3-85.0), 25-hydroxyvitamin D, 38 ng/mL (reference: 10-80), and 1,25-dihydroxyvitamin D, 69 pg/mL (reference: 24-86).

Cut hair was examined under polarization with a light microscope and revealed consistent light and