Expert Shares Challenging Cases From Stanford

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CORONADO, CALIF. — Making the correct diagnosis and choosing the best therapy are standard goals of practice, but sometimes that's easier said than done.

At the annual meeting of the Pacific Dermatologic Association, Dr. Anna L. Bruckner discussed three cases to illustrate that point.

In the first case, a 6-year-old girl with suspected loose anagen syndrome was referred to Dr. Bruckner, who is director of pediatric dermatology at Lucile Packard Children's Hospital in Palo Alto, Calif. In this condition, the anagen hairs are loosely anchored into the scalp so that the hair will fall out with very minor trauma. The hair is short, sparse, and seldom cut. It typically is seen in blond girls aged 2-5 years, but can affect boys and brunettes as well.

Dr. Bruckner did a gentle hair pull test and only two hairs came out. The girl's hair was very short and had a matted appearance in the back. She prescribed 5% minoxidil lotion and scheduled a 3-month follow-up visit. On follow-up, the girl's hair was fuller but it remained short and gentle hair pull tests remained negative.

"We obtained some additional history," said Dr. Bruckner, also assistant professor of dermatology and pediatrics at Stanford (Calif.) University. "Her nails were thin, often peeled, and never required trimming. She had no history of dental anomalies, and she'd had a coarse, deep voice since age 2. Her mother said that she looked different than her siblings." She also had sparse lateral eyebrows, a pear-shaped nose, and a thin upper lip.

The combination of short, sparse hair and abnormal facial features led Dr. Bruckner to consider trichorhinophalangeal syndrome (TRPS) type 1 as the diagnosis. An x-ray of the girl's hand performed after her follow-up visit revealed cone-shaped epiphyses of the phalanges, which confirmed the diagnosis.

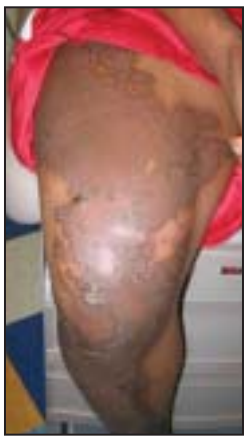
TRPS type 1 is an autosomal dominant disorder characterized by craniofacial and bony abnormalities that include sparse, slow-growing hair and thin lateral eyebrows, a pear-shaped nose, elongated philtrum and thin upper lip, prominent ears, and cone-shaped epiphyses of the phalanges. Variable findings include short stature (the patient was in the 25th percentile for height), nail abnormalities, teeth abnormalities, and a deep voice. The condition is caused by mutations in the TRPS1 gene.

Although there is no specific treatment for TRPS type 1, the parents were happy to better understand their daughter's condition. She has continued to use 5% minoxidil for 6 months with some improvement.

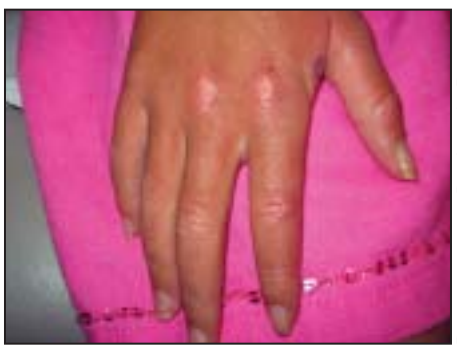
In another case, a 16-year-old African American boy presented with a 1-year history of a rapidly enlarging, pink to brown, scaly plaque on the right lower extremity that extended onto the thigh, shin, and toes.

He had been seen by other dermatologists and previous diagnoses included epidermal nevus and linear psoriasis. Topical treatment with clobetasol, calcipotriene, and tazarotene led to minimal improvement, but the patient was concerned that he was developing significant postinflammatory hyperpigmentation.

Close examination of the skin change revealed a thread-like hyperkeratotic bor-



Scaly plaque is shown on the right lower extremity of the boy with linear porokeratosis. After a course of imiquimod, he opted out of therapy, but his condition is still closely monitored.



PHOTOS COURTESY DR. ANNA L. BRUCKNER

Papules, plaques are seen on knuckles of the girl with erythropoietic protoporphyria.

der. Biopsy of this area showed a cornoid lamella, which is seen in porokeratosis. Dr. Bruckner eventually diagnosed linear porokeratosis, which presents in infancy or childhood. The lesions follow the line of Blaschko.

Treatments include the use of topical retinoids, imiquimod, and fluorouracil. Destructive therapies include cryotherapy, electrodesiccation, laser ablation, and excision. In many cases, however, recurrence of the lesion is common and all of the treatments have potential adverse effects.

After the diagnosis was made, they tried a course of imiquimod. "The boy did not feel that there was any improvement and he developed significant postinflammatory hyperpigmentation," she said.

In this case, Dr. Bruckner and her associates decided that the best therapy was no therapy, but they continue to monitor the lesion for worrisome changes. The decision "was controversial, but it was made with the family. It's something that they're comfortable with at this point," she said.

The third case Dr. Bruckner discussed was that of a 9-year-old girl who was referred by a rheumatologist for evaluation of possible dermatomyositis. The girl had a 4-month history of intermittent redness and swelling of the hands that worsened after prolonged outdoor activities.

The girl was healthy and described one remote episode of burning hands after a hike several years before. She was on naproxen and ranitidine, which had been prescribed by the rheumatologist as treatment for the redness and swelling.

Her family history was unremarkable—she had no muscular weakness or abdominal pain, and the work-up by the rheumatologist was negative for autoimmune disease.

Clinical exam revealed a few waxy papules and plaques distributed over the knuckles. Her hands also had a slightly weather-beaten appearance. A skin biopsy showed cuffs of hyaline material around the superficial blood vessels in the upper dermis, suggesting a diagnosis of erythropoietic protoporphyria (EPP). Confirmatory studies demonstrated that the patient had elevated total red blood cell porphyrins with a predominance of free protoporphyrin.

EPP is the most common type of porphyria in children. It presents between the ages of 1 and 6 years, and symptoms include burning, stinging, redness, and edema, which all occur after sun exposure. The condition is caused by a deficiency of ferrochelatase, which leads to accumulation of protoporphyrin IX. Treatment involves sun avoidance, sunscreens, and beta-carotene 30-150 mg/day. ■