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 dermatology 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.

Many of the girl's friends at school "had long, flowing hair and she wanted to see if there was something we could do about her hair," Dr. Bruckner said. She 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.

Dr. Bruckner 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," Dr. Bruckner recalled. "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 from 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 why their daughter's hair failed to grow normally. She has continued to use 5% minoxidil for 6 months with some improvement.

In another challenging 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. The lesion 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 border. Biopsy of this area showed a cornoid lamella, which is seen in porokeratosis.

Ultimately, Dr. Bruckner diagnosed linear porokeratosis, which presents in infancy or childhood. The lesions follow the line of Blaschko.

When skin conditions lead to social isolation, we need 'parents who are willing to be advocates for their children.'

DR. BRUCKNER

"The presumed pathogenesis is dysregulation of the keratinocytes, which leads to premature apoptosis of keratinocytes," said Dr. Bruckner, also assistant professor of dermatology and pediatrics at Stanford (Calif.) University. "The clinical concerns are for cosmesis and the potential for developing squamous cell carcinoma within the lesion."

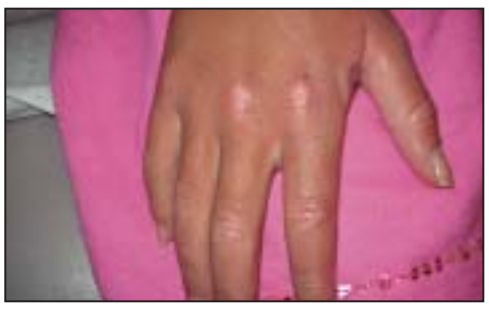
Treatments include the use of topical retinoids, imiquimod, and fluorouracil. Destructive therapies include cryotherapy, electrodesiccation, laser ablation, and excision. "However, in many of these cases recurrence of the lesion is common and all of these treatments have potential adverse effects," she noted. "This raises the question: Is treatment necessary?"

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 ultimately decided that the best therapy was no therapy, but they continue to monitor the lesion for worrisome changes.

This decision "was controversial, but it was a decision that was made with the family," she said. "It's something that they're comfortable with at this point."

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 patient had a 4-month history of intermittent redness and swelling of the hands that worsened after prolonged outdoor activities.



A 9-year-old girl with erythropoietic protoporphyria presented with waxy papules and plaques on her knuckles.

PHOTOS COURTESY DR. ANNA L. BRUCKNER

Scaly plaque is seen on the right lower extremity of a 16-year-old boy diagnosed with linear porokeratosis.

The girl was healthy and described one remote episode of burning hands following 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," Dr. Bruckner said. "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 1 and 6 years of age 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.

The girl developed a sense of social isolation because she attended a school where the children ate lunch and played outside. "She had to eat lunch off in a corner by herself, so she really was not able to interact with her peers when she was at school," Dr. Bruckner said. "This was very distressing for her. In addition, the beta-carotene pills were large and difficult to swallow."

Luckily, she said, the girl had a "tenacious" mother who worked with school officials to create opportunities for her daughter to socialize in shaded or indoor areas during lunch and recess.

"We need to have parents who are willing to be advocates for their children," Dr. Bruckner said. ■

Hemangioma Treatment Does Not Affect Infant Bone Density

CHICAGO — Bone density was no different in children who were treated with oral glucocorticoids for hemangiomas of infancy than it was in healthy controls, based on data presented in a poster at the annual meeting of the Society for Pediatric Dermatology.

Although oral glucocorticoids are considered the first choice of medication for the treatment of infant hemangiomas, concerns persist about the risk that these children will develop osteoporosis because glucocorticoids may prevent the formation of new bone, wrote Dr. Amy J. Nopper, a dermatologist at the Children's Mercy Hospitals and Clinics in Kansas City, Mo.

To assess the possible impact of systemic glucocorticoids on the density of children's bones, Dr. Nopper and her colleagues compared 35 children (mean age 44 months) who received glucocorticoids for hemangiomas for an average of 8.5 months with 35 controls.

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The average treatment dose was 2.2 mg/kg per day of prednisolone. The average body mass index was approximately 16 kg/m^2 for both the treatment and control groups.

The researchers measured the children's bone density after they had been off treatment for a period of at least 1 year. The results showed that the average spinal bone mineral density was the same (0.6 g/m^2) for both the treatment and control groups. The average total bone mineral density also was the same for both groups of children (0.8 g/m^2), and no significant differences appeared in the tibial ultrasound measurements between the two groups.

The results complement findings from other studies that have shown that the use of corticosteroids for the treatment of hemangiomas in early childhood does not prevent children from catching up in growth and achieving normal adult height, noted Dr. Nopper.

—Heidi Splete