Developmental Delay a Key To Hypomelanosis Diagnosis

BY SHERRY BOSCHERT
San Francisco Bureau

STANFORD, CALIF. — If a young child exhibits both global developmental delay and stripes or swirls of skin hypopigmentation on the trunk, get a peripheral blood sample for chromosome analysis.

Hypomelanosis of Ito presents as developmental delay plus swirls or patches of hypopigmentation or depigmentation along the lines of Blaschko, Dr. Louanne Hudgins said at a pediatric update that was sponsored by Stanford University.

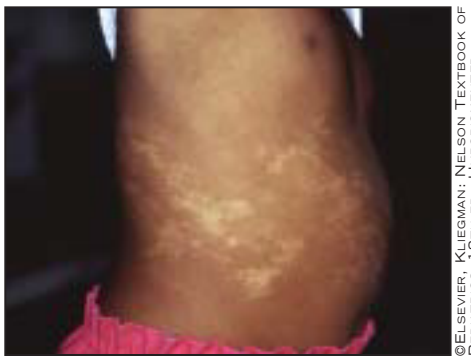
Blaschko's lines are a nonrandom cutaneous distribution pattern of pigment anomalies caused by migration of skin cells that is believed to start during embryogenesis, she explained.

About half of the people with hypomelanosis of Ito will show chromosomal mosaicism, which means that there is more than one cell line in the chromosomes. The skin lesions and developmental delay plus chromosomal mosaicism clinch the diagnosis of this rare disorder, said Dr. Hudgins, professor of pediatrics and chief of medical genetics at Stanford.

Making the diagnosis explains both the skin findings and the developmental delay and eliminates the need for any further workup to find the cause of either problem, she noted.

The diagnosis also can give parents information about the risk for recurrence. Chromosomal mosaicism indicates that a normal cell line is present and that the abnormal cell line probably developed after fertilization took place. "The likelihood that parents would have another child like this would be low," she said.

From 40% to 60% of patients with hypomelanosis of Ito will have structural brain abnormalities or mental retardation with or without seizures.



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Marbled hypopigmented swirls or patches can be seen on the abdomens of patients who have hypomelanosis of Ito.

This risk is the same in all patients with hypomelanosis of Ito, regardless of whether they have chromosome abnormalities or normal karyotypes.

Although the skin lesions can be present at birth, "most of the cases I've seen did not become apparent until later in childhood—around 18 months to 3 years," said Dr. Hudgins, who reported having no conflicts of interest.

If it is not possible to get a peripheral blood sample, take a skin biopsy, preferably from an area bordering both hypopigmented and hyperpigmented cells, she advised. Send the sample to the cytogenetics lab, which will grow the fibroblasts and then analyze chromosomes from the fibroblasts.

If hypomelanosis of Ito is suspected because of skin lesions but the child is meeting developmental milestones, there's no need to do a genetic workup for this disorder, she said. ■

Reassurance Is Best Rx for 'No Worries' Dermatoses

BY BETSY BATES
Los Angeles Bureau

LAS VEGAS — Too often faced with worrisome hemangiomas, grim genetic dermatoses, or serious drug eruptions, Dr. Fred Ghali relishes the chance to tell a family: "No worries."

Such is the case with three common but sometimes unrecognized diagnoses presenting to Dr. Ghali's pediatric dermatology practice in Grapevine, Tex.

He shared these benign conditions with his colleagues at a dermatology seminar sponsored by Skin Disease Education Foundation:

► **Psuedo acne.** Most parents recognize that children are maturing earlier these days, but they still tend to panic when they see what they think is acne developing in their 5- or 6-year-old. Take a good, hard look at the location and pattern of the small white papules on a young child's nose, suggests Dr. Ghali.

The papules are likely "pseudo acne," small milia created when a child constantly rubs his or her nose, often in response to nasal allergies. If these miniature cysts rupture, they may take on an inflammatory appearance that resembles acne.

Less nose-rubbing will help, and topical comedolytics and antibiotics may be prescribed if necessary. However, Dr. Ghali's treatment of choice is pretty simple: "Reassurance to the family."

► **Striking striae.** Mom may gasp when she sees deep, dark, horizontal marks lining the back of her 13- or 14-year-old son.

"It is quite impressive when you see this," Dr. Ghali acknowledged, pointing to the welt-like striations.

He first theorized that the bands of discoloration might be caused by the carrying of heavy backpacks, or might be the result of wacky skateboard maneuvers.

"The bottom line is [adolescents with this condition] have lots of vertical growth over a short period of time. They're usually [white] children who are extremely skinny," he said.

The striae will tend to fade over time, resolving far better than striae of pregnancy. No treatment is necessary.

► **Retention keratosis.** Darkly pigmented, nonpruritic regions in the flexural areas of a child's neck or underarm might point to a diagnosis of acanthosis nigricans. But what about when you see these symptoms in a child of normal weight, with no other signs of metabolic illness?

"You can look like a hero," said Dr. Ghali. "Just walk in with a little bit of alcohol and wipe it off."

Known by many names ("kitschy keratosis" when he was in training), this condition, common to young children, requires just two words to concerned parents: "No worries," said Dr. Ghali.

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Atypical Features of Scalp Nevi Also Seen in Young Adults

BY SHERRY BOSCHERT
San Francisco Bureau

SAN FRANCISCO — Histopathologic features of scalp nevi in children and adolescents that overlap with features of Clark's or dysplastic nevi also can be seen in scalp nevi in young adults, results of a study of 89 hairline and scalp nevi found.

The features may trigger melanoma concerns, so it is important to be aware of them to improve diagnostic accuracy and prevent unnecessary concerns or overtreatment, Dr. Betsy N. Perry said at the annual meeting of the American Society of Dermatopathology.

The nevi in this review came from 84 patients (ages 3-35 years) whose samples were filed in the dermatopathology service at the University of California, San Francisco. They showed characteristics common to other "nevi of special sites" such as melanocytic nevi on acral surfaces, genitalia, flexural areas, the breast, and in and around the ear, she and her associates reported.

In 63 (95%) nevi from 66 children and adolescents (aged 3-18 years) and in 19 (83%) of 23 nevi from young adults (aged 19-35 years), the lesions contained large nests of cells that were composed primarily of pigmented epithelioid melanocytes and/or were distributed irregularly along the dermal-epidermal junction. More often than not, the melanocytes extended onto hair follicles and other adnexal structures in large nests or singly or in small clusters. Adnexal involvement was seen in 44 nevi (67%) from children/adolescents and 14 (61%) from young adults.

The findings support previous reports of these characteristics in scalp nevi of children and adolescents. "These are also seen in young adults, which is something that was not really clear in the literature before," said Dr. Perry of the University of Utah, Salt Lake City.

Suprabasilar scatter was rare, and seen in five (8%) nevi in children and adolescents and in none of the nevi from adults.

Among architectural features, squaring or bridging of the rete was common. Squaring was seen in 51 nevi (77%) in children/adolescents and 22 nevi (96%) in adults, and bridging was found in 61 nevi (92%) in children/adolescents and 20 (87%) in adults. "In some of the lesions, a concomitant congenital pattern was appreciated" in 25 (38%) of nevi in children/adolescents and in 5 (22%) in adults, she said.

Epithelioid melanocytes were very common, and appeared in 64 (97%) of nevi from children/adolescents and 22 (96%) from adults, Dr. Perry emphasized. Other cytologic features included atypical melanocytes in only three (5%) of nevi in children/adolescents and in none from adults.

"When cytologic atypia occurred, it was rare, and could either be in the epidermal or the dermal component," she said.

Dusty melanin commonly was present within keratinocytes, which is not known to have clinical significance but as a practical matter can make it difficult to determine circumscription and to look for melanocytes within these lesions, she added. In 33 (50%) of nevi from children/adolescents and 15 (65%) from adults, dusty melanin was present in keratinocytes.

Stromal features that were observed in most samples included lamellar fibrosis in 62 nevi (94%) from children/adolescents and in all nevi from adults. Lymphocytic infiltrate was seen in 55 nevi (83%) from children/adolescents and in 16 (70%) from adults. Melanophages appeared in 58 nevi (88%) from children/adolescents and in 18 (78%) from adults.

All nevi in the study had been removed from patients because they appeared clinically atypical.

These findings support results of two 2001 studies that characterized scalp nevi in children and adolescents and showed that they share features with "nevi of special sites."

A more recent analysis of atypical nevi of the scalp found features that were not commonly ascribed to either Clark's or "dysplastic" nevi in 4 (10%) of 39 nevi from adolescents but not in 30 nevi from children or 160 nevi from adults (J. Cutan. Pathol. 2007;34:365-9). ■