Developmental Delay a Key To Hypomelanosis Diagnosis

BY SHERRY BOSCHERT
San Francisco Bureau

San Francisco — 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.

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 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, certain 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.

• Pseudo 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.”

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

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 by Hajrullah et al. published in J. Cutan. Pathol. 2007;34:365-9.

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.

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 (30%) of nevi from children/adolescents and 15 (65%) from adults, dusty melanin was present in keratinocytes.

These findings support previous reports of these characters 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.

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.)