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, genital dermatoses, or serious drug eruptions, Dr. Fred Ghali reassures concerned parents: “No worries,” he said.

“No worries,” he said. “These are also seen in young adults, which is something that was not really clear in the literature before,” she said.

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

Re petition 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?

“Should you 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.

SDFP and Skin & Allergy News are subsidiaries of Elsevier.