Incontinentia Pigmenti Lesions Evolve

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LAS VEGAS — If a newborn girl develops vesicles or pustules over the trunk and extremities, think incontinentia pigmenti, Dr. Moise L. Levy said at meeting sponsored by the American Academy of Pediatrics' California Chapters 1, 2, 3, and 4 and the AAP.

"With incontinentia pigmenti you'll have a staged eruption, with vesicles or pustules at the time of birth within the first weeks, followed by more warty or verrucous papules," said Dr. Levy, professor of dermatology and pediatrics at Baylor College of Medicine, Houston.

By the first year of life a swirling gray-brown hyperpigmentation develops. "This is due to incontinence of pigment," he explained. "Melanin has dropped out of the basal layer of epidermis into the upper dermis." The condition occurs primarily in females and is marked by staged cutaneous eruptions.

"You should regard it as a potentially neurocutaneous syndrome," said Dr. Levy, also chief of the dermatology service at Texas Children's Hospital, Houston. "Eosinophilia can be seen during stage 1, which is the vesicular stage."

The gene for incontinentia pigmenti has been mapped to Xq28. Deletions at this site cause mutations in NF-kB essential modulator (NEMO), which governs a variety of inflammatory and immune responses. These mutations can lead to skin lesions, vascular anomalies, and immune dysfunction.

Dr. Levy noted that 80% of incontinentia pigmenti cases are caused by mutations in NEMO. Management of the skin lesions is generally supportive. Careful ophthalmologic and neurologic follow-up is indicated in some cases.

At the meeting, Dr. Levy also discussed the clinical signs of another skin defect that can affect newborns: aplasia cutis congenita. This condition is marked by focal congenital skin defects that most often appear on the scalp or face. It also can occur along the temporal area, a version known as focal facial dermal hypoplasia.

Aplasia cutis congenita is "rarely on the trunk or extremities, though this can happen anywhere," he said. "The etiologies are really varied and may include vascular insufficiency, intrauterine infections, and amniotic membranes. On the scalp, this might be due to defects of closure."

In your work-up, consider these malformation syndromes that may be associated with aplasia cutis congenita: Opitz syndrome, Adams-Oliver syndrome, Trisomy 13-15 syndrome, 4p syndrome, Johanson-Blizzard syndrome, and Xp22 microdeletion.

Some cases of aplasia cutis congenita will have a so-called hair collar sign, which is a ring of dark hair that often surrounds a scalp lesion. The hair collar sign "suggests cranial dysraphism, especially with overlying vascular malformation." ■