Consultant360 Multidisciplinary Medical Information Network

PHOTOCLINIC

PEER REVIEWED

Incontinentia Pigmenti In a Neonate

Authors:
A. Hope Tobey, DO, FAAP, FACOP
Edward Via College of Osteopathic Medicine, Blacksburg, Virginia

Citation:
Tobey AH. Incontinentia pigmenti in a neonate [publisheded online October 26, 2017]. *Consultant for Pediatricians*.

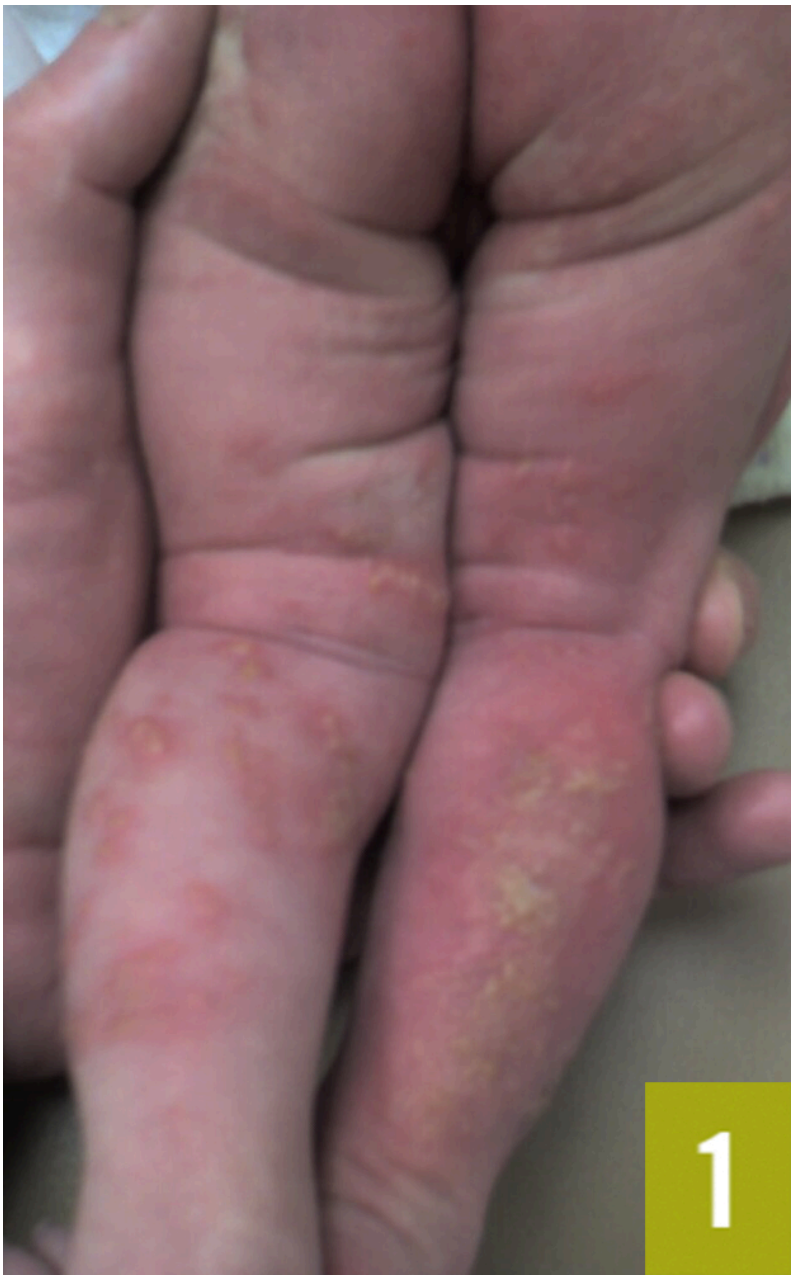
A full-term 10-day-old girl presented to a primary care pediatrics office for a rash that had been present for 7 days. The rash had started on her arms and legs and had spread to her trunk. The number of lesions and intensity of erythema had increased.

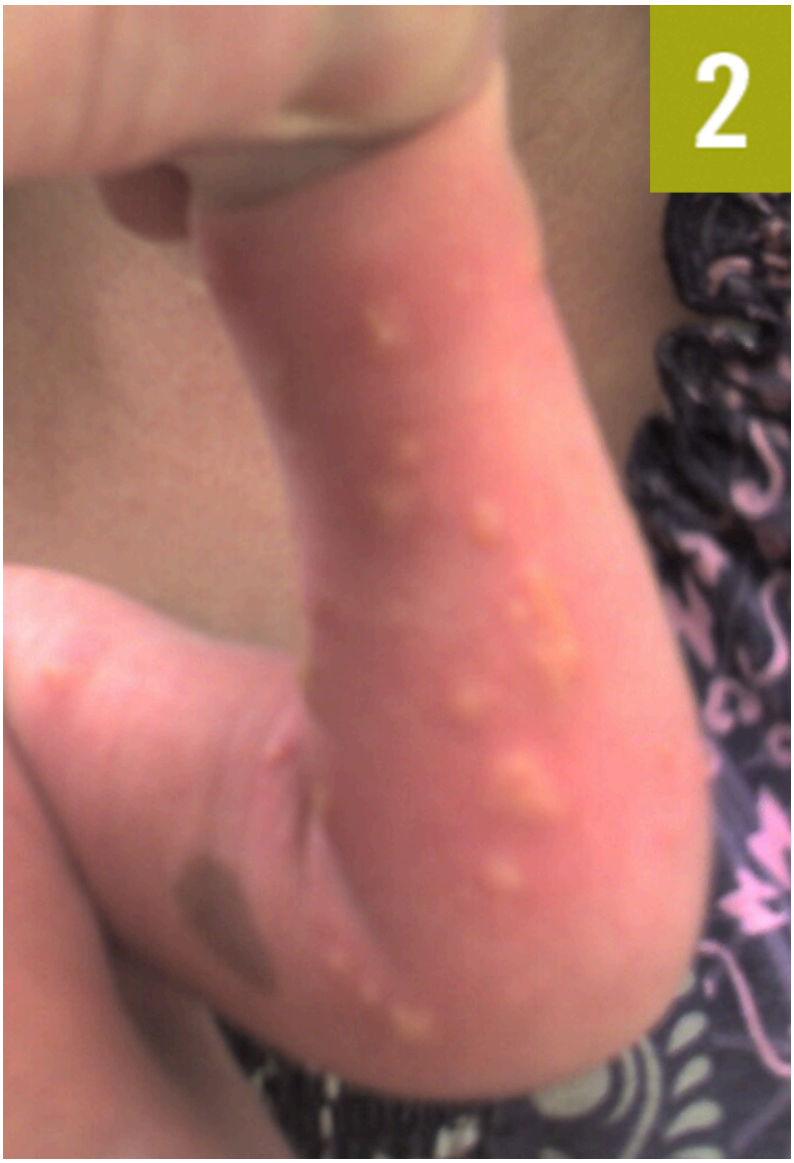
Physical examination. Physical examination demonstrated erythematous patches with multiple vesicular lesions, primarily over the arms and legs, with a few scattered over the trunk (**Figures 1-3**). The face was spared. One of the vesicles was unroofed, and the contents were sent for viral and bacterial cultures. The patient was initially treated with supportive care, and a topical antibiotic cream was applied to the areas with crusting.

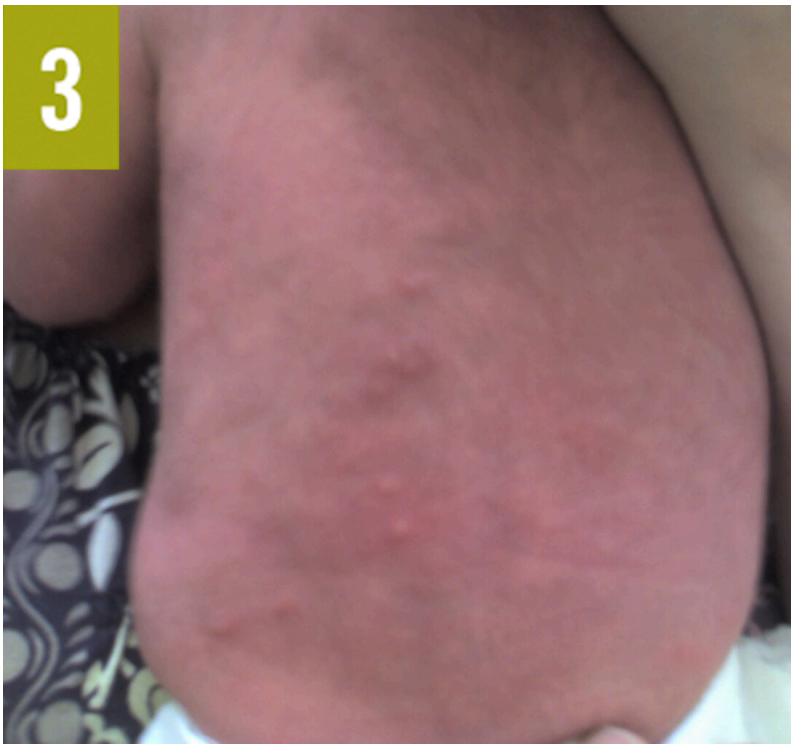
Diagnostic tests. The viral and bacterial cultures were negative. The girl was referred to a dermatologist, who repeated viral and bacterial cultures, the results of which were again negative. With a working diagnosis of bullous impetigo, the dermatologist continued the topical antibiotic and started a 10-day course of oral cephalexin.

There appeared to be initial improvement; however, within a week the rash flared again with the same vesicular lesions. The dermatologist then performed a skin biopsy, the findings of which were consistent with a diagnosis of incontinentia pigmenti (IP) syndrome.

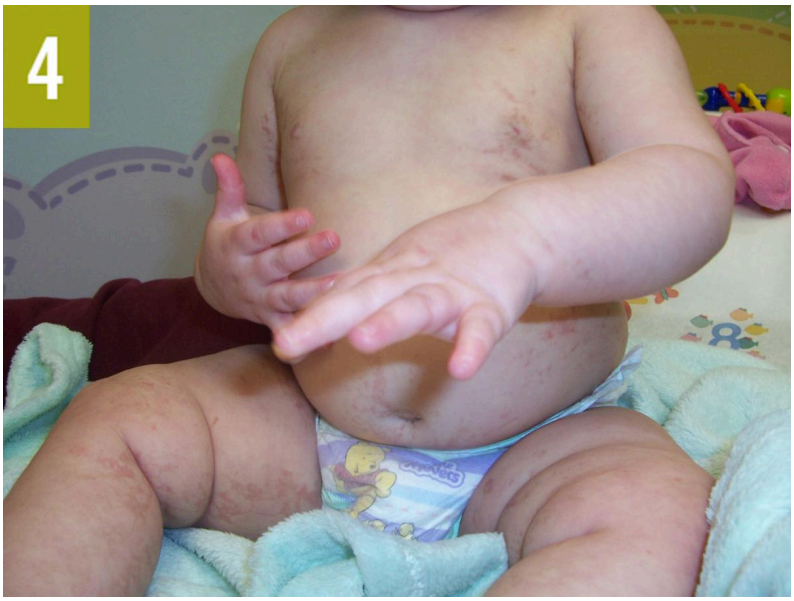
Over the subsequent months, her rash progressed following the typical 4 stages of IP. During stage 1, the vesicular lesions (**Figures 1-3**) seemed to overlap with a brief appearance of the warty lesions that occur in stage 2.




2


3

By age 7 months, the patient had developed the classic hyperpigmented swirling lesions of IP (**Figure 4**). She had yet to develop the fourth stage, which is characterized by linear hypopigmentation and is typically seen in adulthood.



Discussion. IP can be diagnosed clinically based on the classic rash stages.[1] However, the diagnosis is typically confirmed by molecular testing results, which demonstrate a heterozygous pathologic variant in *IKBKG* (formerly *NEMO*) on Xq28.[2] If a pathologic variant is not identified prior to the appearance of the classic swirls of hyperpigmentation, biopsy can be helpful in diagnosis. Histologic findings consistent with IP include eosinophilic infiltration and/or extracellular melanin granules.[1]

IP can also be associated with a defect in tooth formation, alopecia, dystrophic nails, neovascularization of the retina, cognitive delays, seizures, and central nervous system malformations.[3,4] These complications warrant referral to and evaluation by a pediatric neurologist, an ophthalmologist, and a dentist, as well as a pediatric dermatologist and a geneticist.

Treatment of the cutaneous manifestations is largely supportive, because there is no cure, and the lesions typically improve or resolve over time.[4] Makeup is often sufficient to hide residual cutaneous markings. Other possible sequelae should be followed up on and appropriately managed lifelong. Since it is an X-linked dominant disorder, genetic counseling is also warranted, because patients have a 50% chance of transmitting the mutation and have an increased risk for spontaneous abortion of male fetuses.[2]

Patient outcome. After a thorough multidisciplinary evaluation, the patient was found to have a mild form of IP. At the time of this writing, the patient remained healthy with cutaneous findings and no evidence of other abnormalities.

References:

1. Minić S, Trpinac D, Obradović M. Incontinentia pigmenti diagnostic criteria update. *Clin Genet*. 2014;85(6):536-542.

2. Smahi A, Courtois G, Vabres P, et al; International Incontinentia Pigmenti (IP) Consortium. Genomic rearrangement in *NEMO* impairs NF-κB activation and is a cause of incontinentia pigmenti. *Nature*. 2000;405(6785):466-472.

3. Poziomczyk CS, Bonamigo RR, Santa Maria FD, Zen PRG, Kiszewski AE. Clinical study of 20 patients with incontinentia pigmenti. Int J Dermatol. 2016;55(2):e87-e93.

4. Landy SJ, Donnai D. Incontinentia pigmenti (Bloch-Sulzberger syndrome). *J Med Genet*. 1993;30(1):53-59.