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Five Day Old Girl with Rash

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Abstract

A 5 day old girl presented with a 2 day history of rash. Red lesions were noted by her mother on her right leg. On the day of presentation, the patient was seen by her primary care physician for a checkup and was sent to the emergency department for evaluation of the rash. She was otherwise well.

Keywords: Rash; Dermatology

Introduction

She was a term infant born via c-section due to failure to progress. Pregnancy was uncomplicated. The mother was group B strep and serology negative and had no history of oral or genital herpes simplex virus (HSV).

Vital signs were normal. Except for the skin, her exam was unremarkable. Her right leg had an erythematous, blanching, vesiculopapular rash on the lateral aspect of the calf. The medial aspect of the right thigh had erythematous papules arranged in a linear plaque. The rest of her skin was clear (Figure 1).

Incontinentia Pigmenti

Based on the appearance of the rash, a presumptive diagnosis of incontinentia pigmenti (IP) was made. A direct fluorescent antibody (DFA) test for HSV was sent as well as a punch biopsy. The DFA was negative and the biopsy confirmed IP.

Epidemiology and Pathophysiology

IP is a rare, genodermatosis that has X-linked dominant transmission. The incidence is estimated at 1 in 50,000. The aberrant gene appears to encode for nuclear factor κB essential modulator (NEMO) located on chromosome Xq28. NEMO is a transcription factor that regulates the expression of genes controlling immune response and protection against apoptosis induced by tumor necrosis factor. The cells of afflicted males are more prone to apoptosis and this probably leads to the spontaneous abortion of most affected males. In females, IP is usually a benign disease.

Diagnosis

Dermatologic findings are the hallmark of the disease and its diagnosis. There are 4 stages which sometimes occur simultaneously:

Stage 1 (Onset during first 2 weeks of life): Heralded by vesicles and papules on an erythematous base distributed in a linear pattern that can resemble HSV. This generally resolves by 4 months.

Stage 2 (Onset at age 2 to 6 months): The vesicles from stage 1 become verrucous. These lesions usually disappear by age 6 months.

Stage 3 (Onset at age 3 to 6 months): This stage is characterized by hyperpigmented whorls that follow the lines of Blaschko. It usually resolves during the second decade.

Stage 4 (Onset after stage 3): This stage is characterized by permanent, hairless and atrophic patches or areas of hypopigmentation.

Clinical diagnostic criteria have been proposed. When there is a definitive family history, presence of one major criterion strongly suggests the diagnosis of IP. When there is no family history, at least one major criterion should be present. Minor criteria support the diagnosis (Table 1).

Family History	Major Criteria	Minor Criteria
No evidence of IP in a 1st degree female relative	Typical neonatal rash Typical hyperpigmentation on trunk following lines of Blaschko Linear, atrophic, hairless lesions	Dental anomalies
Evidence of IP in a 1st degree female relative	Typical rash Vertex alopecia Dental anomalies Retinal disease Multiple male miscarriages	-

Table: Dermatological findings and diagnosis of the disease

Differential Diagnosis

Because IP manifests itself in multiple ways, the differential diagnosis varies by stage. Most importantly, the vesicular lesions that characterize stage 1 are of greatest concern when faced with an infant that is only days or weeks old.

Stage 1: Herpes simplex virus, varicella zoster virus, bullous impetigo, Langerhans cell histiocytosis, epidermolysis bullosa, and immune-mediated bullous disease.

Stage 2: Verruca vulgaris, linear epidermal nevus, and lichen striatus.

Stage 3: Linear and whorled nevoid hypermelanosis and pigment mosaicism.

Stage 4: Tinea versicolor, vitiligo, and postinflammatory hypopigmentation.

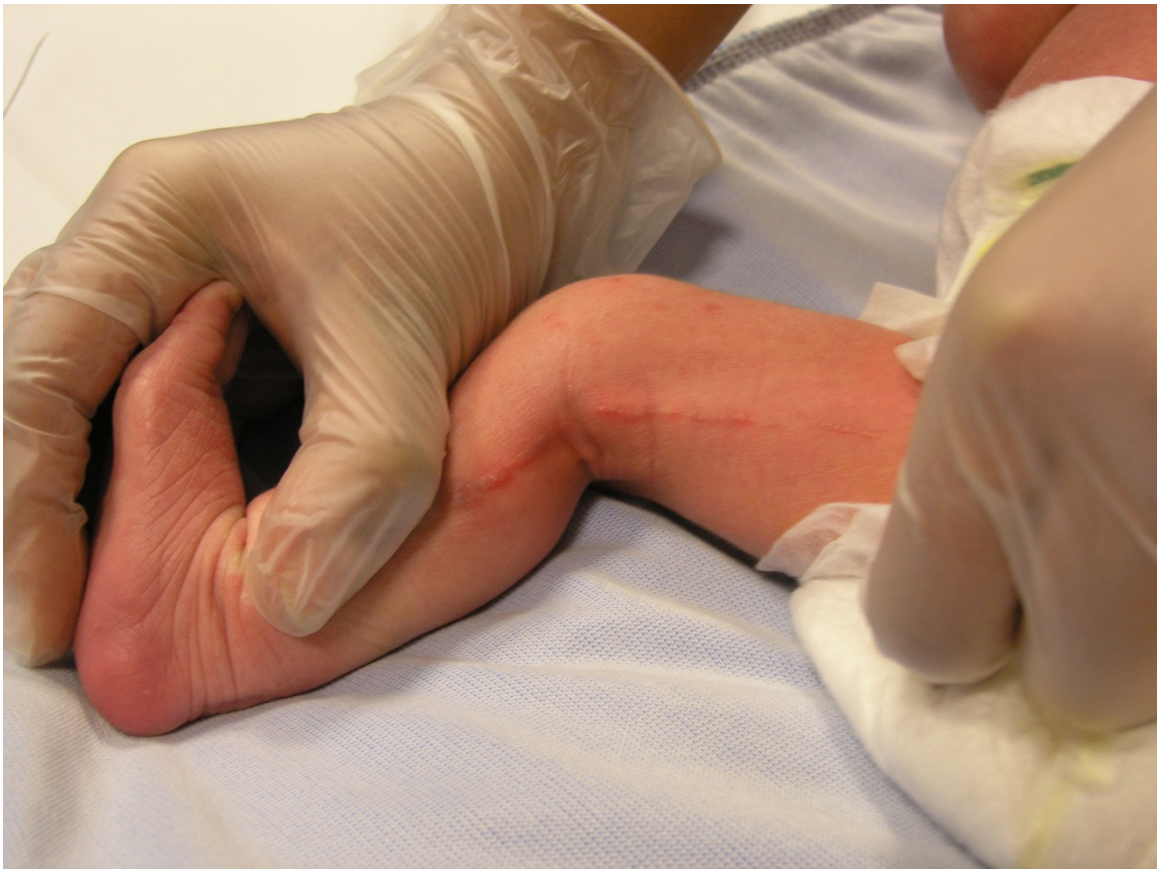

Figure 1: A 5 day old girl presented with a 2 day history of rash.

Non-Dermatologic Manifestations

IP can be a multisystem disease with variable involvement of different organs. So while the skin findings are the keys to diagnosis, it is the presence of other problems that guide management.

Asymmetric ophthalmic problems occur in up to 35% of patients. Findings can include strabismus and optic nerve atrophy. Retinal involvement can lead to detachment. Additionally, there can be an abnormal proliferation of retinal vasculature that is similar to the retinopathy of prematurity. In rare instances, retinal lesions can lead to blindness in the affected eye.

Neurological sequelae occur in roughly 30% of patients and can include seizures and spastic paralysis, and microcephaly. Children with IP and early seizures often have poor neurodevelopmental outcomes. These children require neurology consultation.

Other abnormalities may occur in the teeth, hair, and nails. Abnormalities of the dentition are the most common non-dermatologic manifestations of IP, seen in about 80% of patients. Affected children can have absent teeth, conical teeth, or delayed eruption. All except the latter are lifelong anomalies.

Defects in the nails and hair are also frequent components of IP. Mild vertex alopecia is seen in about 40% of patients. In the nails, there may be pitting or ridging. Subungual and periungual keratotic tumors may appear during the second decade. These lesions can cause lytic destruction of underlying bone.

Management

The acute management of IP, particularly during stage 1, is making a definitive diagnosis. HSV and other infections should be excluded. This can be done on clinical grounds or with cultures. Consultation with a pediatric dermatologist is often helpful. The acute management of IP, particularly in the neonate during stage 1, centers on coming to a definitive diagnosis. Care should be taken to exclude HSV and other infectious causes of vesicular rash. This can be done either on clinical grounds, or with appropriate cultures. Consultation with a pediatric dermatologist is often helpful.

Patients who present with seizure should be managed as with any other infant with seizure. Attention to airway, breathing, and circulation is paramount. Benzodiazapines and fosphenytoin can be used to control the seizures. Strong consideration should be given to empiric treatment with acyclovir for HSV in a seizing infant with vesicular lesions until a definitive diagnosis is made.

In an infant with an otherwise normal physical exam, a formal ophthalmologic examination is all that is necessary in the short term.

This initial exam should take place soon after diagnosis because therapy for abnormal retinal vascular development can be vision preserving. The patient should be followed regularly by ophthalmology until age 3 years. If no problems arise by then, the prognosis for normal vision is excellent.

Seizures or other abnormal neurologic findings in a neonate with IP warrant head imaging with computed tomography or magnetic resonance imaging. Additionally, the brains of patients with retinal involvement should also be imaged. Often seen are focal areas of damage to the corpus collosum, cerebellum, or other parts of the brain. Notably, patients with normal neurologic and ocular findings do not require any imaging.

Older children may present with subungual or preiungula keratotic tumors. As discussed previously, these tumors can lead to lytic lesions in the underlying bone. Because of this, the current standard is to surgically excise the growths.