Two Cases of Incontinentia Pigmenti Simulating Child Abuse

ABSTRACT. In the United States 1.4 million children were maltreated in 1988, resulting in an estimated 2000 to 5000 deaths.1 Largely due to the rising awareness and sensitivity to the horrors of child abuse, the number of deaths declined to approximately 1500 in 1993.2 Guidelines have been published to aid in the identification and management of child maltreatment,3 and reporting of all suspicious cases is mandated by law. In our zealous efforts to protect children, some families are investigated because of misdiagnosed abnormalities, often cutaneous,4 leading to the unintentional injury of both patients and their families.5

In this report, we describe two patients with cutaneous and/or visceral manifestations of incontinentia pigmenti (IP) who were initially thought to be victims of child abuse. Pediatrics 1997;100(4). URL: http://www.pediatrics.org/cgi/content/full/100/4/6; incontinentia pigmenti, child abuse.

ABBREVIATIONS. IP, incontinentia pigmenti; WBC, white blood cells; RBC, red blood cells; Hb, hemoglobin; PT, prothrombin time; PTT, partial thromboplastin time; CT, computed tomography.

CASE REPORTS

Case 1

The patient is a 6-day-old girl transferred from an outside hospital for seizures. She was born at 41 weeks gestation by spontaneous vaginal delivery (birth weight 7 pounds), to an 18-year-old primiparous mother who denied chlamydial, syphilitic, or gonorrheal infection, or substance abuse (alcohol, drugs, tobacco). There was no history of premature rupture of membranes or maternal fever. The delivery was complicated by thin meconium requiring oropharyngeal suctioning without intubation. Vitamin K was given intramuscularly. The infant was discharged to home at 24 hours of life.

On the second day of life, the primary care takers (mother and maternal grandmother) noted seizure activity, described as eye deviation to the right, left upper extremity flexion with adduction, and right upper extremity extension with hypertonicity. These episodes lasted approximately 30 seconds each, occurred three times per day and were associated with cyanosis. The patient was noted to have decreased oral intake and three loose stools on day 3 of life. There was no vomiting or fever reported and no history of trauma or medications. The family history was notable for a seizure in a maternal aunt; no history of sickle cell, bleeding or clotting diseases existed. The maternal grandmother was involved with the Department of Child and Family Services, which handles child abuse, when the patient’s mother was 6 years old and again at the age of 10 years. Both cases were unfounded and dismissed. The patient was seen by a visiting nurse who advised that the infant was a “kicker.” After discussing the case with the Department of Child and Family Services, the nurse referred the infant for further investigation. The state Department of Child and Youth Services was contacted for further investigation. The state Department of Child and Youth Services was notified for consultation. The family brought the infant to the emergency department for a second opinion and further evaluation. The family history revealed that the patient’s mother had two prior miscarriages and the maternal grandmother had three miscarriages as well as three healthy daughters. The mother denied having any dermatologic

upper extremities) and ecchymoses over the buttocks and the lumbar spine (Fig 1 and Fig 2). The heart, lung, and abdominal examinations were normal. The evaluation included a lumbar puncture (1 white blood cell [WBC]/mm3, 12 red blood cell [RBC]/mm3; glucose 86 mg/dL, protein 89 mg/dL), a complete blood cell count (WBC 15 700 k/microL; hemoglobin [Hb] 17.5 g/dL; platelets 87 000 k/microL), coagulation studies (prothrombin time [PT] 12.5 seconds, partial thromboplastin time [PTT] 22.7 seconds) and a computed tomography (CT) brain scan (diffuse cerebral infarcts and edema with sparing of the basal ganglia, thalamus, cerebellum and brainstem) (Fig 3). The patient was given phenobarbital, ampicillin, and ceftriaxone before transfer to a pediatric medical facility. The primary diagnosis was shaken baby syndrome.

On arrival to our hospital, the infant was stabilized in the emergency department and admitted to the intensive care unit. Further anticonvulsant therapy as well as endotracheal intubation was required. Neurosurgical and social work evaluations were instituted immediately. Ophthalmological consultation revealed retinal and visceral manifestations of incontinentia pigmenti. The initial assessment of the emergency medicine and intensive care physicians was that of nonaccidental injury. A report was filed with the Department of Child and Youth Services. A state investigator for the Division of Child Protection interviewed the family and examined the child 24 hours after the patient’s admission to the hospital. The police were notified shortly thereafter and photographed the infant’s dermatologic findings.

The diagnosis became clearer with pediatric dermatologic consultation that recognized the ecchymoses to be hyperpigmented swirls that followed the lines of Blaschko. Within 24 hours, vesicles appeared with pitting along Blaschko’s lines. Further probing of the family history revealed the maternal grandmother had multiple miscarriages; the maternal grandmother and the maternal great grandmother had early-onset cataracts; the maternal grandmother had retinal detachment; and the mother and the maternal aunts had similar skin findings as children (one maternal aunt has persistent skin lesions as an adult). The family history in conjunction with the neurologic, ophthalmologic, and especially the dermatologic findings pointed to the diagnosis of the X-linked dominant genetic disorder IP. Skin biopsy confirmed the diagnosis.

Case 2

The second patient is a 1-month-old Hispanic girl who was brought to the emergency department by her parents because of a worsening skin rash. The neonate was an 8 lb, 3 oz product of a full-term gestation to a 32-year-old woman 6 days after an uncomplicated pregnancy. She was born via cesarean section because of a nuchal cord. There were no problems in the nursery and she went home with her mother. At approximately 2 weeks of age the patient developed vesicular lesions on her back and arms that crusted over shortly thereafter (Fig 4). The patient’s pediatrician referred the infant to a dermatologist who made the diagnosis of impetigo. New skin lesions developed in addition to the impetiginous ones over the patient’s third week of life. During a visit with her pediatrician at 24 days of life, hyperpigmented linear lesions were noted on the patient’s trunk and faintly on the extremities (Fig 5). Poor weight gain was documented (weight 25th percentile, length 50th percentile). The hair, (limited) ophthalmologic, and neurologic examinations were normal. Nonaccidental injury and neglect were suspected and a social worker was notified for consultation. The state Department of Child and Youth Services was contacted for further investigation.

The next day additional hyperpigmented linear lesions were noted by the mother on the infant’s trunk and extremities. The family brought the infant to the emergency department for a second opinion and further evaluation. The family history revealed that the patient’s mother had two prior miscarriages and the maternal grandmother had three miscarriages as well as three healthy daughters. The mother denied having any dermatologic
disorders as a child, though on examination she did have several barely visible areas of decreased pigmentation in linear streaks on the back of her legs. Based primarily on the dermatologic findings, the clinical diagnosis of IP was made by the pediatric emergency physician and confirmed by a pediatric dermatologist. Future evaluations with neurology, ophthalmology, and genetics were arranged; social services was made aware of the diagnosis.

**DISCUSSION**

Suspected nonaccidental injury must be reported to the appropriate authorities. Misdiagnosed cases of child abuse also deserve reporting to prevent recurrent misinterpretation by others. Many examples of cutaneous disorders that were misdiagnosed as a result of suspicious findings have been published.8–16 These are the first published case reports of IP as a potential masquerader of child abuse.

IP is a rare genodermatosis. It is a multisystem, neuroectodermal disorder characterized by dermatologic, dental and, in a minority of patients, ocular and neurologic abnormalities. The name IP describes the characteristic, although nonspecific, histological finding of incontinence of melanin in the superficial dermis.17

The cutaneous manifestations of IP are diagnostic. Although four stages have been described, all stages do not necessarily occur and several stages may overlap.17 The lesions of the first stage, collections of linear vesicles overlying erythema, usually develop within the first 6 weeks of life. This initial inflammatory phase is often accompanied by a marked peripheral blood leukocytosis with eosinophilia.18 These lesions can be mistaken for bullous impetigo, herpes simplex, epidermolysis bullosa, dermatitis herpetiformis, or even second degree burn injury.19 Biopsy sections of lesional skin demonstrate intraepidermal pustules of eosinophils, allowing the diagnosis of IP to be confirmed. By the first few months the second phase is seen, with verrucous plaques, often in a linear configuration.

The lesions of stage 3 are considered the hallmark of IP. The hyperpigmentation can be very localized or extensive, but presents as streaks on the extremities or whorls on the trunk. These pigmented lesions remain static for several years until they fade during childhood or adolescence.19 Some patients have localized areas of persistent pigmentation. In other patients, flares of the vesiculopustular or even the verrucous lesions occur.

The fourth phase of hypopigmented and/or atrophic streaks occurs in 14% and 28% of patients respectively, and may persist into adulthood. Approximately 30% of patients have cicatricial alopecia, which may be the only persistent sign in adult women.18

All of the cutaneous manifestations show pat-
tering along Blaschko’s lines, paths of ectodermal cell migration during embryologic development of the skin. This X-linked dominant disorder is generally lethal for affected boys who do not have a normal X chromosome. However, functional mosaicism occurs in affected girls because of random inactivation of the X chromosome at 12 to 16 days gestation. Expression of the IP as streaks occurs with activation of the mutant gene. Within the spectrum of IP are girls with minimal involvement and others with extensive involvement, as in both of our patients.

Central nervous system manifestations probably require fairly extensive activation of the mutant gene or disturbance of critical brain regions. Seizures, as seen in case 1, are the most common disturbance and have been described in approximately 13% of patients. The CT scan findings of the brain of patient 1 are consistent with the expected neuropathologic findings of hemorrhagic white matter encephalopathy with massive edema. Atrophy eventually develops.

Ocular anomalies occur in one third of IP patients, particularly strabismus and cataracts. (Patient 2 was found to have a moderate left eye esotropia on ophthalmologic follow-up examination at 6 months of age.) Retinal vascular changes, as evidenced in our first patient with hemorrhages and cotton wool spots, are the most frequently reported intraocular abnormalities, and can lead to blindness. Pseudoglioma, a fibrovascular retrolental mass, can evolve to retinal detachment, as in the maternal grandmother of patient 1. This mechanism is thought to be analogous to retinopathy of prematurity.

These two cases stress the importance of disease recognition by pediatric specialists, and of a thorough family and social history. In our first case, the maternal grandmother’s previous involvement with the Department of Child and Youth Services...

Fig 2. The back and buttock region are covered with swirls and streaks of hyperpigmentation and purple discoloration with patches of vesicles, all along the lines of Blaschko.
was considered to be evidence in favor of nonaccidental injury. Victims of child maltreatment are more likely to become abusive parents. Further exploration provided pivotal information against nonaccidental injury, in that it was the mother’s characteristic IP skin lesions that had twice (at age 6 and age 10 years) been misinterpreted as possible intentional injury.

IP is rare and is frequently recognized only by pediatric specialists. This illness is vulnerable to misdiagnosis given that the cutaneous findings alone can mimic traumatic injuries. Herpes simplex is the most common misdiagnosis in the neonate with blisters and seizures. The additional findings in IP of hyperpigmented skin streaks and hemorrhagic manifestations of the eyes and brain easily lead one to consider child abuse. IP should be included in the list of childhood diseases that can be misinterpreted as child maltreatment.

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Fig 3. Diffuse cerebral infarcts and edema.

Fig 4. Forearm vesicles with overlying granulation tissue.
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Fig 5. Streaks of hyperpigmentation on the chest and proximal right arm.
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