CASE REPORT

THE PATIENT

Newborn African American girl

SIGNS & SYMPTOMS

– Depigmented patches covering approximately 15% of newborn's body, surrounded by areas of thickened skin

– Mild scaling and hyperpigmentation

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The authors reported no potential conflict of interest relevant to this article.

THE CASE

A 21-year-old G3P2 mother gave birth to an African American girl via vaginal delivery. Labor had been induced due to gestational hypertension at term. She'd also had a stillborn at term at the age of 16 followed by a second live term birth 3 years ago. During this most recent pregnancy, she'd received adequate prenatal care and had been treated for chlamydia with a single dose of oral azithromycin 1 g.

The newborn had an Apgar score of 9 out of 9 and weighed 6.7 pounds at birth. During a physical examination in the nursery, the infant was found to have large areas of smooth depigmentation on her forehead, right forearm, lower abdomen, and left thigh, with surrounding areas of thickened skin that had mild scaling and hyperpigmentation (**FIGURE**). The depigmented areas involved approximately 15% of the newborn's body. The father and paternal grandmother, who were present at the time of delivery, also had depigmented areas of their skin.

The newborn's tongue was pink and her mucus membranes were moist. No macules or patches were noted on either the oral or vaginal mucosa. Cardiac, pulmonary, and ocular examinations (including evaluation of the retina by ophthalmoscopy) were normal. There was no nystagmus or strabismus. The newborn's extremities were normal, symmetric, and moveable, and she was easily consoled.

THE DIAGNOSIS

We diagnosed the newborn with piebaldism based on her appearance. Piebaldism consists of hypopigmented/depigmented areas and is a clinical diagnosis; no testing is required.

Concerned about the areas of hyperpigmentation, we decided to get a dermatologist's opinion. The dermatology team briefly considered the diagnosis of a large melanocytic nevus with sparing of some areas, but a skin biopsy of a hyperpigmented area on the left leg came back with a normal number of melanocytes.

DISCUSSION

Piebaldism is a rare autosomal dominant disorder characterized by the congenital absence of melanocytes in affected areas of the skin and hair due to mutations of the c-kit gene. The c-kit gene affects the differentiation and migration of melanoblasts from the neural crest during embryonic life.[1] The incidence of piebaldism is estimated to be less than one in 20,000.[2] Both males and females are equally affected, and no race is spared.[2,3]

Affected individuals present with a white forelock and relatively stable, persistent depigmentation of skin with a characteristic distribution from birth.[3] A white forelock arising from a triangular, elongated, or diamond-shaped midline or depigmented macule

FIGURE

Large areas of depigmented patches covering 15% of newborn's body

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on the forehead may be the only manifestation in 80% to 90% of cases.[3] The characteristic distribution of depigmented macules includes a central macule on the forehead, the anterior abdomen extending to the chest, the lateral trunk sparing the dorsal spine, and the mid-arms and legs sparing the hands and feet.[2]

Depigmented macules are rectangular, rhomboid, or irregular in shape and usually have a symmetrical distribution. Typically, islands of hyperpigmentation are present within and at the border of depigmented areas.[4] Piebaldism is associated in rare instances with neurofibromatosis type 1, Hirschsprung's disease, hearing loss, and Waardenburg syndrome.[4]

Histologically, melanocytes are absent or considerably reduced in the depigmented areas and are normal in number in the hyperpigmented areas.[5]

The differential diagnosis of piebaldism includes mosaicism, albinism, and vitiligo.

Cutaneous mosaicism stems from a gene mutation that occurs during embryogenesis and the lesions are distributed along certain patterns and forms. A chromosomal analysis of our patient showed a normal female karyotype that excluded mosaicism.

Albinism is a genetically inherited disorder characterized by partial or complete absence of melanin production in the skin, hair, and eyes.[6] Eye and fundus examinations were normal in our patient, which excluded albinism.

Vitiligo is rarely present at birth but is usually acquired later in life. It results from an immune-mediated destruction of melanocytes and is not genetically inherited, although familial incidence has been reported.[7]

There are no effective therapies

A combination of dermabrasion and grafting of pigmented skin into depigmented areas, with or without phototherapy, has been used in select patients, although no solid data are available on its effectiveness.[3] The lack of effective and safe therapies can make treatment challenging. Piebaldism is usually not medically harmful, but the emotional and psychological effects on the family and the patient as they grow up can be devastating. Therefore, supportive counseling is recommended.

Our patient. Supportive counseling and a follow-up appointment with a dermatologist was planned for our patient and her family.

THE TAKEAWAY

The clinical diagnosis of piebaldism is straightforward based on the presence of a white forelock in the frontal region, the

appearance of depigmented macules since birth that stay relatively stable, and the presence of a similar pattern of depigmented macules in other family members. Histologic or genetic testing is not necessary to establish the diagnosis. Rarely, cases of piebaldism are associated with hearing loss, necessitating a hearing assessment and an audiology exam. Unfortunately, there are no effective treatments for piebaldism. JFP

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