Depigmented patches in a child

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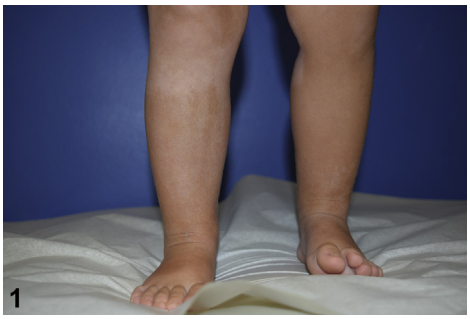

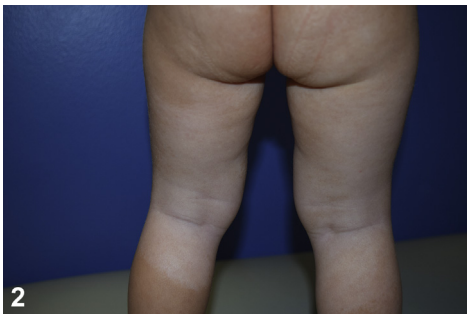

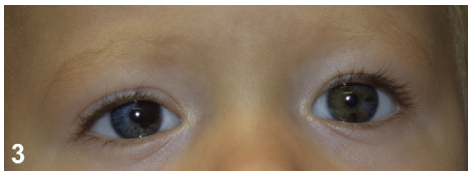

A two-year-old girl presented for evaluation of asymptomatic congenital white patches in addition to new white patches that had appeared over the past few months. She had bilateral congenital sensorineural hearing loss and mild gross motor delays. Family history was negative for similar lesions. Physical examination demonstrated depigmented patches on the lower extremities with poorly circumscribed, feathered edges (Figs 1 and 2). Eye examination was significant for heterochromia iridis and normal placement of the inner canthi (Fig 3). There were no other abnormalities on examination.

Question 1: What is the most likely diagnosis?

A. Vitiligo

B. Piebaldism

C. Pigmentary mosaicism

D. Waardenburg syndrome

E. Tuberous sclerosis complex

Answers:

A. Vitiligo — Incorrect. Although vitiligo presents with depigmented lesions, they are typically not present at birth and are acquired later in life.[1] The existence of true congenital vitiligo is controversial.

B. Piebaldism — Incorrect. Although piebaldism may present with depigmented patches in infants and young children, auditory and facial developmental anomalies, such as heterochromia iridis, are absent in piebaldism.[1,2]

C. Pigmentary mosaicism — Incorrect. Pigmentary mosaicism encompasses a heterogenous group of conditions characterized by varied patterns of hypopigmented or hyperpigmented lesions.

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D. Waardenburg syndrome — Correct. The diagnosis of Waardenburg syndrome (WS) was made based on the Waardenburg Consortium criteria, which requires 2 major criteria or 1 major and 2 minor criteria.[1,3] The patient met 2 major criteria (congenital sensorineural deafness, pigmentary abnormalities of the iris). In addition, she met one minor criteria (depigmented macules or patches).

E. Tuberous sclerosis complex — Incorrect. Tuberous sclerosis presents with hypopigmented, rather than depigmented, macules in addition to other cutaneous abnormalities as well as neurologic, cardiac, renal, and pulmonary disease.[3]

Question 2: Which variant of this condition does the patient mostly likely have?

A. Type 1

B. Type 2

C. Type 3

D. Type 4

E. Type 5

Answers:

A. Type 1 — Incorrect. Type 1 WS is the most common type of WS and is diagnosed clinically using the Waardenburg Consortium criteria.[1,3] Type I WS is distinguished from type 2 WS by the presence of dystopia canthorum. The patient did not have lateral displacement of the inner canthi.

B. Type 2 — Correct. Type 2 WS is similar to Type 1 WS, but the inner canthi are normal.[1,3] This was consistent with the patient's presentation.

C. Type 3 — Incorrect. Type 3 WS, also known as Klein-Waardenburg syndrome, is characterized by musculoskeletal abnormalities.[1,3] The patient had an otherwise unremarkable physical examination and no skeletal abnormalities.

D. Type 4 — Incorrect. Type 4 WS, also called Shah-Waardenberg syndrome, has features similar to type 2 WS but is also associated with Hirschsprung disease or congenital aganglionic megacolon.[1,3] The patient had a negative review of systems, with no history of abdominal distention, emesis or constipation, making type 4 WS less likely.

E. Type 5 — Incorrect. Only 4 types of WS have been described.[1]

Question 3: Which of the following statements is true regarding this diagnosis?

A. It is more common in Caucasian children

B. Visual acuity is commonly affected

C. It is responsible for 2-5% of cases of congenital deafness

D. Skin depigmentation is the most common cutaneous manifestation

E. Mutations in PAX3 are the only known cause of WS

Answers:

A. It is more common in Caucasian children — Incorrect. WS affects all races equally.[1]

B. Visual acuity is commonly affected — Incorrect. WS presents with a broad spectrum of choroidal and iris hypopigmentation, but visual acuity is typically preserved.[4]

C. It is responsible for 2-5% of all cases of congenital deafness — Correct. WS accounts for 2-5% of congenital deafness and affects approximately half of all patients with WS.[3] Hearing loss may be unilateral or bilateral and vary in severity.[2]

D. Skin depigmentation is the most common cutaneous manifestation — Incorrect. A white forelock is the most common cutaneous manifestation of WS and seen in approximately 45% of individuals, while skin depigmentation occurs in 30%.[1,3] Approximately 80-90% of individuals with piebaldism also have a white forelock.[3]

E. Mutations in PAX3 are the only known cause of WS — Incorrect. At least 8 genes, important in melanocyte development, have been associated with WS and contribute to the heterogeneity of the phenotypic presentation.[1] Genetic testing of the patient revealed a MITF (microphthalmia-associated transcription factor) mutation, confirming WS. Parental genetic testing for the MITF mutation was negative, supporting a *de novo* mutation in the patient.

Abbreviations used:

MITF: microphthalmia-associated transcription factor
WS: waardenburg syndrome

Conflicts of interest

None declared.

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