Newborn With Oculocutaneous Albinism

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KEY WORDS
Newborn, oculocutaneous albinism

CHIEF COMPLAINT

A term African American girl with light blond hair, very light skin, and blue eyes was admitted to the normal newborn nursery (Figure).

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HISTORY OF PRESENT ILLNESS

The 28-year-old mother of the newborn girl reported an uneventful pregnancy. She denied a history of chlamydia, gonorrhea, syphilis, human immunodeficiency virus, or environmental or drug exposures. Her medical history was unremarkable, and findings of a review of systems were negative.

MEDICAL AND OBSTETRICAL HISTORY

The mother's medical history was benign except for excision of a gangrenous lesion from her right hand. Her obstetrical history was not significant for any previous genetic disorders. She was identified as gravida 4, para 1. Her early prenatal care was initiated in West Africa, second trimester care was provided in an eastern U.S. city, and care beginning at 35 weeks' gestation was provided at a community health clinic in a major southwestern metropolitan city.

LABOR AND DELIVERY

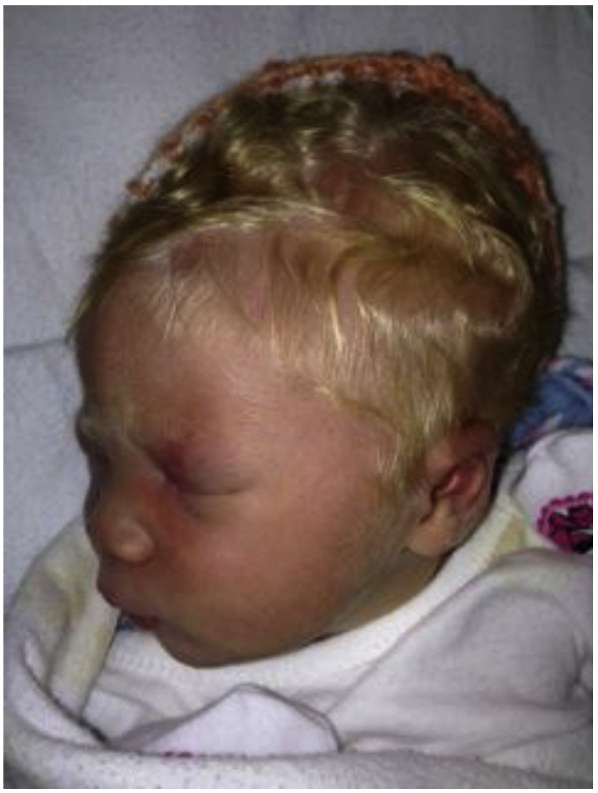
Labor was induced at 40 weeks' gestation because of a low amniotic fluid index. The intrapartum period was uncomplicated, with artificial rupture of membranes 3 hours prior to a vaginal delivery. The infant cried immediately and had APGAR scores of 8 and 9 at 1 and 5 minutes, respectively.

NEWBORN PHYSICAL EXAMINATION

The infant's weight was 3.7 kg, her length was 51.5 cm, her head circumference was 35 cm, and her chest circumference was 33.5 cm; all of these measurements were appropriate for her gestational age of 38+ weeks. Her initial vital signs were normal.

The infant was very light skinned with abundant curly reddish-blonde hair and white eyelashes and eyebrows. She was alert, active, and exhibited appropriate newborn reflexes. Her head, ear, nose, and throat examination were unremarkable. Bilateral red reflex

FIGURE. The albino newborn. This figure appears in color online at www.jpedhc.org. Used with permission.



was present; her eyes were noted to be blue. The infant's cardiovascular and pulmonary examination findings were normal. The vertebral column had no tufts of hair or sacral dimple. The abdomen was soft without organomegaly; the clamped umbilical cord had three vessels. The genitalia examination was normal for a term female. Barlow and Ortolani tests were negative.

A Ballard examination was consistent with 38+ weeks' gestational age. The newborn hearing test was normal. Transcutaneous bilirubinemia tests at 24 and 48 hours of life were in the low to intermediate risk range for the development of hyperbilirubinemia.

ASSESSMENT AND PLAN

Upon initial contact with the infant's mother, the extremely different physical characteristics of the infant compared with the parents and 4-year-old sibling became obvious. These notable differences prompted the health care providers to obtain a genetic consultation.

GENETIC CONSULTATION

A genetics counselor met with the mother and discussed a possible diagnosis of oculocutaneous albinism type 2 (OCA2). Genetic testing for OCA2 to confirm the diagnosis was discussed. The counselor also recommended that an ophthalmologist be consulted to determine any ocular changes—a prominent feature of albinism.

OPHTHALMOLOGY CONSULTATION

After dilating the infant's eyes with 1% tropicamide, the ophthalmologist evaluated the iris, fundus, and extraocular motility. His findings suggested a diagnosis of OCA with the need for ophthalmologic follow-up after completion of the genetic testing. He also provided a plan of care (POC) to ensure that the infant received the most evidence-based treatment to maximize her visual potential.

FOLLOW-UP

During the hospital admission, the infant's mother stated that the family was moving to an East Coast city. Prior to the infant's delivery, the mother had secured a pediatrician in this city and would make arrangements for a genetics referral and follow-up with an ophthalmologist. The medical team provided the information necessary to initiate care as soon as possible.

CASE STUDY QUESTIONS

1. What is OCA, and what are the different types? What is the significance of genetic testing in diagnosing OCA?
2. In proposing an evidence-based POC for the infant, what interventions should be included to ensure maximum visual potential? Why?
3. What interventions should be addressed in providing anticipatory guidance to promote achievement of early developmental milestones (i.e., motor, cognitive, language, self-help, and social) for a visually impaired newborn (VIN)?
4. What are the psychosocial stigmas encountered by albinos? What should a health provider do to lessen these stigmas?

CASE STUDY ANSWERS

1. *What is OCA, and what are the different types? What is the significance of genetic testing in diagnosing OCA?*

OCA is an autosomal-recessive genetic condition characterized by a reduction or absence of melanin synthesis in the skin, hair, and eyes. The prevalence of OCA