

Wobbly Eyes in a Fair Child: Clues to a Rare Diagnosis

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Abstract

Oculocutaneous albinism (OCA) comprises inherited disorders characterized by reduced or absent melanin pigmentation of the skin, hair, and eyes. Ocular involvement may present early with nystagmus and other signs of impaired visual development. When hypotonia or developmental delay accompanies albinism, a careful neurological and syndromic evaluation is required. We report a 7-month-old female infant who presented with generalized hypopigmentation, rapid involuntary eye movements since early infancy, poor head control, and parental concern regarding motor delay. Examination showed milky-white skin; platinum-blond hair, eyebrows, and eyelashes; fine horizontal conjugate pendular nystagmus; iris transillumination; compensatory head posture; and generalized axial and appendicular hypotonia with preserved, symmetrical deep tendon reflexes. A family history of similar generalized hypopigmentation among multiple maternal relatives supported an inherited disorder. Fundal photography provided additional ophthalmic documentation of ocular hypopigmentation. Because neurological involvement raised concern for syndromic albinism, a peripheral blood smear was examined; giant cytoplasmic granules characteristic of Chediak-Higashi syndrome were not identified. The overall clinical picture was consistent with oculocutaneous albinism with infantile nystagmus and hypotonia. This case highlights the importance of multidisciplinary evaluation of infants with hypopigmentation and neurological signs, with attention to syndromic mimics and early visual and developmental intervention.

Keywords: *Oculocutaneous Albinism; Infantile Nystagmus; Hypotonia; Hypopigmentation; Developmental Delay*

Introduction

Infantile nystagmus associated with hypotonia warrants systematic neuro-ophthalmological assessment because the motor findings may coexist with primary neurological disease, syndromic albinism, or impaired development secondary to early visual dysfunction. Oculocutaneous albinism is characterized by reduced or absent melanin pigmentation involving the skin, hair, and eyes. Ocular melanin deficiency is associated with abnormal visual-system development, including foveal hypoplasia and abnormal routing of retinal ganglion-cell axons at the optic chiasm, and may manifest as nystagmus, iris transillumination, reduced fundal pigmentation, strabismus, and reduced visual acuity. We describe an infant with generalized hypopigmentation, characteristic ocular findings, and generalized hypotonia, emphasizing the clinical approach to excluding an important syndromic mimic. [1,2]

Case Presentation

A 7-month-old female infant presented with generalized hypopigmentation and rapid involuntary eye movements noted since early infancy. Her parents also reported progressive concern regarding motor development, particularly poor head control. Family history was notable for similar generalized hypopigmentation among multiple maternal relatives, suggesting an inherited disorder of pigmentation.

Dermatological examination revealed generalized marked hypopigmentation, with milky-white skin and platinum-blond hair, eyebrows, and eyelashes. Ophthalmological examination demonstrated fine, horizontal, conjugate pendular nystagmus with iris transillumination. A compensatory head posture was present, characterized by rightward facial rotation with downward chin tilt. Fundal photography documented a markedly hypopigmented fundal appearance with increased visibility of the underlying choroidal vasculature; the foveal reflex was not clearly identifiable in the available photograph. These photographic findings were compatible with ocular hypopigmentation. Foveal hypoplasia was not considered confirmed from the photograph alone.

Neurological examination showed significant generalized axial and appendicular hypotonia with pronounced head lag. Deep tendon reflexes were intact and symmetrical, making a peripheral lower motor neuron pattern less likely on clinical examination.

In view of the combination of hypopigmentation and neurological findings, syndromic forms of albinism were considered. Peripheral blood smear examination did not demonstrate the giant cytoplasmic granules classically associated with Chediak–Higashi syndrome. The constellation of generalized hypopigmentation, characteristic ocular signs, family history, and fundal hypopigmentation was clinically consistent with oculocutaneous albinism.

Discussion

Oculocutaneous albinism is an inherited disorder of pigmentation in which reduced melanin affects the skin, hair, and visual system. Ocular abnormalities are central to the phenotype. Reduced pigmentation of the iris and retinal pigment epithelium produces iris transillumination and a hypopigmented fundus, while abnormal visual-system development may include foveal hypoplasia and excessive crossing of optic nerve fibres at the chiasm. These abnormalities contribute to impaired visual function and infantile nystagmus.

In the present child, the combination of generalized hypopigmentation, platinum-blond hair, iris transillumination, infantile horizontal nystagmus, and fundal hypopigmentation strongly supported OCA. The family history of similarly affected maternal relatives further supported a heritable pigmentation disorder. The compensatory head posture may reflect an attempt to obtain better visual function at a position in which nystagmus is reduced; however, a specific null point was not documented in the source material.

The associated generalized hypotonia required separate consideration. Severe early visual impairment can adversely affect visually guided motor development and may contribute to delayed acquisition of head control and other motor milestones. Nevertheless, clinically significant hypotonia should not be attributed to visual impairment without evaluation for neurological and syndromic causes. The preserved and symmetrical deep tendon reflexes in this infant did not suggest a classic lower motor neuron pattern, although they do not by themselves exclude all peripheral neuromuscular disorders.

Chediak–Higashi syndrome is an important syndromic differential in a child with hypopigmentation and neurological abnormalities. It is associated with partial albinism, immune dysfunction and characteristic giant lysosomal granules in leukocytes. No giant cytoplasmic granules were identified on the peripheral blood smear in this case, making Chediak–Higashi syndrome less likely. Other syndromic forms of albinism should be considered when suggested by bleeding tendency, recurrent infections, progressive neurological abnormalities, or other systemic features.

The available fundus photograph supports ocular hypopigmentation, but structural foveal hypoplasia should not be claimed solely from this image. When feasible, optical coherence tomography can provide objective assessment of foveal architecture and grading of foveal hypoplasia. Similarly, molecular genetic testing can define the OCA subtype and strengthen genetic counselling, but no genetic result was provided in the source presentation and therefore no molecular subtype is assigned in this report. [1,2]

Conclusion

Generalized hypopigmentation accompanied by infantile nystagmus should prompt detailed dermatological and ophthalmological evaluation for oculocutaneous albinism. When hypotonia or developmental delay is also present, clinicians should evaluate for associated neurological disease and syndromic forms of albinism rather than assuming that all motor findings are secondary to visual impairment. Early multidisciplinary care, including visual habilitation, developmental surveillance, physiotherapy when indicated, and appropriate genetic evaluation, may optimize functional outcomes and facilitate family counselling.

Learning Points

- The combination of generalized hypopigmentation, iris transillumination, fundal hypopigmentation, and infantile nystagmus is strongly suggestive of oculocutaneous albinism.
- Hypotonia in an infant with albinism warrants neurological and syndromic evaluation; it should not automatically be attributed to impaired vision.
- Peripheral blood smear is a useful initial investigation when Chediak–Higashi syndrome is clinically suspected.
- Fundal hypopigmentation can be documented photographically, whereas foveal hypoplasia is better assessed with dedicated ophthalmological evaluation and optical coherence tomography.
- Early multidisciplinary visual and developmental intervention is important in affected infants.

Conflicts of Interest

The authors declare no conflicts of interest.

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