Anophthalmia in a Newborn

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Neonatology was called to evaluate a 1-day-old male who had not yet opened his eyes. The infant was born by normal spontaneous vaginal delivery to a 21-year-old primigravida mother following a full-term pregnancy complicated by gestational diabetes that was well-controlled with insulin. Delivery was uncomplicated. The infant had received erythromycin ointment to his eyes after birth and had been circumcised without event. Physical exam was significant for mild periorbital edema, deeply set orbits and small palpebral fissures that were difficult to separate (Figure 1). The infant also had a high arched palate and micropenis.

MRI showed complete absence of the globes, optic nerves and optic chiasm (Figure 2). The remainder of the brain was anatomically normal. Comparative genomic hybridization demonstrated deletion of the SOX 2 gene, a known mutation associated with anophthalmia. A three-generation pedigree was unremarkable, which was not surprising given that most SOX 2 gene mutations occur de novo. Abnormalities in this gene may be associated with pituitary dysfunction, commonly presenting as micropenis in males. Markers of pituitary function including LH, FSH, T4, TSH and cortisol were normal. Testosterone was mildly low.

After discharge, the infant was to be followed closely by endocrinology. Additional referrals were made to oculoplastic surgery because of the effect the absent optic globes may have on facial growth, and to early intervention services for developmental monitoring.

Discussion

Anophthalmia is a rare ocular defect resulting from abnormal development of the primary optic vesicle. There is complete absence of all eye tissue with no chance for vision. Anophthalmia is associated with central nervous system abnormalities, skeletal defects, genitourinary malformations and metabolic disturbances in more than half of affected individuals.1,2

Anophthalmia should be considered in a newborn with delay in eye opening. Physical exam may be suggestive of this disorder, but findings can be subtle and initially may be missed. Neuroimaging is necessary for diagnosis, and to screen for other brain abnormalities. Further screening for associated endocrine, skeletal and genitourinary conditions should be considered. Genetic counseling and testing for SOX 2 gene mutations may be useful in understanding the risk for recurrence in some families. Other subspecialty referrals may be appropriate as well. MM

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REFERENCES


Learning points

• Anophthalmia should be considered in a newborn with delay in eye opening.
• Early diagnosis, screening for associated conditions and appropriate referrals are key to the ongoing care of children with this rare disorder.

FIGURE 1
Male Infant with Anophthalmia

FIGURE 2
MRI Image of Brain of Child with Anophthalmia