A Case of Anterior Segment Dysgenesis with Iridolenticular Adhesions in Trisomy 18

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Abstract

Trisomy 18 (or Edwards syndrome) has an incidence of 1 in 6,000 to 8,000 live births, making it the second most common trisomy after trisomy 21. Ophthalmologic anomalies include epicanthal folds, hypertelorism, and hypoplastic supraorbital ridges, whereas corneal opacities, microcornea, congenital glaucoma, cataract, retinal depigmentation, retinal vascular tortuosity, colobomatous microphthalmia, and cyclopia are thought to be less common; iridolenticular adhesions have not been previously reported. Our patient was a female with confirmed trisomy 18 with ophthalmologic examination revealing corneal opacities and iridolenticular adhesions. Insofar as corneal opacities are a known entity in trisomy 18 and have been considered by some to be of clinical importance, iridolenticular adhesions may also be a noteworthy manifestation of the disease's anterior segment dysgenesis.

Keywords

- ▶ trisomy 18
- ▶ anterior segment dysgenesis
- ▶ iridolenticular adhesions

Introduction

Trisomy 18 (or Edwards syndrome) has an incidence of 1 in 6,000 to 8,000 live births, making it the second most common trisomy after trisomy 21.¹ Typically it manifests systemically with intrauterine growth retardation, dysmorphic facies, cardiac defects, and clenched fists with overlapping fingers. There is a high mortality rate in the neonatal period with only a 5 to 8% survival rate. The most common cause of death includes cardiopulmonary arrest, congenital heart disease, and pneumonia.

The most common ophthalmologic anomalies include epicanthal folds, hypertelorism, and hypoplastic supraorbital ridges, whereas corneal opacities, microcornea, congenital glaucoma, cataract, retinal depigmentation, retinal vascular tortuosity, colobomatous microphthalmia, and cyclopia are thought to be less common.^{2,3} To the best of our knowledge, iridolenticular adhesions have not been previously reported.

Case Report

The patient was a 2.5-kg female born by cesarean section at term for failure to progress and a non-reassuring fetal heart

tracing. Ophthalmic examination demonstrated shortened horizontal palpebral fissures, 8.5-mm corneas, limbal pannus, stromal opacities in the temporal quadrants of both corneas encroaching on the visual axes, and iridolenticular adhesions (–Fig. 1); the ocular adnexae, anterior segments, and posterior segments were otherwise normal in appearance. There was a hirsute forehead, low-set malformed auricles, widened flat nasal bridge with a prominent nasal tip, redundant neck folds, widely spaced nipples, and bilateral overlapping of the second digit over the third and fifth digits over the fourth.

She was the offspring of a healthy 37-year-old Hispanic gravida two, nonconsanguineous mother. Prenatal ultrasound showed severe polyhydramnios, intrauterine growth restriction, bilateral choroid plexus cysts, and a two-vessel umbilical cord; fetal echocardiogram revealed a large ventriculoseptal defect and a stenotic pulmonic valve. The parents declined amniocentesis. Apgar scores were 5 at 1 minute and 7 at 5 minutes; head circumference was 30.5 cm and birth length was 47 cm. Chest radiograph showed 11 paired, thin ribs, cranial ultrasound showed a small collection of extra-axial fluid in the interhemispheric fissure, and abdominal ultrasound revealed a small right-sided adrenal

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Fig. 1 Eye findings. Dysmorphic features consistent with trisomy 18 with corneal opacities.

hemorrhage and ipsilateral pelviectasis. Karyotype confirmed 47 XX +18. The infant expired on day 14; her parents declined autopsy.

Discussion

In our experience at a large quaternary pediatric referral center in the San Francisco Bay Area, this case is the first instance of corneal opacities and iridolenticular adhesions observed by the genetics services in over 20 years. Insofar as corneal opacities are a known entity in trisomy 18 and have been considered by some to be of clinical importance, iridolenticular adhesions may also be a noteworthy manifestation of the disease's anterior segment dysgenesis.⁴ It is possible that the eye findings in this patient have been previously described as corneal opacities or indeed this could represent new eye findings in trisomy 18. Based on histopathologic studies from the 1960s that showed irregular or absent epithelium and endothelium of the cornea,⁵ one hypothesis for the iridolenticular adhesions may be irregular, "sticky" lens and iris pigment epithelium leading to increased ocular pressure, damage to the structure of the eye, and optical distortion.

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