Infantile Glaucoma and Punctal Atresia in a Child With Caudal Regression Syndrome

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Caudal regression syndrome (CRS) is a rare embryopathy characterized by maldevelopment of the vertebrae, visceral organs, and lower extremities. Patients with CRS have deformities of the sacrococcygeal vertebrae and develop neurogenic bladder from dysfunction of the sacral nerve root.[1-3] Additional anomalies include congenital heart defects, intestinal malrotation, agenesis of the genitourinary system, imperforate anus, and malformation of the lower limbs.[1-3] Ocular findings described in the nonophthalmic literature have been confined to hypertelorism, epibulbar dermoid, and anophthalmia.[3,4] We report a patient with CRS who presented with infantile glaucoma and punctal atresia. A computerized search of the medical literature showed no previous reports of CRS in ophthalmic journals and no previous reports of an association with glaucoma or nasolacrimal anomalies.

CASE REPORT

A 10-week-old infant presented with a 1-week history of bilateral epiphora and photophobia. The patient had neonatal documentation of lumbosacral vertebral anomalies, atrioseptal defect, bifid clitoris, imperforate anus, dislocated hip, and syndactyly of both feet (Fig 1). At 9 weeks, she underwent repair of a malrotated intestine and vesicostomy for neurogenic bladder. The mother's health and prenatal care were excellent, and the pregnancy and delivery were unremarkable. Family history was negative for parental consanguinity or hereditary ocular or systemic disease.

The infant was noted to have hypertelorism, and lower lid punctal agenesis. Both corneas exhibited diffuse stromal thickening, which was more severe in the left eye. Horizontal corneal diameters measured 11.5 mm in the right eye and 12.5 mm in the left eye. Intraocular pressures (IOP) measured serially using a hand-held applanation tonometer were 30 mm Hg in each eye. Portable slit-lamp examination showed no evidence of anterior segment dysgenesis. Fundoscopic examination showed cup-to-disc ratios of 0.5 (right eye) and 0.6 (left eye).

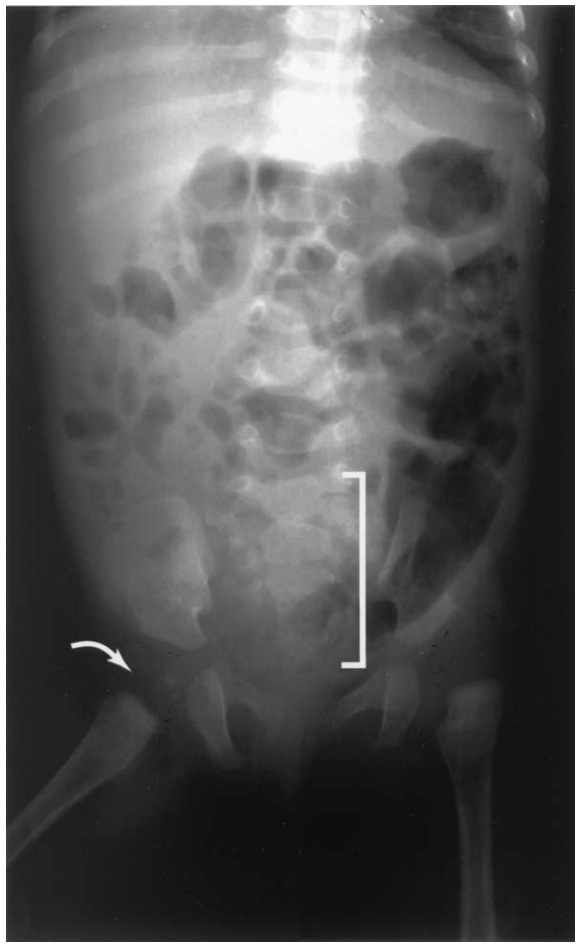

FIG 1. Radiographic features of caudal regression syndrome of our patient at 3 months. Plain radiographs show anomalous, fused sacral vertebrae (bracket) and a congenitally displaced right hip (arrowhead).

The next day, an examination was performed under anesthesia to confirm the diagnosis of glaucoma, after which 90° goniotomies were performed first in the right eye and 1 week later in the left eye. The corneal edema progressively resolved during the next month, and the cup-to-disc ratios decreased to 0.3 in both eyes. At 14 months, bilateral punctoplasty and nasolacrimal duct reconstruction were carried out to repair the punctal atresia. At 3 years, repeat bilateral goniotomies were performed

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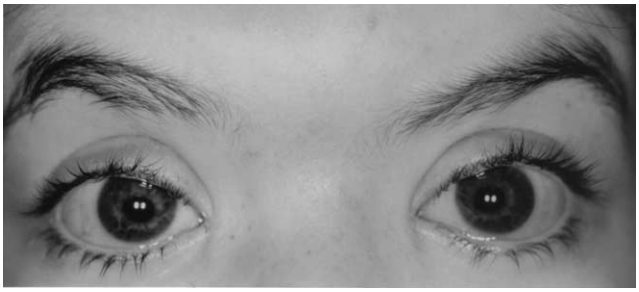

FIG 2. External photograph of our patient at age 13 years with bilateral megalocornea and buphthalmos secondary to infantile glaucoma.

for recurrent increases in IOP. Spectacles were prescribed for anisometropia and moderate myopic astigmatism, and occlusion therapy of the right eye was instituted to treat anisometropic amblyopia of the left eye. At 12 years, transcleral cyclophotocoagulation (TSCP) was used on her left eye for further IOP control, and bilateral TSCP was applied one year later. At 13 years (Figure 2), her best-corrected visual acuity was 20/30 in the right eye and 20/70 in the left eye (cycloplegic refraction: right eye = $-4.50 + 2.25 \times 130$ and left eye = $-6.25 + 2.25 \times 50$). Her IOPs (<20 mm Hg) were stable, with corneal diameters of 13.0 mm and cup-to-disc ratios of 0.3 bilaterally. A-scan ultrasonography documented axial lengths in the right eye of 24.27 mm and in the left eye of 24.52 mm.

DISCUSSION

Caudal regression syndrome occurs in 1/100,000 live births, and there is no apparent gender predilection. It is believed to arise from an embryonic defect of caudal mesoderm development that occurs before the fourth week of gestation.[1,2] Although its etiology remains unclear, maternal diabetes has been associated with CRS, and insulin injection in chick embryos has been shown to cause malformations similar to CRS.[1-3]

The child we report with CRS had punctal atresia, infantile glaucoma, and significant myopia. Primary infantile glaucoma is thought to arise from neural crest maldevelopment, resulting in goniodysgenesis. The low incidence of CRS ($1/10^5$ births) and infantile glaucoma ($1/10^4$ births)—as well as the embryopathic nature of each disorder—suggest a causal linkage rather than chance association. The presence of punctal atresia further supports the notion of embryopathic linkage because the atresia is believed to stem from faulty development of surface ectoderm.[5] The embryopathies of congenital rubella syndrome and fetal alcohol syndrome are also associated with developmental glaucoma, and ectrodactyly ectodermal dysplasia-clefting syndrome has been associated with punctal atresia.[5]

The management of glaucoma in our patient was challenging and required multiple procedures to control IOP. We suggest that patients with CRS be examined early in infancy by an ophthalmologist to rule out nasolacrimal anomalies and developmental glaucoma.

References

1. Escobar LF, Weaver DD. Caudal regression syndrome. In: Buyse ML, editor. Birth defects encyclopedia. Cambridge (MA): Blackwell Scientific; 1990. p. 296-7.
2. Adra A, Cordero D, Mejides A, Yasin S, Salman F, O'Sullivan M. Caudal regression syndrome: etiopathogenesis, prenatal diagnosis, and perinatal management. Obstet Gynecol Surv 1994;49:508-16.
3. Russell LJ, Weaver DD, Bull MJ. The axial mesodermal dysplastic spectrum. Pediatrics 1981;67:176-82.
4. Bini R, Danti DA, Materassi M, Pela I. Report of a new case of axial mesodermal dysplasia complex. Clin Genet 1996;50:407-10.
5. Boerner M, Seiff SR, Arroyo J. Congenital absence of the lacrimal puncta. Ophthalmic Surg 1995;26:53-6.