

# Perinatal/Neonatal Case Presentation

## Bilateral Cataracts, Retinal Detachment and Vitreous Hemorrhage in a Newborn with Congenital Diaphragmatic Hernia

**Pankaj Kumar, MD**  
**Philip J Ferrone, MD**  
**Joyce Fox, MD**  
**Robert Koppel, MD**

Congenital diaphragmatic hernia (CDH) is associated with a wide range of other malformations. We describe a patient with CDH who also had significant eye findings noted at birth.

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### INTRODUCTION

Despite fetal diagnosis and improved perinatal management, the high mortality of patients with congenital diaphragmatic hernia (CDH) is largely due to the presence of secondary pulmonary hypoplasia and other nonpulmonary malformations and chromosomal defects.<sup>1–5</sup> We report a patient with CDH who had unique ophthalmological complication.

### CASE REPORT

A 3.6-kg full-term male infant was transferred for management of persistent pulmonary hypertension secondary to left-sided CDH. The baby was born by a repeat C-section to a 28-year-old gravida 5 para 4, Hispanic woman with negative serology following an uncomplicated pregnancy. The siblings were reported to be normal and healthy. Family history was negative for consanguinity, eye disease and genetic disorders. No abnormalities were reported on prenatal ultrasonography and no amniocentesis was performed. Except for a right undescended testis, the physical examination at birth was unremarkable. Weight, height and head circumference

were within normal percentiles for age. Extracorporeal Membrane Oxygenation (ECMO) was initiated on the second day of life and continued for 14 days. Hernia repair was performed 5 days after ECMO decannulation. There was no facial dysmorphism or obvious external eye abnormality at the time of admission. A dilated funduscopic examination was not performed before ECMO initiation.

Following ECMO, right eye ptosis was noted and an ophthalmology consultation was obtained. On dilated funduscopic examination, bilateral posterior polar type cataracts with a shallow anterior chamber were seen. There was also a retropupillary membrane in both eyes. Both eyes were of equal and normal size. The presence of light perception in either eye was considered unlikely. The posterior segment could not be visualized due to the retropupillary membrane. On B-scan posterior segment disorganization was seen with subretinal and vitreous hemorrhage and total retinal detachments in a closed funnel configuration. A decision was made to attempt rehabilitation of one eye. After lensectomy and vitrectomy were performed on the right eye, uncorrectable retinal dysplasia with a total closed funnel traction retinal detachment and subretinal and preretinal hemorrhage were noted. The child had a normal hearing examination. The child had normal male karyotype (46 XY). The history and the physical findings, other than the eyes, were not consistent with known syndromes. Pulmonary hypertension resolved by the time of discharge.

### DISCUSSION

CDH continues to have high mortality despite recent advances in prenatal diagnosis, neonatal intensive care and surgery. Patients with CDH, as an isolated anomaly, generally have a better prognosis than those with associated malformations.<sup>6</sup> Associated malformations may occur with trisomies 13,18 & 21, nontrisomy chromosomal disorders, or recognizable nonchromosomal syndromes. The most common associated anomalies are cardiovascular and neural tube defects.

Only a few case reports describing ophthalmologic findings with diaphragmatic hernia are available. Gripp et al.<sup>7</sup> reported one patient with CDH and exomphalos–hypertelorism syndrome including bilateral iris colobomata and myopia, facial dysmorphism (hypertelorism and downslanting palpebral fissures), sensorineural deafness, exomphalos, and a history of consanguinity

*Division of Neonatal-Perinatal Medicine (P.K., R.K.), Schneider Children's Hospital, Long Island Jewish Medical Center, The University Hospital and Long Island Campus for the Albert Einstein College of Medicine, New Hyde Park, NY, USA; Department of Human Genetics (J.F.), Schneider Children's Hospital, Long Island Jewish Medical Center, The University Hospital and Long Island Campus for the Albert Einstein College of Medicine, New Hyde Park, NY, USA; and Long Island Vitreoretinal Consultants (P.J.F.) Great Neck, NY, USA.*

*Address correspondence and reprint requests to R Koppel, MD, Division of Neonatal- Perinatal Medicine, Schneider Children's Hospital, 269 –01 76th Avenue, New Hyde Park, NY 11040, USA.*

suggesting an autosomal recessive inheritance. Goddeeris et al.<sup>8</sup> reported on stillbirths with CDH and cloudy corneas, microphthalmia as well as distal limb deformities and cleft palate suggesting a lethal syndrome now widely known as Fryns syndrome. Other ocular findings in Fryns syndrome include irregularities of Bowman's layer, thickened posterior lens capsule and retinal dysplasia.<sup>9</sup> These patients demonstrate an autosomal recessive pattern of inheritance. Microphthalmia and anophthalmia have been described with diaphragmatic defects.<sup>10,11</sup> CDH has also been reported with blepharophimosis, ptosis, and epicanthus inversus that were found to be associated with interstitial deletion of long arm of chromosome 3.<sup>12</sup> Another fetal malformation with bilateral CDH, bilateral anophthalmia, other anomalies and characteristic skin lesions has been described as a severe form of Goltz Syndrome.<sup>13</sup> Stickler syndrome, characterized by flat facies, myopia and arthropathy, has been associated with bilateral cataracts, retinal detachment and diaphragmatic hernia.<sup>15</sup>

Asymmetrical retinopathy with intraretinal hemorrhages has been reported in patients who underwent veno-arterial ECMO for CDH.<sup>14</sup> Unstable circulatory control may predispose patients with CDH to retinal vascular changes independently or with ECMO. However, the same study showed no short-term sequelae associated with this vasculopathy. There are limited data regarding long-term sequelae of common carotid ligation.

Our patient differs from those previously reported as bilateral cataracts and retinal detachment with vitreous hemorrhage have not been previously described in association with CDH. Ptosis is possibly an iatrogenic complication of ECMO cannulation, but the other eye findings were congenital and unrelated to ECMO. In the absence of other physical findings and family history, these findings do not fit into the CDH–Exomphalos–Hypertelorism syndrome<sup>7</sup> or Fryns syndrome. There was no family history of Stickler syndrome, joint problems, cleft palate or hearing loss. Ophthalmologic findings in this infant were not consistent with those seen in Stickler syndrome.

It is difficult to determine whether the above-described eye findings in association with CDH are coincidental or part of a new syndrome not yet described. In view of the range of eye abnormalities associated with CDH, the authors recommend that after initial stabilization, CDH patients should undergo a careful

fundoscopic eye examination in order to enable early diagnosis and treatment if necessary.

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