A Preterm Newborn with Abnormal Development of the Lower Abdomen and Genitalia

Saikiran Deshabhotla, MD, DNB (Neonatology),* Amrut Prajapati, DCH, FIAP,* Baswaraj Tandur, DCH, DNB,* Varma Vidyasagar, DCH*

*Princess Durru Sehvar Children's Hospital, Hyderabad, India

THE CASE

A preterm newborn is noted after delivery to have abnormal development of the lower abdomen and genitalia.

Prenatal and Birth Histories

- Born to a 23-year-old, gravida 1 woman; nonconsanguineous marriage
- Conception after ovulation induction
- Normal prenatal maternal laboratory screening and fetal survey
- Pregnancy complicated by vaginal bleeding noted 5 days before delivery and preterm labor at 32 weeks’ gestation
- Vaginal delivery at an estimated gestational age of 32 weeks
- Apgar scores: 7 and 9 at 1 and 5 minutes, respectively; no resuscitation was required

Presentation (Newborn Day)

Immediately after birth, the neonatology team that was present at delivery examined the infant and noted a lower abdominal mass and ambiguous genitalia (Fig 1). The infant was transferred to the NICU for further evaluation. On admission, the newborn was noted to have central cyanosis and given nasal prong continuous positive airway pressure (CPAP).

PROGRESSION

Vital Signs

- Heart rate: 140 beats/min
- Respiratory rate: 60 breaths/min
- Temperature: 96.8°F (36.0°C)
- Oxygen saturation: 90% on respiratory support of CPAP with positive end-expiratory pressure of 5 cm H₂O

Physical Examination

- Birthweight: 1,560 g (10th percentile)
- Birth head circumference: 28 cm (10th percentile)
- Birth length: 40 cm (10th percentile)
- Head: Normal, open and flat fontanelles; no facial dysmorphism; intact palate
- Lungs: Normal chest shape, mild respiratory distress with moderate intercostal retractions

AUTHOR DISCLOSURE

Drs Deshabhotla, Prajapati, Tandur, and Vidyasagar have disclosed no financial relationships relevant to this article. This commentary does not contain a discussion of an unapproved/investigative use of a commercial product/device.
Cardiovascular: Normal precordial shape and heart sounds, no murmur, appropriate peripheral pulses
Back: Soft, cystic, skin-covered swelling over right para lumbar region (Fig 2)
Abdomen and genitourinary: Bilateral bladder swellings; ambiguous genitalia with bilateral bifid scrotal swellings and absent testes; incontinence of urine and stool from ectopic opening below perineum (Fig 3)
Anus: Imperforate anus; an elephant trunk appearance of the perineum
Extremities: Left-sided talipes equinovarus (suspected agenesis of talus)
Hips: Left-sided hip dislocation

Laboratory Studies
- White blood cell count: 10,000/µL (10 x 10^9/L)
- Differential count: Neutrophils 60%, lymphocytes 35%, eosinophils 3%, basophils 2%
- Serum electrolytes: Sodium 145 mEq/dL (145 mmol/L), potassium 3.0 mEq/dL (3.0 mmol/L), chloride 110 mEq/dL (110 mmol/L)
- Blood urea nitrogen 32 mg/dL (11.4 mmol/L), serum creatinine 0.9 mg/dL (80 µmol/L)
- Mild metabolic acidosis
- Karyotype and genetic studies: This testing was advised but family declined due to financial constraints

Imaging Studies
- Chest radiography: Reticulogranular pattern consistent with surfactant deficiency
- Abdominal radiography: Lumbar hemivertebrae and widely spaced pubic bones
- Abdominal ultrasonography: Absent bladder, hydroureter, hydromelia in the lumbosacral region; absence of uterus, ovary, or testes; both kidneys were visualized and were normal in size and echotexture
- Echocardiography: Moderate-sized patent ductus arteriosus, no other structural abnormality
- Cranial ultrasonography: Normal

Differential Diagnosis
- Bladder extrophy
- Omphalocele-extrophy-imperforate anus-spinal defect (OEIS) complex

ACTUAL DIAGNOSIS
The infant was diagnosed as having OEIS complex. The infant’s specific findings were identified.

MANAGEMENT
The infant received intravenous total parenteral nutrition and continued to receive CPAP until the second day after birth. The parents were counseled about the syndromic nature of the infant’s condition and the concerning long-term prognosis. Because of the poor prognosis, as well as for cultural reasons, the family opted for palliative and supportive care and declined surgical interventions. The infant was transferred to a health facility closer to the family’s residence and continued to receive total parenteral nutrition. The infant died at 45 days of age after developing sepsis and pneumonia.

WHAT THE EXPERTS SAY
OEIS complex is rare, with a reported incidence of 1 in 200,000 to 400,000 births. (1) OEIS complex consists of phallic separation with an epispadias, pubic diastasis, extrophy of the bladder (exposed bladder mucosa), and cloacal extrophy (ventral body wall defect). (2)(3)(4)
OEIS is the result of an isolated defect that occurs during the early blastogenesis stage (4–6 weeks’ gestation) with abnormal development of mesoderm. This contributes later in gestation to infraumbilical mesenchyme, responsible for the formation of a cloacal septum, and caudal vertebrae. Because of the abnormal infraumbilical mesenchyme development, cloacal septation does not take place. Thus, the ureters, ileum, and rudimentary hindgut open into this common cloaca. An associated maldevelopment of the lumbosacral vertebrae in the form of a dilated central canal and herniation (ie, hydromyelia) is a common finding. Abnormal formation of the urorectal septum results in the failure of the urogenital sinus to separate from the rectum. (5)

Even though the etiology of OEIS is unknown and the occurrence is sporadic, several associations have been noted, including teratogenic exposure to antiepileptic drugs, mutations in Homeobox genes (such as HLAB9), and chromosomal aneuploidies (eg, 47,XXX and T18). (3)(6)(7)

Associated anomalies include the following: hypoplastic chest, diaphragmatic hernia, meningocele, tethering of the cord, scoliosis, pelvic kidneys, renal agenesis, multicystic kidney, ureteral duplication, a single umbilical artery, and anomalies of the lower limbs including congenital hip dislocation, talipes equinovarus, and limb agenesis. Boys may have cryptorchidism or a bifid penis with widely separated pubic bones and affected girls have müllerian duct orifices below the bladder mucosa with completely bifid uterine horns, a bifid clitoris, and a short, duplicated, or atretic vagina. (8)

The antenatal diagnosis of OEIS is possible by identifying a midline infraumbilical abdominal wall defect with an irregular cystic mass, absence of the bladder between the 2 umbilical arteries, a lumbosacral neural or vertebral defect, abnormalities of the inferior limbs, and/or a wide pubic arch with symphysis pubis diastasis. (9)(10) Maternal serum α-fetoprotein levels may be elevated. Most cases of OEIS complex are confirmed after birth or at autopsy after the pregnancy is terminated.

Early prenatal diagnosis of OEIS complex allows clinicians to offer families the option to terminate the pregnancy. If a family decides to continue the pregnancy, early diagnosis can allow the family to meet with specialists to plan the appropriate perinatal management. Delivery by cesarean is typically recommended to avoid shoulder dystocia and trauma. During surgical repair, the omphalocele is closed, the gastrointestinal tract is separated from the hemibladders, and the hemibladders are formed into 1 viscus. If feasible, the colon can be pulled through, either at the time of this initial surgery or at a later date. The bladder must be reconstructed using the infant’s stomach and a continence mechanism is constructed by narrowing the outlet and inserting a reversed small bowel nipple. Stool continence is achievable in most patients with the use of enema washouts. In most cases, experts recommend that genetic males should be raised as females as there is often inadequate tissue to create a phallus. (11) The prognosis for infants with OEIS complex is poor. (12)

American Board of Pediatrics
Neonatal-Perinatal Content Specification

- Know the pathogenesis and associated of omphalocele.

References


ANSWER KEY FOR APRIL 2018 NEOREVIEWS
Control of Breathing and Apnea of Prematurity: 1. D; 2. D; 3. D; 4. C; 5. B.
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NeoReviews 2018;19:e265
DOI: 10.1542/neo.19-4-e265

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