Association of imperforate anus and congenital diaphragmatic hernia in one of a twins who conceived by Assisted Reproductive Technology (ART)

Case Report

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Abstract:

Introduction: Combination of congenital diaphragmatic hernia (CDH) and anorectal malformation (ARM) is rare. In this report, we describe a combination of imperforate anus and CDH in one of twins who conceived by Assisted Reproductive Technology (ART).

Case Report: A female preterm newborn at 27 weeks of gestation was referred to our neonatal intensive care unit due to respiratory distress. She was conceived by ART and had combination of imperforate anus and CDH. She expired 18-hours after birth as a result of severe lung hemorrhage and there was no possibility of surgical repair due to poor clinical condition.

Conclusions: In spite of the low incidence of birth defects in the ART-conceived babies, continuing surveillance of them is necessary and the report of their birth defects is helpful.

Keywords: Imperforate Anus, Congenital Diaphragmatic Hernia, Assisted Reproductive Technology

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Introduction:

The incidence of congenital diaphragmatic hernia (CDH) is approximately 1 in 3000 live births and imperforate anus occur in 1 in 2500 to 5000 live births [1, 2, 3]. Although associated anomalies are common in each of these conditions, combination of CDH and anorectal malformation (ARM) is rare [4]. Compared to naturally conceived pregnancies, birth defects occur more frequently in singleton newborns conceived by assisted reproductive technology (ART). On the other hand, the risk of birth defects is higher in twins. Indeed, ART-conceived twins have a much higher risk for birth anomalies [5]. In this report, the combination of imperforate anus and CDH has been described in one of twins who conceived by ART.

Case report

A 32- year- old primigravida woman delivered a male and a female of twins at 27 weeks gestation by emergency cesarean section due to severe preeclampsia and fetal distress. The pregnancy was induced by in vitro fertilization (IVF) technique. Prenatal sonographic scan at 24 weeks had shown diaphragmatic hernia in one of the twins. There were no family history of CDH or other congenital defects. Mother had no history of smoking, alcohol or other illicit drugs intake during pregnancy. After delivery, twins were referred to our neonatal intensive care unit. In twin 1 and twin 2, birth weights were 1200 and 1100 gram, respectively. Twin one only had mild respiratory distress syndrome without any gross anomalies that managed with continuous positive airway

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pressure (CPAP). Twin 2 presented with severe respiratory distress syndrome and hypoxic respiratory failure and was immediately intubated and required to ventilator therapy. On physical examination, breath sounds in the left side were diminished and heart sounds were strongly auscultated on the right side of chest. In addition, she presented with imperforate anus without perineal fistula (Fig 1). She had no deformity of limbs or facial dysmorphism.

Echocardiography revealed a small atrial septal defect (ASD). Arterial blood gas revealed severe respiratory acidosis. Chest x -ray showed herniated bowel loops into the left thoracic cavity which contributed to severe lung hypoplasia and shift of heart to the right side of the mediastinum (Fig 2). No vertebral or lumbosacral anomalies were seen in spinal x-ray. An ultrasound scan of the kidneys was reported normal.

Surfactant was instilled intratracheally and she received respiratory support with high frequency oscillatory ventilation because of no response to the high set up of conventional mechanical ventilation. Respiratory failure was not treated in spite of using above treatments, so surgical repair was not possible because of poor clinical condition. Unfortunately, she expired 18-hours after birth as a result of severe lung hemorrhage and cardiorespiratory collapse.

Fig 1. Imperforate anus without perineal fistula in the patient

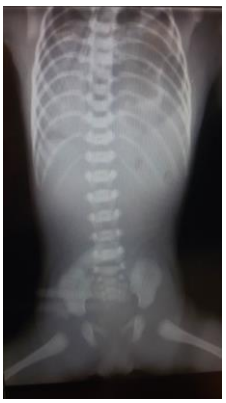

Fig 2. Plain radiography of the patient: herniated bowel loops into the left thoracic cavity and shift of heart to the right side of the mediastinum.

Discussion:

This report has been described a pair of ART-conceived twins, in the one of whom an imperforate anus and a left CDH coexisted. Association of CDH and ARM is rare [6]. Approximately 40-70% of neonates with ARM have additional congenital anomalies and the most commonly affected organ system is genitourinary tract [7, 8].

Other important associations include VACTERL (vertebral, anorectal, cardiac, trachea-esophageal, renal, limb) anomalies and trisomy of chromosome 21 [8, 9]. CDH has been rarely reported as an associated anomaly in ARM [8].

On the other hand, the frequency of additional congenital malformations in patients with CDH ranges from 30-40% [10]. The main involved organs included central nervous system, heart and genitourinary system [10]. It is, however, rare to find neonates with a combination of CDH and imperforate anus [4].

Walters et al.'s reported a newborn with imperforate anus, horseshoe kidney, pulmonary sling complex and diaphragmatic hernia. In their report, CDH had a late and delayed presentation at 4 months of age when the infant was admitted for a definitive laparoscopic assisted pull-through operation [4]. Raut et al.'s described a neonate with a combination of CDH on the right hemithorax and ARM. The patient was a term neonate without respiratory distress or other associated anomalies [6].

Similarly, Chen et al.'s described a neonate with VACTERL association and right sided CDH [10].

Cho et al.'s carried out a long- term retrospective analysis of 103 patients with ARM and described associated malformations of these infants. Only 3 patients had CDH in combination with an ARM that emphasized to the rarity of this co-existence [7].

Mirza et al.'s reported a case of imperforate anus associated with eventration of the diaphragm in a newborn infant who underwent a sigmoid colostomy and plication of diaphragm on second and 8th days of life, respectively [8]. A noticeable point in our report is the presentation of this associated rare anomaly in one of the twins who conceived via IVF technique. Already, a few cases of CDH or ARM have been reported in both twins. Kubiak et al.'s reported a case of isolated imperforate anus in monozygotic twins [3].

Ohn et al.'s described a case of CDH in dizygotic twins. According to Ohn et al.'s report, patients were the sixth survived cases of twins with CDH in the literatures [11].

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CDH in all cases was in the left hemithorax. Indeed, these cases confirmed that although CDH was a sporadic event in majority of cases, the rate of genetic factors in its etiology could not be completely excluded [11].

Concordance rate of birth defects after ART among twins suggests that familial aggregation is a factor in some birth anomalies [12]. It is well known that birth defects are more frequent in twin babies than singleton infants. On the other side, ART conceived pregnancies leads to more twin pregnancies and following more birth anomalies [5].

Increased risk of blastogenesis birth defects has been previously shown arising after ART [13]. Blastogenes is referred to the first 4 weeks of embryo development. This association has been reported for esophageal atresia, cleft palate and lip, septal heart defect and hypospadiasis [12].

One of specific defects in this category is anal atresia [13]. A few studies have also described an increased risk of ARM [13]. Zwink et al.'s in a German case-control study showed the increased risk of ARM among infants who born after ART (IVF and ICSI). This risk was also observed among both singleton and multiple births [14].

Therefore, occurring ASD and ARM in our presented case can be reasonable based on previous studies. However, published studies do not suggest any information on the increased risk of CDH in ART-conceived babies. Although CDH is not likely to be specifically associated with ART, this is the first report of ART-conceived twins in one of whom the combination of CDH and ARM was observed based on search in scholarly articles.

Unfortunately, in contrast to all above reports our presented case had no chance of surgical repair. It was due to severe prematurity, very low birth weight and severe lung hypoplasia.

In spite of the low incidence of birth defects in the ART-conceived babies, continuing surveillance of them is necessary and the report of their birth defects is helpful, too [15,16]. Indeed, most of the neonates conceived after ART are normal. However, there is increasing evidence that ART-conceived babies are at higher risk of birth defects and epigenetic disorders [17]. On the other hand, many factors can influence the neonatal outcome such as risks associated with the ART procedures themselves, higher incidence of maternal nulliparity, advanced maternal age and underlying etiology of parental infertility among others. Therefore, the exact mechanism of occurrence of birth defects and genetic disorders in ART-conceived patients is still unclear [18].

Report of this case may be useful to all caregivers in counseling patients about the birth defects' risk of twin pregnancies conceived following ART. Further researches are needed to elucidate the mechanism of this phenomenon.

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