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Congenital diaphragmatic hernia without antenatal diagnosis

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Introduction: Congenital Diaphragmatic Hernia (CDH) is a rare (approximately 1 in 3,000 newborns) condition but it is associated with high mortality (45-50%) and morbidity due to pulmonary hypoplasia and resistant pulmonary hypertension. Therefore, early diagnosis and proper management are the important keys to survival. CDH can often be diagnosed in utero with ultrasonography and Magnetic Resonance Imaging (MRI). In ultrasound examination polyhydramnios, intrathoracic stomach bubble, mediastinal and cardiac shift, fetal hydrops can be seen with low levels of maternal Serum Alpha-Fetoprotein (AFP). Intrauterine fetal surgery can be used to repair diaphragmatic defects to prevent the hypoplastic lung problems. But still small number of cases is performed and very few centers are capable of performing intrauterine surgery.

Case Presentation: A male, full-term newborn with a birth weight of 3100 g was delivered via normal spontaneous vaginal delivery from 25-year-old gravida 2, para 2 healthy mother. There was routine examination with appropriate antenatal care and monitoring during the pregnancy. Also, she was examined by perinatologist 3 times. But there were no significant antenatal problems. As the following delivery the baby was hypotonic and cyanotic immediately bag-mask positive pressure ventilation with 100% oxygen was given and suctioned. But there wasn't still spontaneous breathing with no sounds from bilateral lungs and the baby was bradycardic. Therefore, he is intubated and ventilated in the delivery room. Also, his abdomen appears scaphoid. There was bowel and stomach in the chest cavity with right shift of heart and mediastinum and bilateral pulmonary hypoplasia on chest X-ray. His first cord gas values were pH: 6.82, pCO_2: 139.2, HCO_3: 8.4, BE -11.7, pO_2: 98, SaO_2 23.8%. He was admitted at neonatal intensive care unit with diagnosis of CDH, but unfortunately, he died on the second day of the life.

Conclusion: Although prenatal diagnosis might be possible approximately 50% of cases by using ultrasound there is still large amount undiagnosed patients with CDH. Also fetal echocardiography and MRI might be used to diagnose antenatally and assess perinatal outcome. When it is diagnosed antenatally intrauterine fetal surgery can be used to repair diaphragmatic defects to prevent the hypoplastic lung problems. But further researches are needed to diagnose and make intrauterine fetal surgery.

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