Congenital diaphragmatic hernia: Early detection is imperative to improve outcome. Case report and literature review.

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Introduction:
Congenital diaphragmatic hernia (CDH) is the production of some of the abdominal or retroperitoneal structures through a defect in the diaphragm into the chest cavity. Typically, this occurs at about 10–12 weeks gestation. Common types include the paraesophageal or Bochdalek hernia, other types include Morgagni’s hernia, diaphragm evagination and central tendon defects of the diaphragm. Infants who are born with diaphragmatic hernia experience respiratory failure due to pulmonary hypoplasia. In babies who survive early neonatal period, there may be feeding difficulties, chronic respiratory disease, pneumonia, and intestinal obstruction. This entity is noted to be associated with other genetic anomalies like Di George syndrome, Chromosome 15, 18, 13, and 21 anomalies, Fryns syndrome, Pallister–Killian syndrome. Associated anomalies include Neural Tube Defects (28%), Cardiac anomalies (8–27%), malrotation (20%), omphalocele and genital anomalies (15%). Ultrasoundologically it is diagnosed when there is solid multiloculated complex cystic mass, mediastinal shift, fetal stomach at the level of heart, decreased AC ratio, herniated viscera into the chest shifting mediastinal structures. (1)

CDH occurs in approximately 1 in every 10,000 births. Making it more commonly affected than females with a ratio of 3:2. (2) Many cases of CDH are diagnosed on the 20-week Fetal Anomaly ultrasound scan. Some may be diagnosed during a third trimester ultrasound or occasionally as early as the dating scan. However, undiagnosed cases still occur and present at birth. The detection rate is 60% based on NHS Fetal Anomaly Screening Programme. Survival for babies born with CDH is approximately 50%, depending on the severity of the CDH and associated abnormalities. Unfortunately, mortality and morbidity remain high despite advances in neonatal intensive care with an overall 1 year survival estimated at 42%.

Case:
In this review we describe a case report where a congenital diaphragmatic hernia was undetected, neither in the abnormal scan nor in two consecutive ultrasound scans, in a baby that was born as an elective cesarean section. The baby was diagnosed postnatally by the newborn acquiring severe breathing difficulties, necessitating admission to the neonatal ward and later transfer to a tertiary centre for further management.

The mother is a 38-year-old lady G3P1A1 with four miscarriages ending before the first trimester. Her past medical history is significant for obesity, depression, hypothyroidism and tobacco disc degeneration. During her prenatal care she was found to have anti-nuclear antibody and natural killer cell activity treated with aspirin, folic acid, and vitamin D. A 12-week ultrasound performed at St. Helier reports a normal abdomen, heart, skin, 1.8m with a very low adjusted birth weight. The twenty-three week anomaly scan showed fetal growth within normal limits in all categories and demonstrated a healthy fetal heart. Both sonographers noted that exam was difficult to the patient’s body habitus (BMI 33.3). All three of the patient’s ultrasounds were performed by sonographers. The remainder of the pregnancy was uneventful except for an episode of bleeding per vagum in 18 weeks that resolved spontaneously. She was scheduled for a cesarean section due to back problems.

A baby boy was born via an elective Caesarean Section at 30+1 weeks at St. Helier’s Hospital. When spinal anaesthesia was initiated the fetal heart rate dropped to 55/min and the decision was made to expedite the delivery. At the time of birth the baby exhibited signs of poor respiratory effort and required resuscitation involving inflation breaths followed by ventilation. The baby was started on 50% Oxygen via CPAP and transferred to the Neonatal Unit 26 minutes after birth. On examination by the paediatric registrar the baby was tachypnoeic at 80 breaths per minute but demonstrated a good air entry bilaterally. The chest X-ray showed a left sided congenital diaphragmatic hernia and the patient was subsequently intubated for ventilation transfer to St. George’s Hospital and surgical management. A left-sided diaphragmatic hernia patch repair was successfully performed without complication 3 days later. The baby’s Neonatal Intensive Care was complicated with respiratory distress syndrome, suspected sepsis and persistent pulmonary hypertension. Despite his dramatic beginning the baby is currently healthy and without breathing problems and is being followed by the respiratory team.

Discussion:
In our case the baby’s CDH went undetected and subsequent emergency post-natal surgery was required. Prenatal detection is possible but can be missed, sensitivity increases if the patient exhibits other anomalies and advancing gestation with a mean age of diagnosis at 24 weeks. (3) Antenatal ultrasound can identify >70% of cases. (4) Severity of the hernia is also directly proportional to mortality. (17) Once the diagnosis has been confirmed then management is based on the severity of the hernia. Larger herniation and right-sided defects are associated with greater lung hypoplasia and increased risk of death. (6) Diaphragmatic defects cannot be reliably measured prenatally. However, herniation and fetal lung volume measurements have become reliable predictors of survival. The degree of liver herniation has been indicated in retrospective studies as a significant predictor of prognosis. (15) For babies born with a proven CDH there is significant debate about delivery time and mode, the use of extracorporeal membrane oxygenation (ECMO) and post-surgical treatment. (6) In cases with mild to moderate CDH detected prenatally, the current treatment is similar to what we have performed; aggressive respiratory support including mechanical ventilation, medical stabilization and surgical closure of the defect. The CDH Study Group found that specialized centres that commonly treat the hernia have survival rates nearing 90%. (7) Patch repairs use synthetic patches (Gore-Loft, Marlex® or Permacol® for larger defects and standard non-absorbable sutures for smaller herniation). (8) Post-operative repair patient also exhibited persistent pulmonary hypertension, which is one of the most common and serious post-surgical complications. However like the majority of properly managed CDH patients he has recovered and is expected to have a normalization of pulmonary pressures. (5, 6, 17)

For isolated severe CDH detected in utero, interventional therapy is only available at a few centres worldwide. There has been important work done with prenatal detection and treatment with Fetal Endoluminal Tracheal Occlusion (FETO), a technique that attempts to decrease lung hypoplasia caused by CDH. A tiny balloon is passed percutaneously into the fetal trachea and inflated. This obstructs the outflow of lung fluid and creates an environment where they can better develop. (1, 5, 8) The Eurofetus group report in 24 poor prognosis fetuses showed increased survival to discharge rates at 50% (11) And the FETO Consortium looked at 210 cases managed by FETO versus expectant management and saw an increase in survival from 24% to 49% with intervention. They concluded that despite a high incidence of PPROM that intervention with FETO did increase survival in patients with poor prognosis (12) Another study showed that FETO resulted in significant improvement in lung size and pulmonary vascularity when compared to neonates who were expectantly managed. (13) However, some articles cite that all randomized trials comparing prenatal intervention to standard intervention show no benefit in less severe cases. (14)

Conclusion:
The early antenatal detection is very important, as it is the key to help in the obstetrician management as well as newborn care. Antenatal diagnosis allows prenatal management (open correction of the hernia in the past and presently reversible fetoscopic trachael obstruction) that may be indicated in cases with severe lung hypoplasia. (13) (18) The diagnosis of CDH is dependent on prenatal imaging and nuchal translucency screening. (5) Treatment after birth may require all the aforementioned interventions to include extracorporeal membrane oxygenation prior to surgical correction. This also allows for in utero transfer to more specialised units where immediate intensive care for the newborn can be offered. It is therefore critical that intensive training be offered to all sonographers and training doctors, in order to maximise the detection rates of congenital diaphragmatic hernia and provide patients with the best care possible.

It is also crucial to have a comprehensive evaluation, including imaging and (high-resolution) genetic studies, should be carried out. The primary purpose of this evaluation is to rule out associated anomalies and to assess the severity of pulmonary hypoplasia in order to offer parents an individualized prognosis. The latter can be done on the basis of the dimensions of the lung, its vasculatization and liver position. Further multidisciplinary consultation should address the neonatal issues as well as later morbidities. Based on this complete evaluation and after extensive counselling, patients should make an informed choice out of the available management options which also include fetal therapy for more severe cases in a number of centres spread over the world.

References: