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Prediction of outcome in isolated congenital diaphragmatic hernia and its consequences for fetal therapy

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Congenital diaphragmatic hernia (CDH) can be diagnosed in the prenatal period either as part of other anomalies or as an isolated birth defect. The clinical impact of this surgically correctable anatomical defect lies in its impairment of lung development. Currently, up to 30% of babies with isolated CDH die from the consequences of lung hypoplasia and/or pulmonary hypertension. Antenatal prediction of outcome essentially relies on the measurement of lung development by the

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so-called lung area to head circumference ratio (LHR). By expressing observed LHR as a proportion of what is normally expected (O/E LHR) at a certain time point in gestation, a prediction of outcome can be made. When O/E LHR is less than 25% of the normal, postnatal death is very likely. In these cases, an antenatal intervention that can improve lung development is currently offered. Currently, this is done by percutaneous fetal endoscopic tracheal occlusion (FETO) with a balloon at 26–28 weeks, and reversal of occlusion at 34 weeks. The feasibility and safety of percutaneous FETO have been established and the procedure seems to improve outcome in severe CDH. The lung response to, and outcome after, FETO depend on pre-existing lung size respectively gestational age at birth. Prenatal decision making can therefore be stratified according to measured lung size.

Key words: congenital diaphragmatic hernia; Fetal surgery; Pulmonary hypoplasia; Tracheal occlusion.

INTRODUCTION

Congenital diaphragmatic hernia (CDH) occurs sporadically; the incidence is 1/2500 to 1/5000 of newborns, depending on whether stillbirths are included. Eighty-four per cent of lesions are left sided (LCDH), 13% right sided (RCDH) and 2% bilateral. Complete agenesis, herniation of the central tendonous part and eventration (thinning of the muscle) are rare manifestations. Less than 2% of cases are familial. The recurrence rate of isolated CDH is 2%. Associated anomalies — an independent predictor of neonatal death — occur in around 40% of cases, with $<$15% of babies surviving in this group.[1,2]

One theory for the cause of CDH is that the pleuroperitoneal canals fail to close at around 9 weeks. By approximately 10–12 weeks, the viscera return to the abdominal cavity and start to herniate into the thorax, where they compete for space with the lungs. It is also possible that the condition is primarily pulmonary. However, we refer to the literature for this discussion and additional concepts on the embryology, molecular and genetic mechanisms behind this disease.[3]

In CDH, the fetal lungs have a reduced number of arteries and of conducting and functional airways. There are fewer alveoli, thickened alveolar walls, increased interstitial tissue and a markedly diminished alveolar air space and gas-exchange surface area. As vascular development parallels that of the airways, in CDH the pulmonary vasculature is also abnormal. There are a reduced number of vessels, adventitial thickening, medial hyperplasia and peripheral extension of the muscle layer into the smaller intraacinary arterioles. Both lungs are affected, the ipsilateral lung more than the contralateral one, and the hypoplasia is progressive beyond 30 weeks.[4] It is only in the postnatal period that these morphological changes become functional, causing ventilatory insufficiency and pulmonary hypertension. Survivors might additionally suffer from variable degrees of pulmonary, gastrointestinal (gastro-oesophagal reflux and feeding problems), orthopaedic, hearing and neurodevelopmental problems.[5]

PRENATAL DIAGNOSIS

Ultrasound screening should result in making the diagnosis of CDH in the prenatal period. The diaphragm can be visualized with high-resolution equipment in the first trimester. In its absence, abdominal organs are visualized when cross-sectioning the thorax to obtain a four-chamber view (Figure 1). Left-sided CDH is most common