

Editorial

Genetic Pathways, Antenatal Care and Congenital Diaphragmatic Hernia: Are We There Yet?

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Congenital diaphragmatic hernia (CDH) remains a challenging diagnostic and treatment anatomical illness for clinicians. Pediatric pulmonologists, cardiothoracic and general surgeons and pediatric intensivists are confronted with these cases. CDH represents a group of congenital defects in the structural integrity of the diaphragm, affecting approximately 1 in 3000 births [1]. While it appears to be an anatomical malformation, CDH has a very complex pathophysiology. Despite the extensive research performed over past 4 decades and the advances in perinatal ultrasound, the early diagnosis is still not available and the mortality rate remains quite high, ranging from 30 to 50% [1].

When identified as a system illness, it is evident that CDH is not an isolated event, and not just a "hole in the diaphragm", but indeed a complex syndrome that includes pulmonary hypoplasia, lung immaturity and left heart hypoplasia that leads to persistent pulmonary hypertension of the newborn [2]. This is particularly important for clinicians dealing with these patients in the acute and critical care settings. The economical impact of CDH can be significant in many patients.

The pathophysiology of CDH has been studied thoroughly in animal models and yet, there are several areas that remain unclear. An exclusive mechanism causing the primary anatomical has not been found yet. In some rat studies, CDH has been noted to be more frequent in the offspring of rats fed with a vitamin A deficient diet [3]. In addition, specific gene deletions have been reported to cause diaphragmatic defects in mice [4].

Although initially CDH is a life-threatening condition in many patients but, those patients who survive, it can become a long-term clinical condition. CDH survivors experience ongoing medical morbidity, including pulmonary nutritional and developmental delays [5]. As the postnatal management of patients with CDH is often complicated by long primary

hospitalizations, several authors have suggested that these patients have significant learning difficulties and low IQs that may interfere with their normal social life and employment status [6].

In this issue of *Current Respiratory Medicine Reviews*, Bourbon provides a historical perspective of the discovery of this elusive illness, as well as the pathophysiology and antenatal therapy currently available and an up-to-date information on clinical research [7]. The author identifies the needs for clinicians dealing with these complex patients. As noted by Bourbon and others, CDH is a genetic disease that still requires significant research in an attempt to identify and promptly stop the anomaly [4]. Recognizing the diaphragm's normal genetic pathways may lead to advanced therapeutic options. However, despite all these advances in diagnosis we "are not there yet!"

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