CHAPTER 36

CONGENITAL DIAPHRAGMATIC HERNIA

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INTRODUCTION

Congenital diaphragmatic hernia (CDH), which is a developmental defect of the diaphragm, causes abdominal viscera to herniate to the chest cavity. These defects are correctable with surgical intervention. Nevertheless; if the herniation begins during in-utero period, it disrupts respiratory system development. Thus, pulmonary hypoplasia and pulmonary hypertension can be originated. A high proportion of CDH patients also have additional abnormalities related to this defect, such as chromosomal abnormalities, major structural malformations in other organ systems, and/or single-gene disorders.

PATHOGENESIS

Defect in normal pleuroperitoneal fold closure between the fourth to tenth weeks of the post-fertilization period is the main pathologic mechanism behind the congenital diaphragmatic hernia. This condition disrupts normal lung development process and has substantial adverse health outcomes, including [1-7]:

- Decrease amount of bronchiolar branching
- Formation of truncated and over-muscularized pulmonary arterial tree, which leads to decrease and constriction in the lumen of pulmonary vessels, remodeling of vascular structure, and constriction of vessels because of adjusted vasoreactivity. A decrease in lumen cross-sectional area leads to an increase in pulmonary resistance.
- Diminished pulmonary mass induces the occurrence of postnatal pulmonary hypoplasia.
- Disrupted surfactant system of the respiratory system disrupts oxygen and carbon dioxide exchange and increases resistance in the lungs.
- Cardiac structures can be hypoplastic ipsilaterally, which can be fatal.

The consequences and adverse outcomes of the defect differ in an affected fetus according to the time when the fetus developed the visceral hernia during the gestational period.

The etiology of this defect in normal diaphragmatic closure is uncertain. Hereditary and/or environmental exposures may be the reason for the abnormal differentiation of mesenchymal cells during the development of the diaphragm and other somatic structures [8-10]. Since the majority of CDH cases occur sporadically with no etiology could be found, there are some reports states that hereditary transmitted cases can be transmitted via autosomal recessive, autosomal dominant, and X-linked patterns [11-14]. Vitamin A deficiency; thalidomide, anticonvulsants, quinine exposures are the possible environmental etiologies that can be related to CDH [24].

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