Clinical aspects of congenital defects and anomalies of lung development

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ABSTRACT. The article presents a comprehensive analysis of contemporary approaches to the diagnosis of congenital lung malformations and anomalies (CLMAs), which account for 5-18 % of all thoracic organ anomalies. Epidemiological aspects, classification, and morphological features of the most common forms are discussed, including congenital pulmonary airway malformation, pulmonary sequestration, congenital lobar emphysema, bronchogenic cysts, as well as rare anatomical variants such as tracheal bronchus or accessory lung. Attention is drawn to the phenomenon of hybrid lesions that combine characteristics of multiple nosologies. The significance of prenatal diagnostics, particularly ultrasound and MRI, is highlighted as essential for timely detection of pathologies and planning the management of newborns.

Considerable attention is given to the genetic factors and pathogenesis of CLMAs, including the role of mutations in the FGF10 and TBX4 genes, as well as associations with primary ciliary dyskinesia. It is shown that malformation development has a polyetiological nature, combining genetic, embryological, and exogenous factors. Clinical manifestations range from asymptomatic cases to severe respiratory failure, and pathognomonic features of individual forms are identified. It is summarized that diagnosis of congenital lung malformations relies on a combination of clinical data, the results of instrumental methods (CT, MRI, bronchoscopy), histological studies, and genetic testing. The findings are crucial for timely selection of optimal therapeutic strategies, prevention of infectious complications, and reduction of the risk of malignant transformation.

KEY WORDS: lungs, congenital malformations, pulmonary sequestration, bronchogenic cysts, prenatal diagnostics, primary ciliary dyskinesia.

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