Review Nat Rev Dis Primers. 2023 Nov 2;9(1):60. doi: 10.1038/s41572-023-00470-1.

Congenital lung malformations

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PMID: 37919294 DOI: 10.1038/s41572-023-00470-1

Abstract

Congenital lung malformations (CLMs) are rare developmental anomalies of the lung, including congenital pulmonary airway malformations (CPAM), bronchopulmonary sequestration, congenital lobar overinflation, bronchogenic cyst and isolated congenital bronchial atresia. CLMs occur in 4 out of 10,000 live births. Postnatal presentation ranges from an asymptomatic infant to respiratory failure. CLMs are typically diagnosed with antenatal ultrasonography and confirmed by chest CT angiography in the first few months of life. Although surgical treatment is the gold standard for symptomatic CLMs, a consensus on asymptomatic cases has not been reached. Resection, either thoracoscopically or through thoracotomy, minimizes the risk of local morbidity, including recurrent infections and pneumothorax, and avoids the risk of malignancies that have been associated with CPAM, bronchopulmonary sequestration and bronchogenic cyst. However, some surgeons suggest expectant management as the incidence of adverse outcomes, including malignancy, remains unknown. In either case, a planned follow-up and a proper transition to adult care are needed. The biological mechanisms through which some CLMs may trigger malignant transformation are under investigation. KRAS has already been confirmed to be somatically mutated in CPAM and other genetic susceptibilities linked to tumour development have been explored. By summarizing current progress in CLM diagnosis, management and molecular understanding we hope to highlight open questions that require urgent attention.

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