Open Peer Review on Qeios

Interstitial lung disease due to ABCA3 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Interstitial</u> <u>lung disease due to ABCA3 deficiency</u>. ORPHA:440402

Interstitial lung disease due to ABCA3 deficiency is a rare genetic respiratory disease characterized by a variable clinical outcome ranging from a fatal respiratory distress syndrome in the neonatal period to chronic interstitial lung disease developing in infancy or childhood with chronic cough, rapid breathing, shortness of breath and recurrent pulmonary infections. Clinical manifestations of respiratory failure include grunting, intercostal retractions, nasal flaring, cyanosis, and progressive dyspnea.