

Opis choroby *

Definicja

Primary pulmonary hypoplasia is a rare, isolated, genetic developmental defect during embryogenesis characterized by congenital malformation of pulmonary parenchyma with absence of other anomalies. Neonatally patients present with decreased breath sounds, small lung volume and severe respiratory distress that is not responsive to aggressive treatment (including surfactant instillation/ mechanical respiratory support). It is usually not compatible with life.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

2257

Kod OMIM

265430

Kod ICD10

Q33.6

Kod ICD11

LA75.2

*Źródło

orphanet