Opis choroby *

Definicja

A rare syndromic esophageal malformation characterized by severe congenital brachyesophagus with midline diaphragmatic hernia and secondary intrathoracic stomach, and vertebral anomalies (in particular rachischisis of the cervical/thoracic spine). Additional reported manifestations include intrauterine growth restriction, short neck, intestinal malrotation, herniation of other abdominal organs, and cleft lip, among others. The condition is mostly fatal in the neonatal or early infantile period.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Serpentine-like syndrome
Zespół przypominający ułożenie węża/serpentynę

Kod ORPHA
514352

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet