Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by a large omphalocele containing liver and small intestine, diaphragmatic hernia, cardiovascular anomalies (e. g. aortic coarctation), variable limb malformations (including radioulnar synostosis, agenesis of the radius and/or thumb, generalized syndactyly, and numerical reduction of toes), and dysmorphic facial features. Additional reported manifestations are unilateral absence of umbilical artery, intestinal malrotation, hypoplastic ovaries, and unilateral renal agenesis, among others. The condition is mostly fatal in the neonatal period.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Gershoni-Baruch syndrome
Gershoni-Baruch syndrome

Kod ORPHA
496693

Kod OMIM
609545

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet