

Opis choroby *

Definicja

Holoprosencephaly-craniosynostosis syndrome is a rare developmental defect during embryogenesis syndrome characterized by the association of primary craniosynostosis (usually involving the coronal and metopic sutures) with holoprosencephaly (ranging from alobar to, most commonly, semilobar) and various skeletal anomalies (typically, hand and feet anomalies including fifth digit clinodactyly, hypoplastic phalanges and cone-shaped epiphyses, small vertebral bodies, scoliosis, coxa valga and/or flexion deformities of hips). Craniofacial asymmetry, microcephaly, brachy/plagiocephaly, short stature and psychomotor delay are additional common features.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Camero-Litwania-Cohen syndrome Zespół Camero, Litwania i Cohena Zespół Genoa Genoa syndrome

Kod ORPHA 2163	Kod OMIM 601370	Kod ICD10 Q04.2
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Kod ICD11
LD20.3

[*Źródło](#)

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