Opis choroby *

Definicja

A rare developmental defect during embryogenesis mainly characterized by severe intellectual disability, short stature, hypogonadism, and distinct facial dysmorphism (including trigonocephaly, prominent forehead, asymmetric and flat face, hypertelorism, epicanthus, downslanting palpebral fissures, ptosis, low-set angulated ears, small mouth, high-arched/cleft palate crowded teeth, microretrognathia), as well as slender hands and/or feet. Variable additional features may include pterygia, hypoplastic nipples, cardiac anomaly, distal muscular wasting, limb contractures, skeletal anomalies (e.g. scoliosis, pectus excavatum, bilateral clubfeet), hypothyroidism, seizures, and cerebral anomalies. Puberty may be delayed.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Haspeslagh-Fryns-Muelenaere syndrome
Zespół Haspeslagha, Frynsa i Muelenaere'a

Kod ORPHA
2994

Kod OMIM
177980

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet