Opis choroby *

Definicja

A rare, genetic, developmental defect during embryogenesis malformation syndrome characterized by severe postnatal growth retardation, craniofacial dysmorphism, which includes a progeroid facial appearance, brachycephaly with hypoplasia of the frontal and parietal tubers and a flat occipital area, narrow forehead, prominent glabella, small orbit, slight bilateral exophthalmos, straight nose, hypoplastic cheekbones, long philtrum and thin lips, skeletal abnormalities (i.e. micromelia, brachydactyly, and severe short stature with short limbs), normal intelligence, Pelger-Huët anomaly of leukocytes, loose skin with decreased tissue turgor, and bilateral optic atrophy with loss of color vision and visual acuity. Recurrent liver failure triggered by fever has been occasionally reported. Radiographs may evidence delayed bone age, late ossification and/or osteoporosis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SOPH syndrome
Zespół SOPH

Kod ORPHA
391677

Kod OMIM
614800

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet