

Zespół niski wzrost-atrofia nerwu wzrokowego- anomalia Pelgera i Huëta

Kod Orpha: 391677 Kod OMIM: 614800

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](#)

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Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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