Opis choroby *

Definicja

A rare inherited cancer-predisposing syndrome characterized by early-onset hepatocellular carcinoma, genomic instability, and progeroid features, such as short stature, low body weight, muscular atrophy, lipodystrophy, bilateral cataracts, and premature hair graying. Dysmorphic craniofacial features include triangular face, small, deep-set eyes, and micrognathia. Kyphoscoliosis, sloping shoulders, mild pectus excavatum, bilateral contractures of the elbows and fingers, bilateral clinodactyly, and pes planus have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Ruijs-Aalfs syndrome
Zespół Ruijsa i Aalfsa

Kod ORPHA
435953

Kod OMIM
616200

Kod ICD10
C22.0

Kod ICD11
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*Źródło

orphanet