

Opis choroby *

Definicja

King-Denborough syndrome is a rare genetic non-dystrophic myopathy characterized by the triad of congenital myopathy, dysmorphic features and susceptibility to malignant hyperthermia. Patients present with a wide phenotypic range, including delayed motor development, muscle weakness and fatigability, ptosis and facies myopathica (with or without creatine kinase elevations), skeletal abnormalities (e.g. short stature, scoliosis, kyphosis, lumbar lordosis and pectus carinatum/excavatum), mild dysmorphic facial features (e.g. hypertelorism, down-slanting palpebral fissures, epicanthic folds, low set ears, micrognathia), webbing of the neck, cryptorchidism, and a susceptibility to malignant hyperthermia and/or rhabdomyolysis due to intensive physical strain, viral infection or statin use.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Koussef-Nichols syndrome Zespół Koussefa i Nicholisa

Kod ORPHA	Kod OMIM	Kod ICD10
99741	619542	G71.2

Kod ICD11
8C72.Y

*Źródło

orphanet