

Opis choroby *

Definicja

Marfanoid habitus-inguinal hernia-advanced bone age syndrome is a very rare developmental defect with connective tissue involvement disorder characterized by tall stature, inguinal hernia, facial dysmorphism (including a long, triangular face, prominent forehead, telecanthus, downslanting palpebral fissures, bilateral ptosis, everted lower eyelids, large ears, long nose, full, everted vermilions, narrow and high arched palate, dental crowding), and radiologic evidence of advanced bone age. Additional manifestations include hyperextensible joints, long digits, mild muscle weakness, myopia, and foot deformities (i.e. hallux valgus, talipes equinovarus).

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

314041

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet