

Opis choroby *

Definicja

Cryptorchidism-arachnodactyly-intellectual disability syndrome is a rare, multiple congenital anomalies syndrome characterized by psychomotor delay, severe intellectual deficit, severe muscle hypoplasia (with absence of subcutaneous fatty tissue), generalized contractures, craniofacial dysmorphic features (dolichocephaly, esotropia, ears of unequal size, high palate), chest and spinal deformities (i.e. sternum shifted to side, kyphoscoliosis), pulmonary anomalies (unilateral hypoplastic bronchial system), arachnodactyly, and genital abnormalities (cryptorchidism, hypospadias, testicular agenesis). Repeated respiratory tract infections and atelectasis are also associated. There have been no further descriptions in the literature since 1970.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Van Benthem-Driessen-Hanveld syndrome
Zespół Van Benthem, Driessena i Hanvelda

Kod ORPHA

1548

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet