

Opis choroby *

Definicja

Ring chromosome 7 syndrome is a rare chromosomal anomaly syndrome, with highly variable phenotype, principally characterized by growth failure, short stature, intellectual disability, dermatological abnormalities (nevus flammeus, dark pigmented nevi, café-au-lait spots), microcephaly and facial dysmorphism (incl. facial asymmetry, small ears, abnormal palpebral fissures, ptosis, epicanthic folds, hyper/hypotelorism). Additional reported features include convulsions, cleft lip and palate, clinodactyly, kyphoscoliosis and genital anomalies (i.e. cryptorchidism, hypospadias, micropenis).

Dane

Klasyfikacja

Zespół wad wrodzonych Ring 7

Synonimy

Ring chromosome 7

Kod ORPHA

1449

Kod OMIM

-

Kod ICD10

Q93.2

Kod ICD11

-

*Źródło

orphanet