

Opis choroby *

Definicja

Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome is characterised by the association of spondylometaphyseal dysplasia (marked by platyspondyly, shortening of the tubular bones and progressive metaphyseal irregularity and cupping), with postnatal growth retardation and progressive visual impairment due to cone-rod dystrophy. So far, it has been described in eight individuals. Transmission appears to be autosomal recessive.

Dane

Klasyfikacja

Choroba

Synonimy

SMD-CRD

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Kod ORPHA

85167

Kod OMIM

608940

Kod ICD10

Q77.8

Kod ICD11

LD24.4

*Źródło

orphanet