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
SMD-CRD

Kod ORPHA
85167

Kod OMIM
608940

Kod ICD10
Q77.8

Kod ICD11
LD24.4

---

*Źródło

orphanet