

Opis choroby *

Definicja

Mandibuloacral dysplasia (MAD) is a rare genetic bone disorder characterized by growth delay, postnatal development of craniofacial anomalies including mandibular hypoplasia, progressive acral osteolysis, mottled or patchy pigmentation, skin atrophy, and partial or generalized lipodystrophy.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych MAD	MAD

Kod ORPHA	Kod OMIM	Kod ICD10
2457	248370	Q87.5

Kod ICD11
LD27.6Z

*Źródło

orphanet