

Opis choroby *

Definicja

Metaphyseal dysplasia-maxillary hypoplasia-brachydacty syndrome is characterized by metaphyseal dysplasia associated with short stature and facial dysmorphism (a beaked nose, short philtrum, thin lips, maxillary hypoplasia, dystrophic yellowish teeth) and acral anomalies (short fifth metacarpals and/or short middle phalanges of fingers two and five). It has been described in several members spanning four generations of a French-Canadian family. The syndrome is likely to be transmitted as an autosomal dominant trait.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2504

Kod OMIM

156510

Kod ICD10

Q77.8

Kod ICD11

LD24.7

*Źródło

orphanet