

Opis choroby *

Definicja

Singleton-Merten dysplasia is characterized by dental dysplasia, progressive calcification of the thoracic aorta with stenosis, osteoporosis and expansion of the marrow cavities in hand bones. Additional features included generalized muscle weakness and atrophy, and chronic psoriasiform skin eruptions. It has been reported in four unrelated patients (male and female) and in a family with multiple affected members (male).

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Singleton-Merten syndrome Zespół Singletona i Mertena

Kod ORPHA	Kod OMIM	Kod ICD10
85191	616298	Q78.8

Kod ICD11
LD24.KY

*Źródło

orphanet