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