Opis choroby *

Definicja

A rare hyaline fibromatosis syndrome characterized by papulo-nodular skin lesions (especially around the head and neck), soft tissue masses, gingival hypertrophy, joint contractures, and osteolytic bone lesions in variable degrees. Joint contractures may cripple patients and delay normal motor development if occuring in infancy. Severe gingival hyperplasia can interfere with eating and delay dentition. Histopathology analysis of involved tissues reveals cords of spindle-shaped cells embedded in an amorphous, hyaline material.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Murray-Puretic-Drescher syndrome
Zespół Murray, Puretica i Dreschera
Zespół Puretica
Puretic syndrome

Kod ORPHA
2028

Kod OMIM
228600

Kod ICD10
M72.8

Kod ICD11
EE6Y

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*Źródło

orphanet