

Dziecięca hialinoza uogólniona

Kod Orpha: 2176 Kod OMIM: 228600

Opis choroby *

Definicja

Infantile systemic hyalinosis (ISH) is a very rare disorder belonging to the heterogeneous group of genetic fibromatoses and is characterized by progressive joint contractures, skin abnormalities, severe chronic pain and widespread deposition of hyaline material in many tissues such as the skin, skeletal muscle, cardiac muscle, gastrointestinal tract, lymph nodes, spleen, thyroid, and adrenal glands.

Dane

Klasyfikacja

Podtyp kliniczny

Kod ORPHA

2176

Kod OMIM

228600

Kod ICD10

E78.8

Kod ICD11

EE6Y

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.