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

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*Źródło

orphanet