

Opis choroby *

Definicja

A rare genetic lipodystrophy characterized by loss of subcutaneous fat layers on the limbs, lipodystrophy in the face and trunk and scleroderma-like skin disorders (thickened skin on the palms and soles and skin pigment changes on the limbs and trunk). Additional clinical signs include joint contractures, reduced relative body weight, a bird-like facial appearance with a beaked nose, micrognathia and insulin-resistant diabetes mellitus.

Dane

Klasyfikacja

Choroba

Synonimy

Combined insulin, insulin-like growth factor 1 (IGF1) and epidermal growth factor (EGF) deficiency
Mieszany Niedobór insuliny, insulinopodobnego czynnika wzrostu (IGF1) i czynnika wzrostu naskórka (EGF)
Zespół Hoepffnera, Dreyera i Reimersa
Zespół podobny do zespołu Wernera spowodowany mieszanym niedoborem czynnika wzrostu
Hoepffner-Dreyer-Reimers syndrome
Werner-like syndrome due to combined growth factor deficiency

Kod ORPHA

1979

Kod OMIM

233805

Kod ICD10

E88.1

Kod ICD11

LD27.6Z

*Źródło

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