

Zespół sztywnej skóry

Kod Orpha: 2833 Kod OMIM: 228020

Opis choroby *

Definicja

Stiff skin syndrome is a rare, slowly progressive cutaneous disease characterized by rock-hard skin bound firmly to the underlying tissues (mainly on the shoulders, lower back, buttocks and thighs), mild hypertrichosis and hyperpigmentation overlying the affected areas of skin, as well as limited joint mobility (mainly of large joints) with flexion contractures. Cutaneous nodules, affecting mostly distal interphalangeal joints, as well as extracutaneous manifestations, including diffuse entrapment neuropathy, scoliosis, a tiptoe gait and a narrow thorax, may be associated. Restrictive pulmonary changes, muscle weakness, short stature and growth delay have also been reported. No vascular hyperreactivity, immunologic abnormalities nor visceral, muscular or bone involvement has been described.

Dane

Klasyfikacja

Choroba

Kod ORPHA
2833

Kod OMIM
228020

Kod ICD10
L98.8

Kod ICD11
EE6Y

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Orphanet - internetowa baza danych dotyczących rzadkich chorób i sierochych leków. ©INSERM 1999 -
Dostępna na stronie www.orphanet.pl