

## 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

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### \*Źródło

orphanet