DISEASE:
Stiff skin syndrome

NAME:	Stiff skin syndrome
DESCRIPTION:	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.
ORPHACODE:	2833
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	FBN1
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