Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome. ORPHA:221043

Hereditary fibrosing poikiloderma-tendon contractures-myopathy-pulmonary fibrosis syndrome is a rare, genetic, hereditary poikiloderma syndrome characterized by early-onset poikiloderma (mainly on the face), hypotrichosis, hypohidrosis, muscle and tendon contractures with varus foot deformity, progressive proximal and distal muscle weakness in all extremities, and progressive pulmonary fibrosis. Mild lymphedema of the extremities, growth retardation, liver impairment, exocrine pancreatic insufficiency and hematologic abnormalities are additional variable features.