Poikiloderma with neutropenia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Poikiloderma with neutropenia. ORPHA:221046

Poikiloderma with neutropenia is a rare, genetic hereditary poikiloderma disorder characterized by early-onset poikiloderma (which typically begins in the extremities, progresses centripetally and eventually involves the trunk, face and ears) associated with chronic neutropenia, recurrent infections, pachyonychia and palmoplantar keratoderma. Growth and/or develomental delay and hepato- and/or splenomegaly are additional reported features.