DISEASE:
Acroosteolysis-keloid-like lesions-premature aging syndrome

NAME:	Acroosteolysis-keloid-like lesions-premature aging syndrome
DESCRIPTION:	A rare, genetic, progeroid syndrome disorder characterized by a prematurely aged appearance (including lipoatrophy, thin, translucent skin, sparse, thin hair, and skeletal muscle atrophy), delayed tooth eruption, keloid-like lesions on pressure regions, and skeletal abnormalities including marked acroosteolysis, brachydactyly with small hands and feet, kyphoscoliosis, osteopenia, and progressive joint contractures in the fingers and toes. Craniofacial features include a thin calvarium, delayed closure of the anterior fontanel, flat occiput, shallow orbits, malar hypoplasia and narrow nose.
ORPHACODE:	363665
SYNONYMS:	Premature aging syndrome, Penttinen type
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PDGFRB
CREATED:	13 May 2019 - 01:02
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