Acroosteolysis-keloid-like lesions-premature aging syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acroosteolysis-keloid-like lesions-premature aging syndrome. ORPHA:363665

Acroosteolysis-keloid-like lesions-premature aging syndrome is 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.