Open Peer Review on Qeios

AGel amyloidosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>AGel</u> <u>amyloidosis</u>. ORPHA:85448

AGel amyloidosis is a rare, systemic amyloidosis characterized by a triad of ophthalmologic, neurologic and dermatologic findings due to the deposition of gelsolin amyloid fibrils in these tissues. Clinical manifestations include corneal lattice dystrophy, cranial neuropathy, especially affecting the facial nerve, bulbar signs, cutis laxa, increased skin fragility, and less commonly peripheral neuropathy and renal failure.