Inclusion body myopathy with Paget disease of bone and frontotemporal dementia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Inclusion body myopathy with Paget disease of bone and frontotemporal dementia.
ORPHA:52430

Inclusion body myopathy with Paget disease of bone and frontotemporal dementia (IBMPFD) is a multisystem degenerative genetic disorder characterized by adult-onset proximal and distal muscle weakness (clinically resembling limb-girdle muscular dystrophy; see this term); early-onset Paget disease of bone (see this term), manifesting with bone pain, deformity and enlargement of the long-bones; and premature frontotemporal dementia (see this term), manifesting first with dysnomia, dyscalculia and comprehension deficits followed by progressive aphasia, alexia, and agraphia. As the disease progresses, muscle weakness begins to affect the other limbs and respiratory muscles, ultimately resulting in respiratory or cardiac failure.