DISEASE:
Inclusion body myopathy with Paget disease of bone and frontotemporal dementia

NAME:	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
DESCRIPTION:	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.
ORPHACODE:	52430
SYNONYMS:	IBMPFD Limb-girdle muscular dystrophy with Paget disease of bone Pagetoid amyotrophic lateral sclerosis Pagetoid neuroskeletal syndrome

XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM
ANALYTE(S):	VCP HNRNPA2B1 HNRNPA1
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