

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Choroba	IBMPFD Dystrofia obręczowo-kończynowa z chorobą Pageta IBMPFD Pagetoidalne stwardnienie zanikowe boczne Pagetoidalny zespół neuro-szkieletowy Pagetoidalny zespół nerwowo-szkieletowy Limb-girdle muscular dystrophy with Paget disease of bone Pagetoid amyotrophic lateral sclerosis Pagetoid neuroskeletal syndrome

Kod ORPHA
52430

Kod OMIM
615424

Kod ICD10
G71.8

Kod ICD11
4A41.21

*Źródło

orphanet