

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

Choroba

Synonimy

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](#)

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