

Opis choroby *

Definicja

Nasu-Hakola disease (NHD), also referred to as polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL), is a rare inherited leukodystrophy characterized by progressive presenile dementia associated with recurrent bone fractures due to polycystic osseous lesions of the lower and upper extremities.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

NHD

NHD

pLOSL

pLO-SL

Wielotorbielowata tłuszczowo-błoniasta

osteodysplazja ze stwardniającą

leukoencefalopatią

PLO-SL

PLOSL

Polycystic lipomembranous osteodysplasia with
sclerosing leukoencephalopathy

Kod ORPHA

2770

Kod OMIM

221770

Kod ICD10

G31.8

Kod ICD11

8A44.3

*Źródło

orphanet