Opis choroby *

Definicja

A rare genetic neurological disorder characterized by pediatric onset of calcifying leukoencephalopathy and skeletal dysplasia. Reported structural brain abnormalities include agenesis of corpus callosum, ventriculomegaly, congenital hydrocephalus, pontocerebellar hypoplasia, periventricular calcifications, Dandy-Walker malformation and absence of microglia. Characteristic skeletal features include increased bone mineral density (reported in skull, pelvic bone and vertebrae), platyspondyly, and under-modeling of tubular bones with widened/radiolucent metaphysis and constricted/sclerotic diaphysis.

Dane

Klasyfikacja
Choroba

Kod ORPHA
556985

Kod OMIM
618476

Kod ICD10
G93.4

Kod ICD11
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*Źródło

orphanet