

Mikrohydranencefalia zależna od NDE1

Kod Orpha: 443162 Kod OMIM: 605013

Opis choroby *

Definicja

NDE1-related microhydranencephaly is a rare, hereditary syndrome with a central nervous system malformation as major feature characterized by extreme microcephaly and growth restriction, severe motor delay and mental retardation, and typical radiological findings of gross dilation of the ventricles resulting from the absence (or severe delay in the development) of cerebral hemispheres, hypoplasia of the corpus callosum, cerebellum, and brainstem. Associated features are thin bones and scalp rugae.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

MHAC
MHAC

Kod ORPHA

443162

Kod OMIM

605013

Kod ICD10

Q04.8

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.