

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	Synonimy
Zespół wad wrodzonych MHAC	MHAC
	MHAC

Kod ORPHA	Kod OMIM	Kod ICD10
443162	605013	Q04.8

Kod ICD11
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*Źródło

orphanet