Opis choroby *

Definicja

A rare genetic central nervous system malformation characterized by dysplasia of the superior cerebellum (especially the vermis), brainstem asymmetry, dysplasia of the basal ganglia, and cortical irregularities with asymmetric abnormalities in gyral size and orientation, as well as varying sulcal depth, but without lissencephaly, pachygyria, or polymicrogyria. Clinically, patients present global developmental delay with motor development usually being more affected that speech. Variable features are abnormal eye movements including oculomotor apraxia, strabismus, seizures, and behavioral problems.

Dane

Klasyfikacja
Choroba

Synonimy
Brain stem asymmetry-superior cerebellar and basal ganglia dysplasia syndrome
Zespół asymetrii pnia mózgu, dysplazji konarów górnych móżdżku i zwojów podstawnych

Kod ORPHA
467166

Kod OMIM
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Kod ICD10
Q04.8

Kod ICD11
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*Źródło

orphanet