Opis choroby *

Definicja

A rare genetic neurological disorder characterized by severe pseudo-TORCH syndrome with signs of brain damage and occasionally systemic manifestations resembling the sequelae of congenital infection, but in the absence of an infectious agent. Characteristic features include microcephaly, white matter disease, cerebral atrophy, cerebral hemorrhage, and calcifications, among others. Affected individuals typically have seizures and respiratory insufficiency and die in infancy.

Dane

Klasyfikacja
Choroba

Kod ORPHA
481665

Kod OMIM
617397

Kod ICD10
Q04.8

Kod ICD11
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*Źródło

orphanet