Opis choroby *

Definicja

A rare, severe, genetic neurometabolic disorder associated with clinical manifestations related to impaired synthesis of dopamine, noradrenaline, adrenaline and serotonin. Clinical manifestations are typically characterized by early-onset muscular hypotonia, movement disorders (oculogyric crisis, dystonia), developmental delay, ptosis and non-motor symptoms (sleep disturbance, irritability, excessive sweating, and nasal congestion).

Dane

Klasyfikacja
Choroba

Synonimy
AADC deficiency
Niedobór AADC

Kod ORPHA
35708

Kod OMIM
608643

Kod ICD10
G24.8

Kod ICD11
5C59.00

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*Źródło

orphanet