Opis choroby *

Definicja

A rare and severe inborn metabolic disease characterized clinically by the association of severe-to-profound neurodevelopmental impairment, severe visual impairment, ante-postnatal growth impairment, severe scoliosis and, frequently, early-onset epilepsy.

Dane

Klasyfikacja
Choroba

Synonimy
5-amino-4-imidazole carboxamide ribosiduria
5-amino-4-imidazole carboxamide ribosiduria
Niedobór ATIC
AICA-ribosiduria due to ATIC deficiency
AICAR transformylase/IMP cyclohydrolase deficiency
ATIC deficiency

Kod ORPHA
250977

Kod OMIM
608688

Kod ICD10
E79.8

Kod ICD11
5C55.0Y

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*Źródło

orphanet