Opis choroby *

Definicja

Hyperprolinemia type 2 is an autosomal recessive proline metabolism disorder due to pyroline-5-carboxylate dehydrogenase deficiency. The condition is often benign but clinical signs may include seizures, intellectual deficit and mild developmental delay.

Dane

Klasyfikacja
Choroba

Synonimy
Delta-1-pyrroline-5-carboxylate dehydrogenase deficiency
Niedobór dehydrogenazy delta 1-pirolino-5-karboksylanu

Kod ORPHA
79101

Kod OMIM
239510

Kod ICD10
E72.5

Kod ICD11
5C50.8

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*Źródło

orphanet