

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-karboksyłanu

Kod ORPHA

79101

Kod OMIM

239510

Kod ICD10

E72.5

Kod ICD11

5C50.8

*Źródło

orphanet