

Opis choroby *

Definicja

A rare, genetic, neurometabolic disease characterized by prenatal and postnatal growth retardation, hypotonia, failure to thrive, large and late-closing fontanel, development delay, cutis laxa, joint laxity, progeroid appearance, and dysmorphic facial features. In addition, corneal opacities, cataracts, myopia, seizures, hyperreflexia and athetoid movements have also been associated.

Dane

Klasyfikacja

Podtyp etiologiczny

Synonimy

Delta-1-pyrroline 5-carboxylate synthetase deficiency
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Niedobór syntetazy karboksylanu delta-1-piroliny
Niedobór P5CS
Zespół nerwowo-skrótny, typ Bicknella
Neurocutaneous syndrome, Bicknell type
P5CS deficiency

Kod ORPHA

35664

Kod OMIM

219150

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet