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
Delta-1-pyrroline 5-carboxylate synthetase deficiency Niedobór syntetazy karboksylanu delta-1-piroliny
Niedobór P5CS
Zespół nerwowo-skórny, 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