

Mnogie łagodne bruzdy-obręcze na kończynach

Kod Orpha: 2505 Kod OMIM: 616734

Opis choroby *

Definicja

A rare genetic disease characterized by benign circumferential skin creases, mainly on the limbs, due to folding of excess skin. The creases often improve spontaneously in childhood. Patients also exhibit variable degrees of intellectual disability, short stature, cleft palate, and facial dysmorphism (including epicanthal folds, microphthalmia, broad nasal bridge, low-set, posteriorly rotated ears, and microstomia, among others). Variable additional features have been reported, such as seizures, infantile hypotonia, hearing impairment, strabismus, and urogenital anomalies. Brain imaging may show hypoplastic corpus callosum or mildly dilated ventricles.

Dane

Klasyfikacja

Choroba

Synonimy

CCSF

CCSF

Zespół Kunze'a i Riehma

Wrodzone obwodowe fałdy skórne

Circumferential skin creases, Kunze type

Congenital circumferential skin folds

Kunze-Riehm syndrome

Kod ORPHA

2505

Kod OMIM

616734

Kod ICD10

Q82.8

Kod ICD11

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[*Źródło](#)

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Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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