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

orphanet