Opis choroby *

Definicja

Oculocerebral hypopigmentation syndrome, Cross type is a rare congenital syndrome characterized by cutaneous and ocular hypopigmentation, various ocular anomalies (e.g. corneal and lens opacity, spastic ectropium, and/or nystagmus), growth deficiency, intellectual deficit and other progressive neurologic anomalies such as spastic tetraplegia, hyperreflexia, and/or athetoid movements. The clinical picture varies among patients and may also include other anomalies such as urinary tract abnormalities, Dandy-Walker malformations, and/or bilateral inguinal hernia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cross syndrome
Zespół Crossa

Kod ORPHA
2719

Kod OMIM
257800

Kod ICD10
E70.3

Kod ICD11
EC23.20

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*Źródło

orphanet