## Opis choroby *

Definicja

A group of multiple congenital anomalies syndromes associated to KLHL7 biallelic variants, ranging from a phenotype partially overlapping the Bohring-Opitz syndrome (BOS) to a phenotype overlapping the Crisponi/Cold-Induced Sweating syndrome (CS/CISS), with some patients presenting features of both conditions.

Dane

**Klasyfikacja**
Grupa fenomenów

**Kod ORPHA**
603699

**Kod OMIM**
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**Kod ICD10**
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**Kod ICD11**
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*Źródło

orphanet