

## 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