

## Opis choroby \*

### Definicja

A rare genetic disease characterized by congenital contractures of the distal interphalangeal joints, progressive stiffness of the shoulders and neck, keloid scarring, increased optic cup-to-disc ratio, and renal stones. Additional reported features include arthritis, osteoporosis, hypoplastic flexion creases, clinodactyly, anxiety, and facial dysmorphism (such as sloping forehead, prominent supraorbital ridges, downslanting palpebral fissures, prominent ears, and high arched palate). Female carriers exhibit a variable, milder phenotype.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA	Kod OMIM	Kod ICD10
482606	-	-
<b>Kod ICD11</b>		
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### \*Źródło

orphanet