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
482606

Kod OMIM
-

Kod ICD10
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Kod ICD11
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*Źródło

orphanet