

Opis choroby *

Definicja

Dermochondrocorneal dystrophy is characterised by osteochondrodystrophy of the hands and feet, corneal dystrophy and the presence of skin nodules clustered around the metacarpophalangeal and interphalangeal joints, around the nose and ears and on the posterior surface of the elbow. Gingival lesions may also be present. It has been described in less than 20 patients. Transmission is autosomal recessive.

Dane

Klasyfikacja

Choroba

Synonimy

François syndrome

Zespół François

Kod ORPHA

79149

Kod OMIM

221800

Kod ICD10

L98.8

Kod ICD11

EC4Y

*Źródło

orphanet