

## Opis choroby \*

### Definicja

A rare genetic disease characterized by sclerosing dysplasia affecting the diaphyseal and metaphyseal regions of the long bones, as well as the skull and metacarpals, in association with skin changes like those seen in ichthyosis vulgaris and premature ovarian failure with bilateral hypoplasia of the ovaries. Patients present in adulthood, primarily with swelling of the extremities and occasional mild pain in the legs.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Sclerosing dysplasia of bone-ichthyosis-  
premature ovarian failure syndrome  
Stwardniająca dysplazja kości - rybia łuska -  
przedwczesne wygasanie czynności jajników

#### Kod ORPHA

75325

#### Kod OMIM

609993

#### Kod ICD10

M85.8

#### Kod ICD11

LD2F.1Y

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#### \*Źródło

orphanet