

## Opis choroby \*

### Definicja

Rothmund-Thomson syndrome type 2 is a subform of Rothmund-Thomson syndrome (RTS; see this term) presenting with a characteristic facial rash (poikiloderma) and frequently associated with short stature, sparse scalp hair, sparse or absent eyelashes and/or eyebrows, congenital bone defects and an increased risk of osteosarcoma in childhood and squamous cell carcinoma later in life.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Poikiloderma of Rothmund-Thomson type 2  
Poikilodermia Rothmunda i Thomsona typu 2  
RTS2  
RTS2

#### Kod ORPHA

221016

#### Kod OMIM

268400

#### Kod ICD10

Q82.8

#### Kod ICD11

LD2B

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#### [\\*Źródło](#)

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