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

orphanet