

Opis choroby *

Definicja

Rothmund-Thomson syndrome type 1 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, and rapidly progressive bilateral juvenile cataracts. In contrast to RTS2 (see this term), patients with RTS1 do not appear to have an increased risk of developing cancer.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

Poikiloderma of Rothmund-Thomson type 1
Poikilodermia Rothmunda i Thomsona typu 1
RTS1
RTS1

Kod ORPHA

221008

Kod OMIM

618625

Kod ICD10

Q82.8

Kod ICD11

LD2B

[*Źródło](#)

orphanet