

Opis choroby *

Definicja

Rothmund-Thomson syndrome (RTS) is a genodermatosis presenting with a characteristic facial rash (poikiloderma) associated with short stature due to pre- and postnatal growth delay, sparse scalp hair, sparse or absent eyelashes and/or eyebrows, juvenile cataracts, skeletal abnormalities, radial ray defects, premature aging and a predisposition to certain cancers.

Dane

Klasyfikacja

Choroba

Synonimy

Poikiloderma of Rothmund-Thomson
Poikilodermia Rothmunda i Thomsona
RTS
RTS

Kod ORPHA

2909

Kod OMIM

268400

Kod ICD10

Q82.8

Kod ICD11

LD2B

*Źródło

orphanet