Open Peer Review on Qeios

Rothmund-Thomson syndrome type 2

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Rothmund-Thomson syndrome type 2. ORPHA:221016

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.