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## Rothmund-Thomson syndrome

### What is Rothmund-Thomson syndrome?

Rothmund-Thomson syndrome is a rare inherited disease that affects the skin, eyes, bones and internal organs. At least 300 cases have been reported in medical journals since a case was first described by Rothmund in 1868. Rothmund-Thomson syndrome is also known as poikiloderma congenitale.

### What is the cause of Rothmund Thomson syndrome?

Rothmund Thomson syndrome is due to a genetic defect, in which there are mutations in the RECQL4 gene on Chromosome 8. This gene encodes for the enzyme DNA helicase which unwinds DNA (deoxyribonucleic acid). The abnormal gene makes the chromosomes unstable, altering the growth of cells in many tissues.

The defect is inherited as a autosomal recessive trait. This means an abnormal gene must come from each parent.

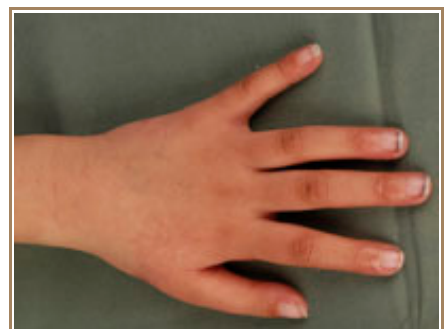
### What are its clinical features?

Affected children may be identified early in life by their small size, their tendency to sunburn easily, and from the appearance of their skin, teeth and bones. Rothmund Thomson syndrome is slightly more common in females than males.

Rothmund-Thomson syndrome



Early poikiloderma



Abnormal carrying angle

Abnormal thumb of younger sister

Telangiectasia on back of hand

Affected organ	Clinical features
Skin	<ul style="list-style-type: none"> <li>• Photosensitivity: sunburn-like redness, swelling and blisters on cheeks and face; may extend to involve buttocks and extremities. Noted during the first year of life in 90%.</li> <li>• Poikiloderma: variegated pigmentation, telangiectasia (prominent tiny blood vessels) and skin thinning; usually evident on cheeks, hands and buttocks by 3–5 years of age.</li> <li>• Thin eyebrows and sparse scalp hair.</li> <li>• Abnormal, brittle nails.</li> </ul>
Eyes	<ul style="list-style-type: none"> <li>• Cataracts: lens opacities occur in 50% of children aged 3–7 years and are often bilateral.</li> <li>• Corneal lesions are less common.</li> </ul>
Bones	<ul style="list-style-type: none"> <li>• Bony defects affect over 50% of children, who are often of short stature.</li> <li>• These include dysplasia (abnormal growth), sclerosis (thickening and hardening) and cystic abnormalities of the long bones.</li> <li>• Small hands and feet; absent or malformed radii and thumbs.</li> <li>• Osteoporosis and bone hypoplasia (bone thinning) are common with aging. Pathological fractures may occur with minimal trauma.</li> <li>• Other changes include: widened long bone epiphyses (part of the bone where growth occurs), iliac bone hyperplasia (excessive growth), trabeculated metaphyses (middle part of the long bone).</li> </ul>
Dentition	<ul style="list-style-type: none"> <li>• Agenesis (absent formation of teeth).</li> <li>• Microdontia (small teeth).</li> <li>• Delayed and ectopic eruption of teeth.</li> <li>• Supernumery teeth.</li> </ul>
Reproductive system	<ul style="list-style-type: none"> <li>• Hypogonadism (reduced function of the gonads) in 25%.</li> <li>• Juvenile-appearing genital organs.</li> <li>• Amenorrhoea (lack of menstruation).</li> <li>• Sterility.</li> </ul>
Endocrine system	<ul style="list-style-type: none"> <li>• Parathyroid adenoma.</li> <li>• Disturbed thyroid function.</li> </ul>
Gastrointestinal system	<ul style="list-style-type: none"> <li>• Chronic nausea and vomiting.</li> <li>• Diarrhoea.</li> </ul>
Intellect	<ul style="list-style-type: none"> <li>• Intellectual impairment in up to 30%.</li> </ul>

## What are the complications?

The main concern is an increased susceptibility to cancer.

### Skin cancer

[Skin cancers](#) including [basal cell carcinoma](#), [squamous cell carcinoma](#) and [intraepidermal carcinoma \(Bowen\)](#)

[disease](#)) are common in older children and adults with Rothmund–Thomson syndrome. They often arise on the face, neck and limbs. It is postulated that skin cancers occur because of defects in DNA repair after exposure to ultraviolet radiation.

### **Bone cancer**

The second most common type of cancer is osteosarcoma, which may develop in late childhood or adolescence. Osteosarcoma may arise within pre-existing bone dysplasia so it may be difficult to diagnose on X-ray.

Treatment of osteosarcoma in Rothmund–Thompson syndrome is similar to in patients without the syndrome.

### **Other malignancies**

Other cancers affecting individuals with Rothmund–Thomson syndrome include:

- Gastric adenocarcinomas
- Fibrosarcomas
- Hodgkin lymphoma
- Malignant eccrine poroma

## **Management**

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Children with Rothmund–Thomson syndrome are often followed up by a paediatrician, dermatologist, orthopaedic surgeon, dental surgeon and/or other specialists. Clinicians should bear in mind the risk of cancers, and should monitor and investigate as appropriate.

[Sun protection](#) is very important because of photosensitivity and increased risk of skin cancer. This should include seeking shade, fully covering clothing and broad spectrum [sunscreens](#).

Genetic counselling is important for family members

*Draft 14 June 2008*

### **Related information**

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#### **References:**

- Bologna J et al. *Dermatology* 2nd Edition (2008) pages 1343–1345
- Cumin I et. al. Rothmund–Thomson Syndrome and Osteosarcoma. *Medical and Pediatric Oncology* 36: 414–416 (1996)
- Mallory S et. al. What syndrome is this? *Pediatric Dermatology* Vol. 16 No. 1 59–61, 1999
- Duker N. Chromosome Breakage Syndromes and Cancer. *American Journal of Medical Genetics* 115:125–129 (2002)
- Hicks J et. al. Clinicopathologic features of osteosarcoma in patients with Rothmund–Thomson Syndrome. *Journal of Clinical Oncology* 25: 370–375 (2007)

#### **On DermNet NZ:**

- [Photosensitivity](#)

#### **Other websites:**

- [Rothmund–Thomson Syndrome](#) – emedicine dermatology
- [Rothmund–Thomson Syndrome](#) – MedicineNet.com
- [Rothmund–Thomson Syndrome](#) – NCBI Gene Reviews
- [Rothmund–Thomson Syndrome; RTS](#) – OMIM
- [Rothmund–Thomson Syndrome](#) – Genetics Home Reference
- [Rothmund Thomson Syndrome](#) – NORD National Organisation for Rare Disorders

#### **Books about skin diseases:**

See the [DermNet NZ bookstore](#)

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