

## Rothmund-Thomson Syndrome Foundation

The Rothmund-Thomson Syndrome Foundation is a [501\(c\) \(3\) nonprofit organization](#). It was incorporated in November, 2009 by the Kimmel Family after the loss of their daughter Victoria who had RTS. Currently, we operate with nine volunteer board members who raise money to support the Foundation's goals.



**The Mission** of the Rothmund-Thomson Syndrome Foundation is to foster worldwide awareness and education about RTS and related disorders and to encourage research through support of scientists and physicians devoted to studying the clinical spectrum, molecular and cellular basis of RECQL4 and related syndromes.



## *Ways to Give*

The Research of Rothmund-Thomson Syndrome (RTS) and related disorders is ongoing at Baylor College of Medicine, in affiliation with Texas Children's Hospital. Your support will help us continue to promote RTS awareness to the general public, healthcare professionals, and individuals affected with RTS and their family members. Your donations will also enable us to sponsor annual conferences and regional seminars, and expand research activities worldwide.

### Donate by check

Please make checks payable to the **RTS Foundation** and send directly to the address below:  
RTS Foundation  
4307 Woodward Court  
Chantilly, VA 20151

### Donate by credit card

For more information on how to make a donation by credit card, please visit our website: [www.rtsplace.org](http://www.rtsplace.org)

### Get Involved

Visit our website: [www.rtsplace.org](http://www.rtsplace.org) for news and upcoming events.

**Facebook** Join us at Rothmund-Thomson Syndrome Foundation(RTS). A support group has been established for patients and their family members and is currently active and growing.



## *Understanding Rothmund-Thomson Syndrome*



*How common is it?*

*What are the features?*

*How is it diagnosed?*

*How is it inherited?*

*Are resources available?*

[www.rtsplace.org](http://www.rtsplace.org)

## What is RTS?

Rothmund-Thomson syndrome (RTS) is a rare genetic disorder that can affect people of all races and nationalities. Symptoms often first appear during infancy, usually between three and six months of age, in the form of a skin rash on the cheeks. Patients can have just a few or several features of the syndrome.

## HOW COMMON IS RTS?

RTS is a rare disorder, and the exact number of cases worldwide is not known. There have been a few hundred cases of RTS reported in the medical literature. RTS is not specific to any one race or nationality. However, because it is a recessive disorder, consanguineous (related by blood) families have an increased risk of disease.

## CLINICAL FINDINGS

RTS is characterized by the following clinical features:

- A unique skin rash, that begins in infancy usually on the cheeks, and spreads to the arms and legs and persists for life. This rash is called *poikiloderma*, and consists of areas of increased and decreased pigmentation, prominent blood vessels, and thinning of the skin.
- Sparse/absent scalp hair, eyebrows, and/or eyelashes
- Small stature
- Diarrhea and/or vomiting in early childhood
- Bone deformities
- Dental and nail abnormalities
- Juvenile cataracts
- Cancer: skin (usually basal cell or squamous cell carcinomas) or bone (osteosarcoma)

## DIAGNOSIS OF RTS

Diagnosis of RTS is primarily made through evaluation of clinical features. In general, having the classic rash (poikiloderma) is the main finding for making the diagnosis of RTS. A skin biopsy may also be helpful in showing changes that are consistent with poikiloderma. Patients may have an "atypical" rash that is not quite classic for RTS, but if they have at least two other of the clinical features associated with RTS, then they are considered to have "Probable RTS." In most cases of RTS, the clinical diagnosis can be confirmed by molecular testing of the *RECQL4* gene.

## What is *RECQL4*?

*RECQL4* is a gene located on chromosome 8. Genes are packages of DNA, or genetic material, that help in the formation of proteins which carry out specific functions in the body. *RECQL4* is the only gene that is currently known to be associated with RTS. Patients with mutations, or changes, in both copies of their *RECQL4* genes can develop RTS. However, only about two-thirds of patients with a clinical diagnosis of RTS will have mutations found in their *RECQL4* genes. Therefore, there are likely other genes that can also cause RTS. DNA sequence analysis and mutation testing of the *RECQL4* gene are available through the Baylor College of Medicine Medical Genetics Laboratory. Presence of mutations in *RECQL4* can confirm a diagnosis of RTS; however, absence of mutations in *RECQL4* does not necessarily mean that the patient does not have RTS, since one-third of patients clinically diagnosed with RTS do not have mutations in this gene. Researchers at Texas Children's Cancer Center have shown that presence of *RECQL4* mutations is associated with increased risk for developing osteosarcoma. They are continuing to study the function of the *RECQL4* protein to try to understand why lack of this protein leads to the findings seen in RTS.

## INHERITANCE

Since RTS is usually inherited in an autosomal recessive pattern, affected individuals inherit one abnormal (mutated) gene from each parent. If an individual only has a mutation in one gene, then he/she is a carrier (e.g., parents of RTS patients are carriers). Therefore, at conception there is a 25% chance of the offspring of two carriers to be affected with the disease, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected ("normal" *RECQL4* genes).

## ADJUSTING TO RTS

Most parents of an individual affected with RTS wonder if their child will ever be able to lead a normal life. Ongoing research studies conducted at Baylor College of Medicine and Texas Children's Hospital have found that RTS patients can lead very fulfilling lives. Patients appear to have normal life spans (in the absence of cancer), and several have gone on to have families of their own.



## RTSF Medical Advisory Board Members

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Contact us at: [rtssupport@rtsplace.org](mailto:rtssupport@rtsplace.org)

For more information on Rothmund-Thomson Syndrome, please visit:  
**Rothmund-Thomson Syndrome Foundation**  
<http://www.rtsplace.org>

### **Gene Reviews**

<http://www.genetests.org>