Rubinstein-Taybi syndrome
This guide is not meant to take the place of medical advice.

Please consult with your doctor about your genetic results and health care choices. The information in this guide was up to date at the time it was written in 2019. But new information may come to light with new research. You may find it helpful to share this guide with friends and family members or doctors and teachers of the person who has Rubinstein-Taybi syndrome.
What is Rubinstein-Taybi syndrome?

Rubinstein-Taybi syndrome happens when there are changes to the CREBBP gene or the EP300 gene. These changes can keep these genes from working as they should.

**Key role**
The CREBBP and EP300 genes play a key role in controlling the activity of other genes.

**Symptoms**
Because the CREBBP and EP300 genes are important in the development of the body, many people who have Rubinstein-Taybi syndrome have:

- Intellectual disability
- Growth problems and short height
- Behavior issues
- Changes in facial features and the hands and feet
What causes Rubinstein-Taybi syndrome?

Our genes contain the instructions, or code, that tell our cells how to grow, develop, and work. Every child gets two copies of the CREBBP and EP300 genes: one copy from their mother, from the egg, and one copy from their father, from the sperm. In most cases, parents pass on exact copies of the gene to their child. But the process of copying genes is not perfect. A change in the genetic code can lead to physical issues, developmental issues, or both.

Sometimes a random change happens in the sperm or egg. This change to the genetic code is called a ‘de novo’, or new, change. The child can be the first in the family to have the gene change.

New/De Novo genetic changes

Genetic change occurs in egg or sperm or after fertilization

Child with de novo genetic change in autism gene
De novo changes can take place in any gene. We all have some de novo changes, most of which don’t affect our health. But because the CREBBP and EP300 genes play important roles in development, de novo changes in these genes can have a meaningful effect.

About 60 percent of cases of Rubinstein-Taybi syndrome are caused by changes in the CREBBP gene. About 10 percent are caused by changes in the EP300 gene. The cause of the remaining cases is not well understood.

Research shows that Rubinstein-Taybi syndrome is often the result of a de novo change in the CREBBP or EP300 genes. Many parents who have had their genes tested do not have the gene change found in their child who has the syndrome. In some cases, Rubinstein-Taybi syndrome happens because the gene change was passed down from a parent. This is called dominant inheritance.
Why does my child have a change in the CREBBP or EP300 genes?

No parent causes their child’s Rubinstein-Taybi syndrome. We know this because no parent has any control over the gene changes that they do or do not pass on to their children. Please keep in mind that nothing a parent does before or during the pregnancy causes this to happen. The gene change takes place on its own and cannot be predicted or stopped.
What are the chances that other family members or future children will have Rubinstein-Taybi syndrome?

Each family is different. A geneticist or genetic counselor can give you advice on the chance that this will happen again in your family.

The risk of having another child who has Rubinstein-Taybi syndrome depends on the genes of both birth parents.

- If neither birth parent has the same gene change found in their child, the chance of having another child who has the syndrome is on average 1 percent. This 1 percent chance is higher than the chance of the general population. The increase in risk is due to the very unlikely chance that more of the mother’s egg cells or the father’s sperm cells carry the same change in the gene.

- If one birth parent has the same gene change found in their child, the chance of having another child who has the syndrome is 50 percent.

For a symptom-free sibling, a brother or sister, of someone who has Rubinstein-Taybi syndrome, the risk of having a child who has the syndrome depends on the symptom-free sibling’s genes and their parents’ genes.

- If neither parent has the same gene change found in their child who has the syndrome, the symptom-free sibling has a nearly 0 percent chance of having a child who has Rubinstein-Taybi syndrome.

- If one birth parent has the same gene change found in their child who has the syndrome, the symptom-free sibling has a small chance of also having the same gene change. If the symptom-free sibling has the same gene change as their sibling who has the syndrome, the symptom-free sibling’s chance of having a child who has Rubinstein-Taybi syndrome is about 50 percent.

For a person who has Rubinstein-Taybi syndrome, the risk of having a child who has the syndrome is about 50 percent.
How many people have Rubinstein-Taybi syndrome?

As of 2019, about 300 people in the world with Rubinstein-Taybi syndrome had been described in the medical literature. The first case of Rubinstein-Taybi syndrome was described in 1995. Scientists estimate that the condition happens in 1 in 100,000 to 1 in 125,000 live births.

Do people who have Rubinstein-Taybi syndrome look different?

People who have Rubinstein-Taybi syndrome often have distinct features. Appearance can vary and can include some but not all of these features:

- Short height
- A small head, also called microcephaly
- Facial features that are different from those of other family members
- Broad thumbs that may be positioned differently on the hand
- Big first toes
How is Rubinstein-Taybi syndrome treated?

At this point, there are no medicines designed to treat the syndrome. A genetic diagnosis can help people decide on the best way to track the condition and manage therapies. Doctors can refer people to specialists for:

- Genetics consults.
- Development and behavior studies.
- Neurological studies.
- Orthopedic studies.

A developmental pediatrician, neurologist, or psychologist can follow progress over time and can help:

- Suggest the right therapies. This can include physical, occupational, speech, or behavioral therapy.
- Guide individualized education plans (IEPs).

Specialists advise that therapies for Rubinstein-Taybi syndrome should begin as early as possible, ideally before a child begins school.

People who have Rubinstein-Taybi syndrome may have a higher risk of developing tumors. These may be cancerous or non-cancerous. Your doctor can recommend whether you need additional screening or to consult a specialist.
Rubinstein-Taybi syndrome is rare. As of 2019, studies described around 300 people who have the syndrome.

This section includes a summary of information from major published articles. It highlights how many people have different symptoms. To learn more about the articles, see the Sources and references section of this guide.

Behavior and development concerns linked to Rubinstein-Taybi syndrome

**Speech**

Ninety percent of people who have the syndrome have speech delay and persistent difficulty with language fluency.

**Learning**

Most people have some degree of intellectual disability.

**Mental health**

Some people have anxiety or obsessive-compulsive disorder.

**Behavior**

Some people have autism.
Medical and physical concerns linked to Rubinstein-Taybi syndrome

**Growth**

*Short height and small head size* are common among people who have the syndrome.

**Feeding and digestion issues**

About 70 percent of people have gastrointestinal concerns.

**Eyes and eyesight**

People who have Rubinstein-Taybi syndrome may have eye issues, including *crossed eyes*, *blocked tear ducts*, also called nasolacrimal duct obstruction, *cataracts*, *gaps in a part of the eye*, also called coloboma, glaucoma, and issues with the cornea.

**Birth defects**

Some people have *broad thumbs* and *distinct facial features*. A small percentage have *heart disease* at birth.

**Cancer risk**

About 5 percent of people who have the syndrome develop *cancerous or non-cancerous tumors*, including certain kinds of brain tumors. *Blood and bone marrow cancers*, also called leukemia, are more common in people who have Rubinstein-Taybi syndrome.
Simons Searchlight is another research program sponsored and run by the Simons Foundation Autism Research Initiative, also known as SFARI. As part of the next step in your research journey, Simons Searchlight offers you the opportunity to partner with scientists and other families who have the same gene change. Simons Searchlight is a registry for more than 200 genetic changes that are associated with neurodevelopmental conditions, including autism spectrum disorder. Simons Searchlight makes it easier for researchers to access the information they need to advance research on a condition.

To register for Simons Searchlight, go to the Simons Searchlight website at www.simonssearchlight.org and click “Join Us Today”.

- Learn more about Simons Searchlight
  www.simonssearchlight.org/frequently-asked-questions

- Simons Searchlight page on CREBBP
  www.simonssearchlight.org/research/what-we-study/crebbp

- Simons Searchlight Community CREBBP Facebook group
  www.facebook.com/groups/2566586560099702

- Simons Searchlight Community EP300 Facebook group
  www.facebook.com/groups/484880595623787
Sources and References

The content in this guide comes from published studies about Rubinstein-Taybi syndrome. Below you can find details about each study, as well as links to summaries or, in some cases, the full article.

  www.ncbi.nlm.nih.gov/books/NBK1526


  www.ncbi.nlm.nih.gov/pubmed/29506490