

BBSOAS

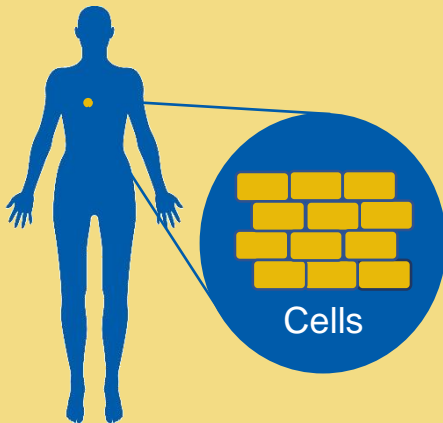
**Bosch-Boonstra-Schaaf
optic atrophy syndrome**

Easy read guide

This leaflet is for people with BBSOAS.



Before we learn about BBSOAS, first let us learn a little bit about our bodies.



Our bodies are built from millions of tiny building blocks called cells.



Our cells contain genes.
Genes are the instructions that tell our bodies how to grow and work.

NR2F1

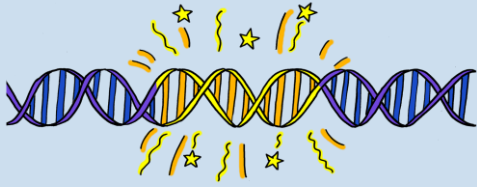
FOXP1

PUF60

ANKRD11

We all have thousands of genes.

Each gene has its own special name and function.



Sometimes we have a change in a gene.

The change can affect how the gene works.

NR2F1

A change to a gene called NR2F1 causes BBSOAS.



What is BBSOAS?

BBSOAS is a genetic condition.



People with BBSOAS live all over the world and they all have a change in the NR2F1 gene.



Lots of people with BBSOAS have an eye problem or can't see very well without glasses.



Some people with BBSOAS find some things harder to do than other people with BBSOAS.



Some people find it difficult to learn how to read.



Some people find it difficult to learn how to speak.



Some people need help to understand things.



Some people find it difficult to make friends.



Some people worry a lot.
There is help for people who worry a lot.



People with BBSOAS might also have other difficulties and strengths.



Why do I have BBSOAS?

BBSOAS happens by chance.

Most people are the first person in their family to have it.



We all have gene changes.

We only have a genetic condition when important genes are changed.



If you had a child, they could have BBSOAS too.

You could talk to a genetics doctor about this.



How do I know I have BBSOAS?

You can only find out by having a genetic test.



It is good to know that you have BBSOAS so you can get extra help if you need it.

We all need help sometimes!



Can the gene change be removed?

A gene change cannot be removed or replaced.



Knowing you have BBSOAS is important because it might help doctors and other professionals to find ways to help you.



If you would like to know more about BBSOAS, you can talk to someone who knows about genetics or a BBSOAS group or a charity like Unique.

Inform Network Support



Understanding Chromosome & Gene Disorders

Rare Chromosome Disorder Support Group

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Join Unique to meet other families and get information and support.

Unique is a charity without government funding, existing entirely on donations and grants. If you can please make a donation via our website at: www.rarechromo.org/donate Please help us to help you!

Websites that you might find helpful

<https://www.learningdisabilities.org.uk/>

<https://www.turning-point.co.uk>

<https://www.unitedresponse.org.uk>

BBSOAS Foundation (US)

Website: www.BBSOASfoundation.org

Facebook: <https://www.facebook.com/BBSOASfoundation>

Instagram: <https://www.instagram.com/BBSOASfoundation/>

Twitter: <https://x.com/BBSOASfoundation>

BBSOAS Facebook Support group

Parent Support Group: <https://www.facebook.com/groups/667174239987278>

Wiedemann -Steiner Syndrome: Info for Families, Friends, and Professionals
<https://www.facebook.com/groups/339084866256825>

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