



PORETTI BOLTSHAUSER SYNDROME

- **WHAT IS PTBHS?**
- **WHAT ARE THE SYMPTOMS?**
- **HELPFUL THERAPIES**
- **UNIQUE & CONNECTED**



**A FLYER CREATED BY
UNIQUE & CONNECTED**

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WHY THIS FLYER?

This flyer is written by Alissa, chair(wo)man of the foundation Unique and Connected.

As an individual with Poretti Boltshauser Syndrome, PTBHS, she knows what it means to have an extremely rare diagnosis. There is a lack of understandable information out there. To fix this issue, the foundation Unique and Connected was created.

“I know from personal experience how frustrating it can be, to not have a lot of information about the syndrome online. I know how it feels to be misunderstood by doctors or professionals. I understand the worries and the feelings of insecurity that come along with having such a rare diagnosis.

This flyer is here because I want you all to be able to spread the information about the syndrome you, or a loved one, has. To be able to show people around you what it means, and to hopefully create a better understanding of PTBHS.”

Feel free to download this flyer and send it to other people.

Unique and Connected is a foundation and everything we do is to help the people with rare diagnoses.

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WHAT IS PORETTI BOLTSHAUSER SYNDROME?

Poretti Boltshauser Syndrome (PTBHS) is a rare condition that someone is born with. It affects the cerebellum, the part of the brain that helps coordinate movement and balance. Because of this, people with PTBHS may experience challenges with coordination, muscle control, and development.

One important thing to know is that PTBHS is non-progressive. This means that the symptoms do not get worse over time – they stay relatively stable throughout life.

Sometimes, children with PTBHS have cysts in the cerebellum, although this is not always the case.

PTBHS was only identified in 2014, which makes it a very recently discovered syndrome. Because of this, many individuals may still be undiagnosed or misdiagnosed. The condition is caused by a change in the LAMA1 gene. In many cases, both parents are carriers of this gene without knowing it. However, recent discoveries show that this is not always the case, and the genetics can sometimes be more complex.

Often, it takes time before it becomes clear that a child has PTBHS. Every child develops in their own way, and the signs may only gradually become noticeable.

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9

POSSIBLE SYMPTOMS

Every person with PTBHS is unique. Not everyone will experience the same symptoms, and the severity can vary greatly from person to person.

Some of the challenges that may be seen include:

- *low muscle tone*
- *developmental delays*
- *balance issues*
- *issues with talking*
- *swallowing issues*
- *very nearsighted*
- *Ocular Motor Apraxia*
- *variable retinal dystrophy*
- *problems learning*
- *problems with focusing or concentrating (ADHD)*
- *autistic features*
- *constipation, IBS*
- *problems with getting potty trained*
- *seizures*
- *problems with their energy levels / fatigue*



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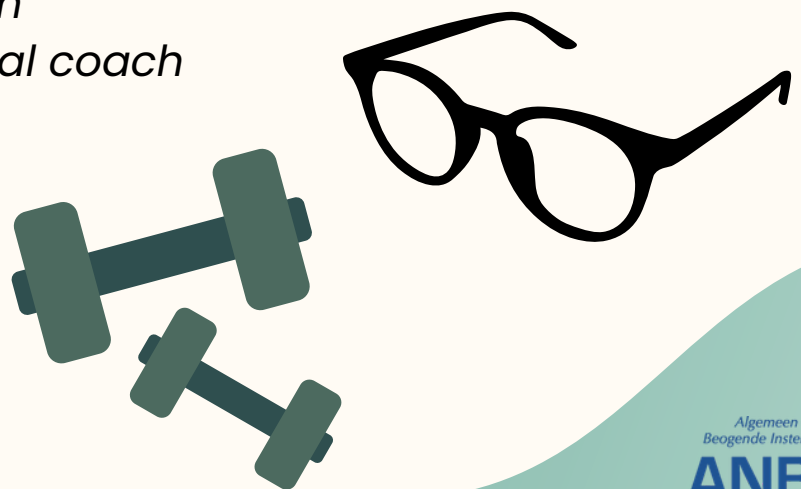
HELPFUL THERAPIES

There is currently no cure for Poretti Boltshauser Syndrome. However, there are many ways to support someone in their development and daily life.

With the right guidance and care, children and adults with PTBHS can continue to grow, learn, and find their own way.

Some forms of support that may be helpful include:

- *glasses or eye patches for a lazy eye*
- *physical therapy*
- *speech therapy*
- *occupational therapy*
- *rehabilitation doctor*
- *special education*
- *medication to improve focus*
- *psychiatrist or psychologist*
- *a dietitian*
- *a personal coach*



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UNIQUE & CONNECTED

The foundation Unique and Connected has been founded to help people navigate through all the information, or rather the lack of it, on the internet about extremely rare syndromes.

OUR MISSION

Our mission is to connect people affected by extremely rare conditions, provide accessible and reliable information, and build a supportive community where no one feels alone in their diagnosis.

To achieve that goal, we want to provide tools and guidance to help people navigate the problems they may face with their condition.

With our foundation we want to create an environment where individuals and families can find information and support about their condition – no matter where they live.



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9