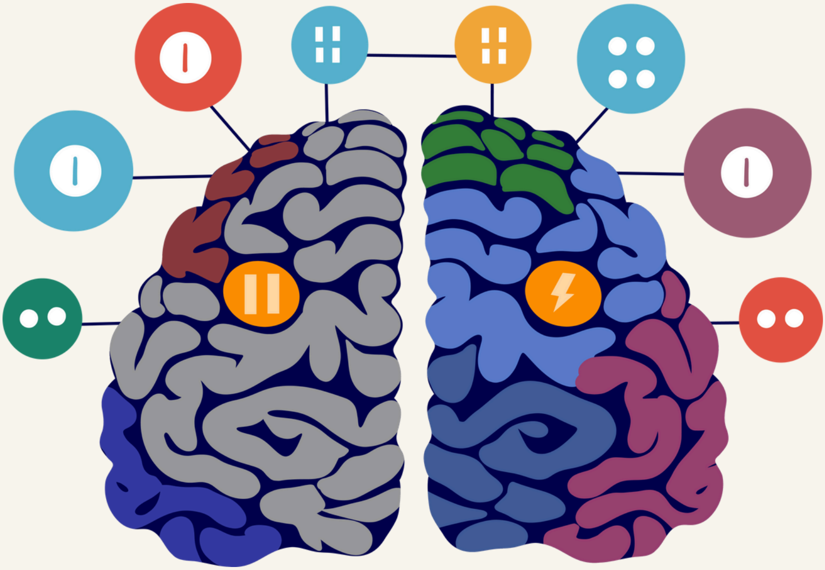
PORETTI BOLTSHAUSER SYNDROME



by

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WHAT IS PBS?

Poretti Boltshauser Syndrome (PBS) is a congenital non-progressive cerebellar ataxia that affects the eyes and the cerebellum.

Non-progressive means that the symptoms don’t get worse throughout life. Congenital means that you are born with it. Ataxia means lack of coordination or muscle movements.

The syndrome is very rare, due to it only being found out about in 2014. So many children who may have it, have not been diagnosed yet.

WHAT DOES THIS MEAN?

The defect in the LAMA1-gene causes the lack of a protein called Laminine A1. This protein plays a big part in forming the eyes, muscles and brain.

Usual in children with PBS, the eyes and the cerebellum are affected.

The cerebellum is involved in all kinds of things, like moving, balancing, coördination of muscles, learning and developing, speak, read and understanding and showing of emotions.

POSSIBLE SYMPTOMS

There is a huge variety in people with PBS, because there is a wide range of symptoms and not everybody shows all of them. The possible symptoms are:

- 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
- autistic features
- constipation, IBS
- problems with getting potty trained

As children with PBS age, they may show improvement at some of these symptoms. Others might have different symptoms from this list.

CONNECTING WITH OTHERS

Feeling alone with a diagnosis, let alone a diagnosis so rare as PBS, is something that I want to prevent from happening.

Know that there are others like you or like your child. Visit our Facebook group (link below) or visit the websites porettiboltshausersyndrome.info and unique-connected.org.

POSSIBLE THERAPIES

Please note that not every country or region provides each type of therapy listed below. This is just a list of possibilities you can consider.

- *glasses or eye patches with a lazy eye*
- *physical therapy*
- *speech therapy*
- *occupational therapy*
- *rehabilitation doctor*
- *special education*
- medication to improve focus
- *psychiatrist*
- *medication for constipation (fibres)*
- *a personal coach*

ABOUT THIS FOLDER

My name is Alissa and I founded the **Unique & Connected** foundation. This foundation is about helping spreading awareness about rare syndromes such as Poretti Boltshauser syndrome.

When I grew up, this syndrome could not yet be diagnosed. This resulted in feeling alone a lot. I didn't feel like people really understood me. And how could they? I was born in 1998 and information could not easily be found.

Today I can help make a difference, by spreading this information with those who need it.