# SHARE OUR STORY AND SUPPORT FAMILIES AFFECTED BY PVNH AND OTHER NEURONAL HETEROTOPIA DISORDERS

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*MEDICAL DISCLAIMER:*
*This document was prepared solely for educational purposes.* PVNH Support & Awareness™ *does not provide medical diagnosis. The information in this pamphlet is not intended nor implied to be a substitute for professional medical advice. Always seek the advice of your physician or other qualified health provider prior to starting any new treatment or with any questions you may have regarding a medical condition.*

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In memory of
ELLA DUPONT BEDASSIE

PVNH caused by a Filamin A mutation

August 7, 2008—March 27, 2009

PVNH
support & awareness™

*the international neuronal heterotopia disorders community*

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On August 7, 2012, World PVNH Disorder Awareness Day™ was celebrated for the first time worldwide, and its official proclamation was signed in Canada, the United States, New Zealand, Italy, the Netherlands, France, Spain, England and Germany by PVNH researchers and specialists.

Meet a few of our members from around the globe. Top to bottom, left to right:
Anthony, Celeste, Sophie, Jackson, Ella, Jonathan, Ghislaine, Elise and Sophia.
All are affected by rare heterotopia disorders.

They are a few members of the 500+ families in 33 countries supported by PVNH Support & Awareness™ since 2009.

October 2009 -
PVNH Support & Awareness™ was created as a legacy for Ella Dupont Bedassie. Ella was 7 months and 20 days in March 2009 when she passed away from respiratory failure, the cause of which was not known at the time.
Six months later, confirmation came that she had the rare disease X-Linked Periventricular Nodular Heterotopia, caused by a Filamin A (FLNA) gene mutation.

## WHAT IS PVNH?

And why create an awareness day for it?

Periventricular nodular heterotopia (PVNH) is a rare neuronal migration disorder characterized by the presence of nodules of neurons (gray matter) in the wrong spot in the brain. It can also be referred to as BPH (bilateral periventricular heterotopia), BPNH (bilateral periventricular nodular heterotopia), PNH (periventricular nodular heterotopia), subependymal heterotopia, or simply gray matter heterotopia (GMH). There is also another form of neuronal heterotopia disorder: subcortical band heterotopia (SBH), also called double cortex. A brain MRI investigation usually confirms the diagnosis.

PVNH is one of those rare disorders you may not know you have. Seizures occur in up to 90% of patients, and learning difficulties are not uncommon, but not everyone has these symptoms. However, some individuals have motor, cognitive and developmental delays and some patients have connective tissue problems which can affect joints and blood vessels and lead to gastrointestinal, pulmonary and heart complications.

There is no cure for PVNH,
only symptoms can be treated.

If unrecognized and untreated,
these problems can be fatal.
Help us spread the word to educate patients and medical communities alike around the world.

## TELL ME MORE*

PVNH is a widely variable disorder with many causes and potential outcomes ranging from normal development and functioning to profound impairment.

Its incidence is unknown. In some families the condition and seizures are passed down through generations and considered 'familial'. In others, an individual may be the first in a family to show symptoms, representing a sporadic occurrence.

We know the most about a genetic form of PVNH due to mutations in the FLNA (Filamin1/Filamin A) gene located on the X-chromosome. This 'classic' form of bilateral PVNH with a high incidence of seizures was once thought to lead to early lethality in males. However, male patients with FLNA gene mutations have been identified in recent years. Some have passed the gene / condition down to their daughters. Most commonly, females present with seizures by late childhood / adolescence, though the range of onset is very broad. Seizures can start much earlier or never occur.
Other reported complications include vascular / clotting problems that may need monitoring. Most of the time, a family history of heterotopia and seizures in multiple individuals is due to FLNA mutation. Even if a patient does not have a family history, if there are bilateral and multiple nodules, the FLNA gene should be checked. Different types of mutations in the FLNA gene can lead to skeletal as well as developmental defects.

PVNH may be present as just one or two isolated nodules of varying sizes (less likely to be related to FLNA mutations). Little is known about its basis, causes, outcomes. In some familial and sporadic cases of typical PVNH, the FLNA gene is normal, leading researchers to believe that other genetic causes exist. New genes have been discovered as causing PVNH. PVNH has also been reported in individuals with other known genetic conditions, including Fragile X and Williams syndromes, among others. It can also be found in association with various chromosome abnormalities and deletion syndromes.

Reported distinguishable types of PVNH include:
(1) Bilateral, multiple and symmetrical
(2) Bilateral single nodules
(3) Bilateral and asymmetrical
(4) Unilateral

## WHAT CAN YOU DO?

Unite. Join our support community comprised of more than 400 affected families from 32 countries. Educate yourself. Engage the conversation in social media. Donate to our cause and / or fundraise. Share stories. Buy and wear our PVNH branded clothing / accessories.

By educating yourself and others, you become an advocate. Begin by advocating for yourself and your child if a neuronal heterotopia disorder has affected you personally. Or you may advocate for a friend, or family member. In this process you will become an advocate for everyone affected by neuronal heteropia disorders to make lives better. We need your help.

You can talk / blog / post about PVNH. You can reach out to medical professionals or organizations that have an interest in PVNH. You can help us fundraise to support PVNH families.

On August 7, 2012, we started an annual tradition of recognizing everyone affected by neuronal heterotopia disorders, acknowledging World PVNH Disorder Awareness Day™ around the world. On this day, wear a pink and/or yellow piece of clothing / accessory and tell people why you are doing so. Take a photo and share it with friends and with us on social media to show you care. Interact with us, and other families affected by PVNH and other neuronal heterotopia disorders in social media.

**Alone we are rare, together we are strong.
And it is together that we will positively impact
the lives of families affected by PVNH
and other neuronal heterotopia disorders.**

**Unite. Educate. Advocate. Find a Cure.**

*Sources: Gene Reviews, National Library of Medicine, Genetics Home Reference, MedLink® Neurology