



PMGawareness.org

**Unlocking the Mysteries of Polymicrogyria**

# **Patient Brochure**



**PMG AWARENESS ORGANIZATION, INC.**

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## OUR MISSION

The PMG Awareness Organization is an international non-profit organization of families, caregivers, and medical professionals who care about those with Polymicrogyria. Our mission is to build a community of support to enhance the quality of life for those affected by Polymicrogyria through education, advocacy and promoting awareness.

## WHAT IS POLYMICROGYRIA?

Polymicrogyria (PMG), is a condition characterized by abnormal development of the brain before birth. While a baby is still in the womb, neurons are migrating to where they are supposed to go. For some reason, the neurons become disorganized and land in the wrong places. The surface of the brain normally has many ridges or folds, called gyri. In children born with Polymicrogyria, the brain develops too many folds, and the folds are unusually small. The name of this condition literally means too many (poly-) small (micro-) folds (gyria) in the surface of the brain. These small folds do not process the information it receives like a normally formed brain. This causes problems with functioning in the body much like how the effects of a stroke can affect the brain. The impairments that are seen as a result of PMG depend on what part of the brain is involved and how severe the deformity is.

There are several different forms of PMG (see the RESOURCES tab on our website for more information). Even though multiple children may be diagnosed with the same form, PMG affects every single child differently. Common problems associated with PMG in general are swallowing and speech difficulties, reflux, seizures of varying degrees (about 90% are affect with seizures at some point in their lives), development delays, lack of muscle coordination, impaired cognition of varying degrees and cerebral palsy, but there can be many others.

For some, PMG can also cause problems with vision, cognition, hearing, breathing and maintaining body temperature. Sometimes it comes with other diagnoses such as cerebral palsy, epilepsy, hydrocephalus, macrocephaly, and microcephaly (certainly not an all-inclusive list). It is important to note that many cases are mild enough to go undiagnosed and the child is labeled as "developmentally delayed" or having a "seizure disorder" or "cerebral palsy." It is



imperative to take the extra step to find out why a child is being labeled with these disorders and have them undergo an MRI (magnetic resonance imaging exam) to look at the brain. PMG, at the present time, can only be diagnosed through MRI results. It is also important to note that a specialized radiologist who knows what they are looking for should read the results of the MRI. Currently, many children are falling through the cracks and being misdiagnosed, especially when their symptoms are mild. This causes them to miss out on early medical intervention and therapies while their brains are still growing which could help them make significant more progress in the future.

## WHAT ARE THE SYMPTOMS OF POLYMICROGYRIA?

Symptoms of PMG can vary widely and depend on how much and what part(s) of the brain are affected. However, there are some symptoms that are fairly common in children with PMG. These include, but are not limited to:

- Difficulty with speaking, swallowing or chewing
- Epilepsy (recurrent seizures), mild to severe
- Developmental delays related to neurological impairment of the muscles or cerebral palsy
- Mild to severe intellectual disability

## WHAT CAUSES POLYMICROGYRIA?

Polymicrogyria can result from both genetic and environmental etiologies. It can occur as an isolated event, or as a symptom or part of other brain abnormalities. After years of collecting data and as research in this area has grown, experts say that the most common cause, probably by far, is related to a prenatal infection from a virus called Cytomegalovirus, also known as CMV. Polymicrogyria has also been recently found to be linked to the Zika Virus and is the most

common cause of PMG in Brazil and South America (see the RESOURCES tab for more information on both CMV and the Zika Virus).

Recent studies by experts have deemed the term "PMG" to refer to several different development disorders or birth defects of the brain that all have an irregular appearance of the brain surface in common. They have split "PMG" into different categories; classic PMG, Cobblestone Malformation, Tubulinopathy-associated dysgyria and other rare patterns. While all are rare, almost 50 genes have been associated with Polymicrogyria or PMG - like malformations, which fall into several groups listed below:

- **PMG Genes** (all very rare) – PAX6, FOXP2, BICD2, EOMES (TBR2), WDR62, NDE1, CEP135, PI4KA
- **Tubulinopathy Genes** – TUBA1A, TUBA8, TUBB2B, TUBB, TUBB3, DYNC1H1, KIF5C, KATNB1
- **Warburg Micro Syndrome Genes** – RAB3GAP1, RAB3GAP2, RAB18, TBC1D20
- **Cobblestone Malformation Genes with Congenital Muscular Dystrophy Plus** – AG1, POMT1, POMT2, POMGnT1, FKTN, FKRP, LARGE, B3GALNT2, B3GNT1, GALNT2, GTDC2, ISPD, TMEM5
- **Cobblestone Malformation Genes Other** – LAMA2, LAMB1, LAMC3, GPR56/ADGRG1, COL3A1, SRD5A3, ATP6V0A2, SNAP29
- **Brain Overgrowth and PMG Genes** – PIK3CA, PIK3R2, PTEN, AKT3, MTOR, CCND2



**In addition, PMG has been seen with two relatively common chromosome deletion syndromes:**

deletion 1p36.3, also known as Monosomy 1p36 and deletion 22q11.2, also known as DiGeorge Syndrome (*see the RESOURCES tab for more information about these syndromes*).

As internet research, MRI's and other imaging technology advances, so are the numbers of children who are being diagnosed with PMG. Unfortunately, a lot of children who have less severe cases of PMG frequently go undiagnosed. It is often not found unless there is a significant event in the child's life that leads to an MRI.

But even with an MRI, it can still go undiagnosed unless a specialized radiologist reads the MRI.

It is difficult to make a predictable prognosis because each individual is very unique in their presentation of this disorder.

## **WHY IS IT IMPORTANT TO GET A DIAGNOSIS?**

The answer to that question is multi-fold:

- Without a diagnosis, many children are not afforded proper medical care and other benefits
- Without accurate statistical data, the number of cases seem smaller and therefore funding for research and treatment is poor
- As more children are diagnosed, the public becomes more aware of their needs
- Early diagnosis is CRITICAL – delay of diagnosis often means that therapy and treatment during the “golden era of childhood development” (the first three years) is missed, leaving the child with a greater chance of permanent loss of developmental goals
- EVERY parent/family of an undiagnosed child deserves to have an answer!



## **HOW IS POLYMICROGYRIA TREATED?**

Treatment and management of PMG depend on the needs of the individual. Commonly used, but not limited to, are physical therapy for gross motor impairment, orthotic devices and surgery for patients with spastic motor impairment. Speech therapy is widely used for language and feeding impairment. Augmentative and alternative communication devices are also commonly used to aide in speech deficits. Occupational therapy is used for fine motor difficulties and antiepileptic drugs are used to treat and control seizure.



## **UNLOCKING THE MYSTERIES OF POLYMICROGYRIA**

### **HOW YOU CAN HELP:**

PMG Awareness Organization consists of an all-volunteer Board of Directors who can always use more dedicated and passionate people! The organization is always looking for people to lead or join various committees. There are many great things in the works and many annual events that take place every year, which take a lot of preparation and planning such as our Annual Virtual 5K Run/Walk Event, Rare Disease Day, Giving Tuesday, etc. It is our desire and goal to hold a bi-annual conference to bring PMG families and medical professionals together to discuss PMG and help find ways to advance research. But we **NEED YOUR HELP!**

If you are interested in volunteering in some capacity, please contact us! We want to hear from you, and we encourage you to join our cause and spread the word in any way you can.

As always, any monetary contribution is greatly needed and appreciated. You can make a tax-deductible donation to the organization on our website.

### **FOLLOW PMGA ON SOCIAL MEDIA:**



**Please feel free to print out this Patient Brochure and take it with you to all doctors and therapy appointments.**

This information will let them know our organization exists and will help them provide others diagnosed in the future with hope, encouragement and most of all, support!