Polymicrogyria

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Description

Polymicrogyria is a condition characterized by abnormal development of the brain before birth. The surface of the brain normally has many ridges or folds, called gyri. In people 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.Polymicrogyria can affect part of the brain or the whole brain. When the condition affects one side of the brain, researchers describe it as unilateral. When it affects both sides of the brain, it is described as bilateral. The signs and symptoms associated with polymicrogyria depend on how much of the brain, and which particular brain regions, are affected.Researchers have identified multiple forms of polymicrogyria. The mildest form is known as unilateral focal polymicrogyria. This form of the condition affects a relatively small area on one side of the brain. It may cause minor neurological problems, such as mild seizures that can be easily controlled with medication. Some people with unilateral focal polymicrogyria do not have any problems associated with the condition.Bilateral forms of polymicrogyria tend to cause more severe neurological problems. Signs and symptoms of these conditions can include recurrent seizures (epilepsy), delayed development, crossed eyes,

problems with speech and swallowing, and muscle weakness or paralysis. The most severe form of the disorder, bilateral generalized polymicrogyria, affects the entire brain. This condition causes severe intellectual disability, problems with movement, and seizures that are difficult or impossible to control with medication.Polymicrogyria most often occurs as an isolated feature, although it can occur with other brain abnormalities. It is also a feature of several genetic syndromes characterized by intellectual disability and multiple birth defects. These include 22q11.2 deletion syndrome, Adams-Oliver syndrome, Aicardi syndrome, Galloway-Mowat syndrome, Joubert syndrome, and Zellweger spectrum disorder.

Frequency

The prevalence of isolated polymicrogyria is unknown. Researchers believe that it may be relatively common overall, although the individual forms of the disorder (such as bilateral generalized polymicrogyria) are probably rare.

Causes

In most people with polymicrogyria, the cause of the condition is unknown. However, researchers have identified several environmental and genetic factors that can be responsible for the disorder. Environmental causes of polymicrogyria include certain infections during pregnancy and a lack of oxygen to the fetus (intrauterine ischemia).Researchers are investigating the genetic causes of polymicrogyria. The condition can result from deletions or rearrangements of genetic material from several different chromosomes. Additionally, mutations in one gene, ADGRG1, have been found to cause a severe form of the condition called bilateral frontoparietal polymicrogyria (BFPP). The

ADGRG1 gene appears to be critical for the normal development of the outer layer of the brain. Researchers believe that many other genes are probably involved in the different forms of polymicrogyria.

Learn more about the genes associated with Polymicrogyria

ADGRG1

TUBB2B

Inheritance

Isolated polymicrogyria can have different inheritance patterns. Several forms of the condition, including bilateral frontoparietal polymicrogyria (which is associated with mutations in the ADGRG1 gene), have an autosomal recessive pattern of inheritance. In autosomal recessive inheritance, both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.Polymicrogyria can also have an autosomal dominant inheritance pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Other forms of polymicrogyria appear to have an X-linked pattern of inheritance. Genes associated with X-linked conditions are located on the X chromosome, which is one of the two sex chromosomes. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to

their sons.Some people with polymicrogyria have relatives with the disorder, while other affected individuals have no family history of the condition. When an individual is the only affected person in his or her family, it can be difficult to determine the cause and possible inheritance pattern of the disorder.

Other Names for This Condition

PMG

Additional Information & Resources

Genetic Testing Information

Genetic Testing Registry: Bilateral frontoparietal polymicrogyria

Genetic Testing Registry: Bilateral parasagittal parieto-occipital polymicrogyria

Genetic Testing Registry: Complex cortical dysplasia with other brain malformations 7

Genetic and Rare Diseases Information Center

Bilateral frontoparietal polymicrogyria

Bilateral perisylvian polymicrogyria

Bilateral frontal polymicrogyria

Bilateral generalized polymicrogyria

Bilateral parasagittal parieto-occipital polymicrogyria

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Clinical Trials

ClinicalTrials.gov

Catalog of Genes and Diseases from OMIM

POLYMICROGYRIA, BILATERAL PERISYLVIAN, X-LINKED; BPPX

CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS 14A (BILATERAL FRONTOPARIETAL); CDCBM14A

CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS 7; CDCBM7

POLYMICROGYRIA, BILATERAL TEMPOROOCCIPITAL; BTOP

Scientific Articles on PubMed

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