WHAT IS HEMIMEGALENCEPHALY?

Understanding a rare condition

Introduction

Hemimegalencephaly is an extremely rare condition where one half of the cerebral cortex is larger than the other. The neurons in the enlarged hemisphere are typically malformed. It can include various types of cortical dysplasias and other brain malformations. Often, the corpus callosum is poorly formed or absent. The ventricles of the brain may be enlarged as well due to hydrocephalus .

Types of Hemimegalencephaly

- Isolated hemimegalencephaly: Only the cerebral cortex is involved with no associated unilateral body overgrowth, skin involvement, or systemic involvement of the organs;
- Syndromic hemimegalencephaly: Not only is the brain involved, but also the skin and some organs. Conditions such as linear nevus syndrome, “cafe au lait” spots, ash leaf marks , and Hypomelanosis of Ito. Skin involvement may take months or years after birth to appear. Hemihypertrophy , where one half of the body is larger than the other, may also be present. In rare cases, syndromic hemimegalencephaly can include facial infiltrating lipomatosis, Proteus syndrome, and issues with collagen which may lead to hyperextensible joints and other challenges.
- Total hemimegalencephaly: Includes enlargement of cerebellum and sometimes brain stem and can be isolated or syndromic.

Cause

Hemimegalencephaly is caused by a spontaneous mutation which occurs on the mTOR pathway at approximately the third week of gestation. It is genetic but is not hereditary. There are no reported cases of two or more children with hemimegalencephaly in one family or that it is caused by poor maternal diet or ingestion of any substance during pregnancy.

Diagnosis

Hemimegalencephaly is very difficult to diagnose in utero. Ultrasound imaging can only detect some, but not all, forms of brain malformations. Because it is a spectrum condition it can often go undetected in utero.

Seizures

In most cases, hemimegalencephaly will cause the child to have seizures. Seizures can begin in very early infancy and are often drug-resistant. Some children may not have any seizures associated with hemimegalencephaly.

There are many different types of seizures associated with hemimegalencephaly. It is important to note here that seizures in childhood, especially in early infancy, can be catastrophic. Stopping seizures as soon as possible is imperative.

A small number of children are able to maintain relative seizure control throughout their lives, but most will have drug-resistant epilepsy that requires brain surgery.

The shorter time seizing before surgery, the more likely the child will have a better developmental outcome .

Prognosis

Hemimegalencephaly is a spectrum condition, meaning there is a wide range of functional outcomes that are poorly reported in research literature. Most children with hemimegalencephaly have some level of intellectual disability. Some can attain near normal intellectual development if seizures are controlled. Others may have profound intellectual and motor disabilities. All children with hemimegalencephaly have hemiparesis, a type of cerebral palsy which affects one side of the body, which may range from very mild to severe. Some may have homonymous hemianopsia, a visual impairment where half the visual field is lost. Most are able to walk and talk, but not all. Some may be tube fed while others can eat a normal diet orally. Like any spectrum condition, there are outliers.

Functional outcomes are closely tied to seizure control and whether the brain malformation is present on both sides of the brain. It is extremely important that the child’s neurologist is proactive in ensuring that the child has no seizures. Epilepsy surgery – most often hemispherectomy – should be considered early if seizure control is not gained.

How young can hemispherectomy be performed?

Hemispherectomy is an extremely delicate and difficult brain surgery which should only be performed by an experience pediatric epilepsy surgeon. The youngest reported case is 41 days; however, anecdotally, an infant as young as 28 days has had hemispherectomy surgery for hemimegalencephaly.

Hemispherectomy for hemimegalencephaly is particularly difficult for many reasons: often the blood vessels are malformed, making them difficult to locate and cut during surgery, and can sometimes cause excessive bleeding. Also, the overall brain malformation makes surgery difficult because the familiar landmarks the surgeon uses to identify various structures are often missing. The epilepsy surgeon should not only have experience performing hemispherectomy, but should also be experienced performing hemispherectomy on infants with hemimegalencephaly.

> *The Hemimegalencephaly Family Support Network is the world’s largest support group for parents of children with hemimegalencephaly. It offers a website and Yahoo! group, but its most active support platform is now found in this closed Facebook group which includes parents from all over the world.*