Agenesis of the Corpus Callosum

BIRTH DEFECT RESEARCH FOR CHILDREN

What is Agenesis of the Corpus Callosum (ACC)?

Agenesis of the Corpus Callosum means a complete or partial absence of the bridge between the left and right sides of the brain. In and of itself, the corpus callosum is not necessary for life or health, but conditions associated with ACC can lead to serious medical problems including abnormalities of cerebrospinal fluid or mental retardation.

In a normal brain, a huge mass of nerve fibers, called the corpus callosum, connects the two hemispheres (halves) of the brain. The fibers of the corpus callosum start growing when the fetus is about 3½ months old, and by 5 months the corpus callosum has usually reached the general shape and structure found in an adult brain.

Agenesis of the Corpus Callosum

How often does ACC occur?

The occurrence among the general population is unknown, but in the Saguenay-Lac region of northeastern Quebec the incidence was 1 in 2117 live births. This may be due to a unique mutation that causes 1/23 residents to be carriers.

What causes agenesis of the corpus callosum?

Agenesis of the corpus callosum may have genetic causes. It has been found in a recessive form in several studies. Other studies suggest that some sort of in-utero interference with the developing brain (perhaps vascular in nature) may sometimes be the cause. ACC is not usually a single malformation, but typically involves other syndromes such as microcephaly (abnormally small brain or head), growth retardation, or recurrent bronchopneumonia.

How do the two types of agenesis of the corpus callosum differ?

Two primary types of ACC have been recognized.

- Type I, the most common, is not associated with other disorders, and is characterized by absent to mild symptoms, normal intelligence to mild/moderate retardation, and some impairment of visual, motor, and coordination skills.

- Type II is associated with other disorders and is usually characterized by severe symptoms that may include mental retardation, seizure disorder, hydrocephaly (water on the brain), microcephaly (abnormally small head), spasticity (spasms), failure to thrive, and other complications.

How is ACC detected?

Diagnosis of agenesis of the corpus callosum can often be difficult. Ultrasonography can often identify ACC in the second trimester of pregnancy (18-20 weeks gestation). Magnetic Resonance Imaging can provide further information, especially in the case of late detection, around 28-30 weeks gestation.

The development of 3D echographic imaging offers an even more precise prognosis.

In children who have not been diagnosed prenatally and who have no immediate symptoms, ACC may remain undetected, or may not be discovered until later in life.

What treatment is available for ACC?

Most doctors recommend periodic check-ups by specialists in pediatrics and neurology to monitor associated conditions like seizure disorder or hydrocephaly (a build up of fluid in brain).

Associated disorders such as seizures, hydrocephaly, mental retardation and/or spasticity may require treatment with medication, shunting, special rehabilitation, and/or physical therapy.

Prognosis

Most children do not die because of the absence of the corpus callosum, and if they do experience mental retardation, it is usually non-progressive. Some affected children appear to be free of any symptoms; yet testing may reveal defective integration for performance of visual or fine motor tasks.

Fact Sheet by:

Birth Defect Research
Children, Inc.
www.birthdefects.org