

Open neural tube defects (ONTDs), which includes spina bifida and anencephaly, is a range of <u>birth defects</u> that can include the brain and/or spinal cord. This is generally something that happens very early in pregnancy, and can vary widely in severity and expected outcome. Although the cause of most cases of ONTDs are not known, there are a few known risks factors, such as maternal obesity, uncontrolled maternal diabetes, and exposure to certain antiseizure medications. Taking folic acid (a type of vitamin B) before and during pregnancy has been shown to reduce the risk for ONTDs during pregnancy.

ONTDs can often be diagnosed by <u>ultrasound</u>, but may also be identified by doing a <u>diagnostic procedure</u>, such as <u>amniocentesis</u>.

The two most common types of ONTDs are spina bifida and anencephaly. Spina bifida occurs when the bones in the spine do not fuse correctly. This can cause part of the spinal cord to bulge through the spine. There is a wide variability with spina bifida: some individuals may have a very small hole and may not even know they have it, while others can have more severe openings that cause significant medical complications. The size of the opening and where it is on the spine can sometimes give more information as to what to expect. To learn more about spina bifida, visit the Spina Bifida Association on their <u>web site</u>.







While spina bifida can be quite variable, anencephaly is almost always life-limiting. Anencephaly is when all or part of the brain is missing. Most babies with anencephaly do survive to delivery, and those who do often pass away shortly after birth.





Click <u>here</u> to learn more about scheduling a genetic counseling appointment for pregnancyrelated questions.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Related Articles

• Down Syndrome

Down syndrome, also known as trisomy 21, is a genetic condition caused by an extra copy of the 21st chromosome. Each individual with Down syndrome is unique. Individuals with Down syndrome have variable health issues, including developmental delays and some degree of intellectual disability. What is life like for people...

• <u>Trisomy 18 (Edwards Syndrome)</u> Trisomy 18, also known as Edwards syndrome, is a genetic condition caused by an extra chromosome 18. Babies with trisomy 18 generally have many complex medical complications, including heart defects, growth restriction, a small abnormally shaped head, and clenched fingers with overlapping fingers. Because of the severity of medical complications...



• Trisomy 13 (Patau Syndrome)

Trisomy 13, also known as Patau syndrome, is a genetic condition caused by an extra chromosome 13. Babies with trisomy 13 generally have many complex medical complications, including heart defects, brain and spinal cord abnormalities, very small or poorly developed eyes, cleft lip and/or cleft palate, and low muscle tone...

• <u>Sex Chromosome Variations</u> X and Y chromosome variations are conditions that result when a baby have a different number of sex chromosomes, also known as X & Y chromosomes, than we would expect to see in some or all of the cells in their body. Typically, girls have two X chromosomes while boys...

<u>Microdeletion Syndromes</u>

There are many different kinds of genetic conditions. Some are caused by whole extra or missing chromosomes (such as Down syndrome), while others are caused by changes in a specific gene (such as sickle cell anemia). There is a third group of genetic conditions called microdeletion syndromes. A microdeletion is...