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Presenting Cases of Delayed Diagnosis or Missed Diagnosis of Spina Bifida and Other Serious Birth Defects

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Last year, approximately 3.8 million babies were born in the U.S. Approximately 120,000 of these babies were affected by some form of birth defect, meaning roughly every 4-and-a-half minutes, a baby with a birth defect is born. Some birth defects are more serious than others. Some serious birth defects can be treated during the prenatal time period, providing significant improvement in outcomes for these children. For babies to receive treatment for birth defects during the prenatal time period, their conditions must be diagnosed correctly and timely. Failure to diagnose birth defects used to result in no more than unprepared parents. Now, this failure to diagnose can result in loss of opportunity to improve quality of life.

Presently, the most common birth defects are heart defects, cleft lip/palate, Down's syndrome and spina bifida. Spina bifida is the most serious birth defect that does not affect a child's life expectancy, which has made treatment of this condition the focus of prenatal surgeons across the country.

WHAT IS SPINA BIFIDA?

Spina bifida means "cleft spine." It is a congenital defect that occurs when the infant's spine and spinal cord are not completely formed during the first few weeks of pregnancy.

When an unborn child's neural tube does not develop properly during pregnancy, the baby experiences complications with their vertebrae and spinal cord. Spina bifida ranges in severity with the mildest form being "occulta" (meaning hidden) and the most severe form being "myelomeningocele."

This most severe form of spina bifida causes the spinal cord and nerves to be exposed through an opening in the spine in the lumbar or thoracic regions. The exposure of the baby's nerves and spinal cord to the mother's amniotic fluid causes these tissues to become injured, often result in paralysis. The hole or opening in the spine disrupts the pressure and flow of cerebral spinal fluid, and some babies develop a buildup of fluid in their brains called "hydrocephalus."

HOW SERIOUS IS SPINA BIFIDA?

The effects of spina bifida vary based upon the severity of the defect. Those with mild spina bifida ("occulta") may suffer from little to no complications, while those with myelomeningocele suffer from varying degrees of:

- Mobility issues due to damage to the spinal cord
- Orthopedic problems such as scoliosis
- Problems with bowels and bladder control
- Tethered spinal cord
- Breathing problems during sleep
- Cognitive deficits
- Hydrocephalus (often requiring multiple brain surgeries)

Fatalities from spina bifida are rare but can occur from infections (such as meningitis) or other infections that occur following surgeries performed on the brain to treat hydrocephalus.

RISK FACTORS FOR SPINA BIFIDA

Although the causes of spina bifida are unknown, research indicates the following risk factors may contribute to the development of the birth defect:

Folate deficiency. Folate, the natural form of vitamin B-9, is important to the development of a healthy baby. The synthetic form, found in supplements and fortified foods, is called folic acid. A folate deficiency has been found to increase the risk of spina bifida and other neural tube defects.

Family history of neural tube defects. Couples who have had one child with a neural tube defect have a slightly higher chance of having another baby with the same defect. However, most babies with spina bifida are born to parents with no known family history of the condition.

Some medications. For example, anti-seizure medications, such as valproic acid (Depakene), have been found to increase the occurrence of neural tube defects when taken during pregnancy. This might happen because they interfere with the body's ability to use folate and folic acid.

Diabetes. Women with diabetes who do not have well-controlled blood sugar have a higher risk of having a baby with spina bifida.

Obesity. Pre-pregnancy obesity is associated with an increased risk of neural tube birth defects, including spina bifida.

Increased body temperature. Some evidence suggests that increased body temperature (hyperthermia) in the early weeks of pregnancy may increase the risk of spina bifida. Elevating your core body temperature due to fever or using a sauna or hot tub has been associated with a possible slightly increased risk of spina bifida.

If you have any of the known risk factors for spina bifida, talk with your doctor before you decide to become pregnant to determine if you need a larger dose or prescription dose of folic acid, even before a pregnancy begins.

If you take medications, tell your doctor. If you plan ahead, some medications can be adjusted before your pregnancy to diminish the potential risk of spina bifida.

FAILURE TO DIAGNOSE SPINA BIFIDA

Failure to timely diagnose spina bifida can result in the inability to treat the most serious form of this birth defect, myelomeningocele.

Spina bifida should be diagnosed during a first-trimester ultrasound or at the 20-week fetal anatomy ultrasound. Early signs of this birth defect can also be detected with blood tests. If a timely diagnosis of spina bifida is made, patients can be screened to determine if they are candidates for prenatal surgical treatment of the birth defect.

For some patients, the most severe form of spina bifida can be treated with fetal surgery to close the fetal spine before the baby is even born. Fetal surgery must be performed between approximately 20 to 26 weeks of gestation, making timely diagnosis critical. Babies treated surgically during the prenatal time period are expected to have significantly improved outcomes compared to children who are treated surgically after they are born.

Babies treated with prenatal surgery experience significantly less occurrence of hydrocephalus (which decreases need for brain shunting surgeries) and increased ability to walk with or without assistive devices.

Given the clear guidelines for diagnosis of spina bifida during the prenatal time period, the failure to diagnose this birth defect using ultrasound and/or blood tests is usually considered negligent. In cases where the diagnosis of spina bifida is missed or delayed beyond the time period when fetal surgery can correct the defect, the injured infant/parent may have cause to file a legal action for monetary compensation.

WHEN TO FILE A LAWSUIT

If your newborn is suffering from spina bifida or another serious birth defect that was undetected until after he or she was born, your child may have a claim for damages for delayed or missed diagnosis.

To succeed in such a claim, the State of Colorado requires a claimant (plaintiff) to have expert opinions to support claims of liability (who is at fault), causation (what harm did the mistake cause) and damages (the amount of harm caused by the mistake).

The development of damages cases in spina bifida or other missed birth defect cases is complex involving the need to prove which medical treatments and expenses could have been avoided if early detection and treatment occurred. Medical malpractice attorneys must work with top professionals and experts to explore potential claims and damages related to missed or delayed diagnosis of serious birth defects such as spina bifida.

These claims take significant time to investigate, making it important for parents to contact an attorney quickly after learning of diagnosis of birth defect. •

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