

Neural Tube Defects

BIRTH DEFECT RESEARCH FOR CHILDREN



What is Neural Tube Defects?

(Anencephaly, Encephalocele, Spina Bifida)



Neural Tube Defects



Neural Tube Defects are serious birth defects which involve incomplete development of the brain, spinal cord and/or the protective coverings of the organs. There are three types of NTDs — anencephaly, encephalocele, and spina bifida.

Anencephaly: Babies born with this birth defect have underdeveloped brains and incomplete skulls. Most infants born with anencephaly do not survive more than a few hours after birth.

Encephalocele: A birth defect that results in a hole in the skull through which tissue protrudes. Although in the past, most babies with encephalocele do not live, today some children survive because of early surgery to correct this defect. Children who survive may be severely retarded.

Spina Bifida: A birth defect of the spinal column (backbone) that is sometimes called “cleft or open spine”. Spina Bifida can range from a mild defect that causes no problems to a serious condition involving muscle paralysis, loss of feeling, infection and loss of bowel and bladder control. 70-90% of children born with the more severe types of spina bifida will also have hydrocephalus, a build up of fluid in the brain. Hydrocephalus is controlled by surgically implanting a drain or “shunt” to relieve the fluid pressure on the brain.

There are three types of spina bifida:

1. **Spina Bifida Occulta:** An opening in one or more of the bones of the spinal column which does not involve any damage to the spinal cord. Approximately 40% of all Americans have this physical difference without even knowing it.
2. **Meningocele:** A more serious form of spina bifida in which the meninges, the protective covering around the spinal cord, pushes out through an opening in the spinal column. The sac containing the exposed part of the spinal cord is called the “meningocele”. Early surgery can usually repair this type of spina bifida with little or no damage to the nerves in the spinal cord.

3. **Myelomeningocele:** The most severe form of spina bifida. The spinal cord containing the deeper nerves of the spinal column protrudes from the open spine without a protective covering of the skin. Spinal fluid may leak out and infection can be a serious problem. This defect usually occurs at the lower end of the spine resulting in paralysis of the baby’s legs along with poor bladder and bowel control.

How many children are born with Neural Tube Defects?

In North America, NTD’s occur in one or two out of every 1,000 births. The risks increase to 2% if you have already had one child born with an NTD. A family history of NTDs also increases the possibility of having a child with one of these defects. If you or your partner has an NTD, your risk of having a child with spina bifida, anencephaly or encephalocele increases to between 3-5%.

What Causes NTD’s

Neural Tube Defects are in a category of birth defects called polygenic or multifactorial. This means that NTDs are caused by one or more genes interacting with some triggering factor. Environmental triggers for NTDs that are being studied include viruses, vitamin and mineral deficiencies, chemicals, drugs, and maternal illness like diabetes.

How can you help a child with a NTD?

Prenatal testing: Early intervention is important. If you know ahead of time that your baby may be born with an NTD, you can arrange for prompt and expert medical treatment. A blood test has been developed which can help determine whether an unborn baby has a serious NTD. The AFP screening test measures the amount of a substance called alpha-fetoprotein in the mother’s blood. The most NTDs will often leak AFP resulting in an increase in AFP levels in the mother’s bloodstream.



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The AFP test should be performed between 16 and 18 weeks of pregnancy. The serum AFP screening test may detect more than 90% of the cases of anencephaly and 64-80% of babies with spina bifida. Combining AFP screening with ultrasound and amniocentesis (a test of the fluid surrounding the baby) increases the reliability of the test.

Medical treatment: When a baby is born with one of the more severe forms of spina bifida, surgery is usually performed within 24 hours of birth. Doctors try to remove the cyst protruding from the spine and cover the wound with muscle and skin. Even this, however, may not prevent paralysis of the legs and lack of feeling. Fluid must be drained from the brains of babies who develop hydrocephalus. A special tube called a shunt is inserted to help this excess fluid pass out of the body.

Physical therapy: It is extremely important for children with spina bifida to begin physical therapy. The therapist teaches parents how to exercise their baby's feet and legs to prepare for walking with leg braces and crutches.

Some children will need instruction to use wheelchairs. The physical therapist can also help parents teach children bladder and bowel control.

Support groups: National organizations provide the latest information on spina bifida and other NTDs through newsletters, brochures, and conferences as well as training in rehabilitation. By joining local support groups, parents of children with NTDs can share experiences and helpful strategies on coping with the challenges of raising their children.

Education: It is important for your child to be placed in the least restrictive school setting as early as possible. Children with spina bifida may require special building modifications to attend school in wheelchairs or wearing braces. Federal funds have been provided throughout the nation to help renovate school buildings with ramps and elevators and other structural changes that make it easier for handicapped children to attend regular classes.

Can NTDs be prevented?

Recent research has shown that taking vitamins with the B vitamin folic acid prior to and during early months of pregnancy may significantly reduce the occurrence of Neural Tube Defects. Folic acid is also in dark leafy greens, beans, peas and in many fortified foods like cereals and bread.

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