

### What are “neuromuscular diseases”?

This term covers a number of disease patterns deriving primarily from neurological or muscular diseases and characterized by a variety of symptoms of different degrees of severity (such as disorders affecting posture and the locomotor system, mental disabilities, or damage to sensory perception). Many of these diseases have their onset in childhood and may, in addition to the large number of prevailing individual symptoms, lead to the development of a scoliosis over the course of the disease due to neuromuscular damage to the postural apparatus.

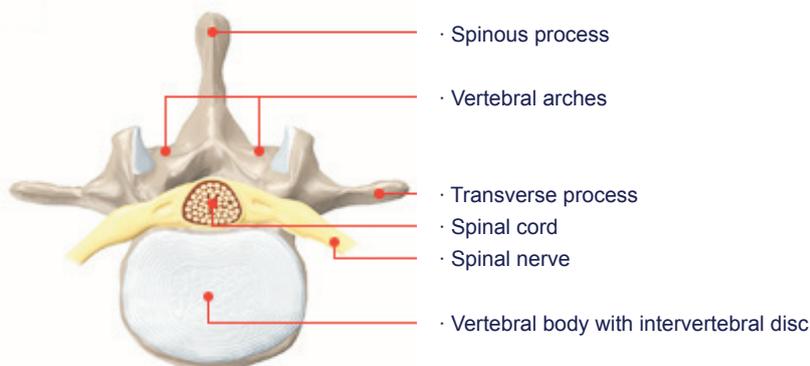
### What is meningomyelocele?

This is a congenital disease in which a defect in the embryonal formation of the neural tube results in an inhibition malformation of the central nervous system, where parts of the meninges and spinal cord (myelon) emerge from the incompletely closed vertebral arches and may lie open and unprotected. Every year, around 500 children are born in Germany with a neural tube defect.

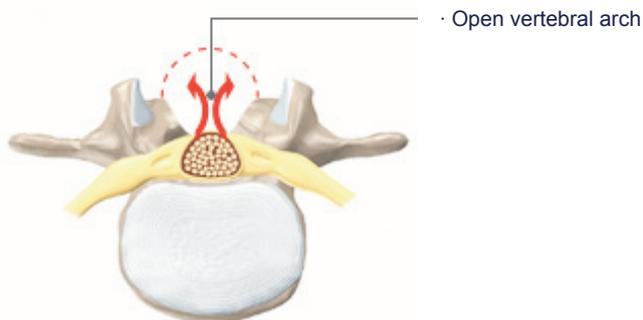
### How does meningomyelocele develop?

During embryonal development, between the 20th and 28th week of pregnancy, the neural tube is formed from the medullary plate. The neural tube then develops into the brain, spinal cord, spinal column and skull. To understand this process of organ formation, imagine folding a sheet of paper (the medullary plate) until the edges of the sheet touch and fuse, creating a tube (the neural tube). In a neural tube defect the tube does not close completely, resulting in damage affecting the further development and formation of the brain, spinal cord and spinal column.

#### • Lumbar vertebra, view from above



- Open vertebra arch, through which parts of the spinal meninges or spinal cord may emerge towards the back, unprotected.



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Due to the incomplete closure of the neural tube, the vertebrae also do not form completely and the vertebral arches remain open. Portions of the spinal meninges or spinal cord may emerge dorsally (towards the back) through these openings. A spinal column with open vertebral arches is known as a spina bifida (“split or open spine”). If the spinal cord and spinal meninges lie open and unprotected as well, the term spina bifida aperta is used. If the spinal column and spinal cord deformity is not visible under the protective skin (i.e. “hidden”), the term spina bifida occulta is used.

If only the spinal meninges emerge from the opening and form a sac bulging out from the back that is filled with liquor (cerebral fluid), this is called a meningocele.

If the spinal meninges and spinal cord (myelon) both bulge out toward the back through the open vertebral arches, this is called a meningomyelocele.

The protuberance of the spinal meninges and the spinal cord inhibits the circulation of the liquor, resulting in cerebral fluid congestion in the cerebral ventricles, a condition known as hydrocephalus (“water on the brain”). If the neural tube does not close in the segment from which the brain is supposed to develop, the resulting condition is termed anencephaly, i.e. the brain is not formed.

Depending on the location and extent of the defect, varying levels of paralysis occur in the arms, legs, intestine, and urinary bladder. Newborns with anencephaly are not viable.

### What are the causes of meningomyelocele?

The pathogenic causes of neural tube defects (spina bifida, meningocele, meningomyelocele, and anencephaly) have not yet been clarified. The following are under discussion as causal factors:

- Genetic factors
- Folic acid metabolism

Folic acid is a water-soluble B-complex vitamin (Vitamin B9). It plays an important role in embryonal development of the brain, spinal cord, and spinal column. It was assumed for quite a long time that a folic acid deficiency was responsible for the development of neural tube defects. More recent studies have shown that many neural tube defects develop at normal blood folic acid levels, so that a folic acid deficiency is less likely to be the cause. It is presumed that a dysfunction in folic acid metabolism or the formation of autoantibodies to the folic acid receptors hinder the integration of folic acid into the cells. These two possible causes can be compensated for if a woman planning a pregnancy takes of folic acid supplements to reduce the probability

of the occurrence of a neural tube defect. Since defects of the neural tube are occur during the first weeks of a pregnancy, it makes sense to supplement folic acid early when planning a pregnancy, such as immediately after discontinuing birth control pills.

### How are neural tube defects diagnosed?

Neural tube defects can be detected in ultrasonic images as early as the 10th week of pregnancy. Between the 16th and 18th weeks of pregnancy the alpha-1-fetoprotein (AFP) test is carried out on the mother's blood or amniotic fluid. AFP is a protein produced by the fetus. An elevated AFP level indicates a neural tube defect.

### What are the symptoms of meningomyelocele or spina bifida?

- High incidence in lumbar spine or sacrum
- Varying degrees of paralysis, complete and partial, dysesthesias depending on localization of findings and extent of spinal cord damage. The paralyzes cause imbalances in the skeletal muscles, in turn resulting in shortened muscles (contractures) and abnormal positioning of the hip, knee and foot joints. Meningomyelocele is very frequently associated with congenital anomalies, which significantly contribute to the progression of a spinal column malposition.
- Dysfunction of the voiding of urinary bladder and colon with frequent urinary tract infections.
- Impaired sexual function.
- In children with spina bifida, the Arnold Chiari syndrome may develop as an additional anomaly, i.e. a complex anomaly in the cranial fossa and at the transition from brain to spinal column, in most cases accompanied by meningomyeloceles in the lumbar spine and hydrocephalus.
- Hydrocephalus (water on the brain)

The liquor (cerebral fluid) is produced in the 4 cerebral ventricles in the interior of the brain and serves to protect the brain and spinal column like a cushion. The brain and the spinal cord are surrounded by the soft meninges - the pia mater and arachnoid membrane - and from the hard meninx - the dura mater. The space between the two soft cerebral and spinal meninges is called the subarachnoid space. The liquor produced in the cerebral ventricles flows through the subarachnoid space to the spinal canal, where it flows around the spinal cord. If the flow of this fluid is hindered, for example by a meningomyelocele, liquor congestion may expand the cerebral ventricles, potentially exerting pressure on the surrounding brain structures and damaging nervous tissue. In infants and small children, whose cranium is not yet closed (open fontanelles), hydrocephalus is characterized by an increase in the size of the head and a bulge at the fontanelles. Potential causes of hydrocephalus besides spina bifida are tumors, inflammations, or hemorrhages in the brain or spinal cord. In a case of congenital hydrocephalus, the increased cerebral pressure must be surgically reduced immediately after birth so as to avoid further damage to the brain. To this end, a venting tube system is inserted into the cerebral ventricles. The excess liquor drains through the tube and under the skin into the abdominal cavity where it can be resorbed.

This system of tubes is known as a ventriculoperitoneal shunt.

- Development of a cerebral seizure condition (epilepsy)
- Tethered cord

Nearly all patients with a meningomyelocele show adhesions of the spinal cord with surrounding tissues in MRT images and approximately 40% of children with this condition experience worsening neurological symptoms over the course of further development caused by tensile pressure on the spinal cord resulting from longitudinal growth.

- Decubital ulcers on the back due to increasing gibbus formation (hump) and on the trochanter major of the thigh bone.

- Orthopedic symptoms

- The course of development of a scoliosis depends on the extent of the vertebral arch defects and the spinal level of the paralysis. It normally develops in the thoracolumbar (transition from thoracic to lumbar spine), lumbar or lumbosacral (transition from lumbar spine to sacral region) areas, with a compensatory contralateral curve above the paralyzed area. If the scoliotic and kyphotic curvature of the spine progresses the acquired ability to sit and walk may be lost. In the lungs, this condition may be accompanied by respiratory dysfunction with frequent pulmonary infections caused by the increasing pressure of the deformed ribcage on the lungs.
- Gibbus (hump) formation caused by increasing kyphotic deformation of the spinal column with the risk of decubital ulcer formation (decubitus)
- The paralysis of the skeletal muscles results in malpositions and contractures in the hip, knee and foot joints.
- Reduced levels of calcium carbonate in the bones due to inactivity (osteoporosis) with an increased risk of bone fractures.

### How is spina bifida/meningomyelocele treated?

The most important goal of treatment of children with spina bifida is to provide them with the maximum level of mobility and independence possible through optimized therapeutic concepts. This complex disease therefore requires a cooperative effort by a team of specialists (pediatric surgeon, neurosurgeon, pediatrician, neurologist, urologist, physiotherapist, logotherapist, ergotherapist, and orthopedic technician).

- **Closure of the “open spine”**

Children born with spina bifida must be operated on immediately after birth. The open spine must be closed within the first 24-48 hours after birth if possible, since otherwise the risk of infection of the spinal cord and spinal meninges is too great. In this operation, the parts of the spinal cord and spinal meninges protruding into the meningomyelocele are gently pushed back into the spinal canal. The defect is then covered with muscles, muscle sheath, and skin. Plastic surgery methods may be required to close larger defects.

For a number of years now, it has been possible to close the open spine in a fetal surgery procedure done between the 18th and 25th week of pregnancy. The operation can be carried out in open surgery through the abdominal wall and womb, similar to the procedure used in a cesarean section, or in the closed womb using minimally invasive techniques.

- **Treatment of hydrocephalus**

If the child also suffers from hydrocephalus, a shunt must be implanted to counteract the liquor congestion and increased cerebral pressure. This shunt consists of a system of venting tubes, which connect the cerebral ventricles and the abdominal cavity. The tube is inserted into a cerebral ventricle through a burr hole (a drilled hole), then placed under the skin to drain off the liquor, usually into the abdominal cavity (ventriculoperitoneal shunt). When the cerebral pressure rises, the excessive cerebral fluid runs through the shunt into the abdominal cavity, where it is resorbed.

The shunt system regulates the cerebral pressure so as to avoid further damage to the child's brain. In some cases in which a peritoneal connection is not feasible, the shunt can be laid to the atrium of the heart (ventriculoatrial shunt).

**· Treatment of scoliosis and gibbus formation**

If the spinal column curvature increases, children with neuromuscular diseases such as spina bifida should undergo early corrective surgery before they are done growing in order to preserve the ability to sit and prevent further damage to the spinal column and progressive worsening of cardiovascular and respiratory function.

**· Treatment of the “tethered cord” syndrome**

Should increasing neurological symptoms arise from tension on a spinal cord fixed by adhesions, a gentle release of the spinal cord adhesion may become necessary (neurolysis).

**· Treatment of contractures and malpositions**

Tendons and muscles often become foreshortened (contractures) during the course of the illness, which may cause a considerable loss of function. In such cases, surgical tendon lengthening or transections (tenotomies) are carried out to improve mobility. In rare cases, corrective surgery of the hip, lower legs or foot deformities may be required.

**· Treatment of urinary bladder paralysis**

This type of paralysis results in frequent urinary tract infections, which is why either a special technique for emptying the bladder, or autocatherization, should be learned.

**· Physiotherapy**

Improvement and preservation of locomotor function to achieve a maximum level of independence for affected children requires the implementation of a wide variety of physiotherapeutic treatments, and exercises. Classic physiotherapy, neurophysiological treatments such as those designed by Bobath or Vojta, manual techniques, and craniosacral therapy or myofascial relaxation techniques should be used as needed.

**· Orthopedic technology**

Support from orthopedic technology can facilitate many everyday activities. To avoid muscle foreshortening and malpositions, positioning splints worn mainly at night may become necessary. Braces that support the spinal column, functional orthotic devices, or walking and standing aids, such as the swivel-Walker or parawalker, may improve patient independence.

**What other neuromuscular diseases are there?**

The best-known neuromuscular diseases are:

- **Infantile cerebral palsy**
- **Spinal muscular atrophy**
- **Duchenne muscular dystrophy**

These terms describe a progressive muscle disease beginning in the first two years of life with a weakness of the pelvic muscles that then spreads to the shoulder girdle muscles and develops into a generalized muscular weakness. The relevant trait is carried on the X chromosome, meaning that only boys are affected. Life expectancy is about 20 years. Patients suffering from this condition die of cardiac or respiratory insufficiency due to affected heart or respiratory muscles with repeated bouts of severe pneumonia.

- **Syringomyelia, syringobulbia**

Syringomyelia is a disease of the spinal cord affecting mainly the cervical and thoracic spine, while syringobulbia affects the extended spinal cord (medulla oblongata). Cavities (syringes) containing no nerve cells form in the spinal cord, resulting in dysfunctions of varying severity (disturbance of depth perception, sense of position, uncertain gait, muscular atrophy with altered body statics and the development of scoliosis). The causes leading to formation of these cavities may be both congenital (Arnold Chiari malformation, spina bifida and other primary diseases) and acquired (tumors or infections of the spinal cord).

- **Poliomyelitis**

Polio is caused by RNA viruses from the group of the picorna viruses.

After it attacks the central nervous system, poliomyelitis damages the second motor neurons in the spinal cord, causing paralyses and meningitis.

No antiviral therapy has been developed as yet. The best protection is preventive vaccination.

- **Spinocerebellar ataxia (Friedreich's ataxia)**

Friedreich's ataxia is an inherited spinal locomotor system disorder, characterized by poor locomotor coordination with uncertain stance and gait (stance and gait ataxia). The disease is progressive. A possible cause is iron oversaturation of the mitochondria, resulting in the formation of free radicals, which then damage the nerve cells.

In addition to other complications, such as cardiac muscle enlargement (cardiomyopathy), this ataxia may also cause the development of scoliosis.

- **Arthrogyposis multiplex congenita (AMC)**

This is a congenital disorder of the connective tissues and nervous system resulting in the disruption of the development of muscles (muscle weakness, missing muscle groups) and therefore in poorly developed joints with rigidification. The disease may be associated with malpositions of the spinal column and severe anomalies in the central nervous system.