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COMMON SPINA BIFIDA DEFECT APPEARANCE AND CLINICAL SIGNIFICANCE

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Annotation: This article discusses spina bifida, one of the neural tube defects. Special attention is paid to the etiology, pathogenesis, clinical manifestations of the disease, diagnostic methods and modern treatment and prevention measures. Rehabilitation approaches aimed at improving the quality of life of children born with spina bifida are also highlighted.

Keywords: Spina bifida, neural tube defect, congenital anomaly, neurosonography, prenatal diagnosis, surgical treatment, rehabilitation, folic acid prevention.

Introduction:The aim of the study is to help children diagnosed with spina bifida make the decision that their guardians, parents and relatives will be fully aware of this disease.In addition, the diagnosis of spina bifida includes medical care, treatment, and practical advice for children with infected children. O Spina bifida: Spina bifida, also known as neural tube defect, is a birth defect that occurs when the spine and spinal cord are not formed properly. In the embryonic period, it is considered a defect in the development of the neural tube. In normal cases, the neural tube forms in the early stages of pregnancy and closes on the 28th day after conception. In infants with spina bifida, part of the neural tube does not close or develop properly, leading to defects in the spine and spinal bones. Spina bifida can be mild or severe, depending on the type of defect, location, size, and complications. If necessary, early treatment of spina bifida includes surgery, but the problem is not always solved by such treatment. The defect can occur along the entire length of the spine and manifest itself by the fact that part of the spinal cord and surrounding tissues protrude outward rather than inward. Spina bifida (SB) is the incomplete closure of the posterior parts of the vertebrae due to developmental disorders and the clinical picture that arises in connection with this condition. This is the second by the most common cause of childhood disability after cerebral palsy. In the classical classification, SB is divided into two types: closed (spina bifida occulta, SBO) and open (spina bifida aperta, SBA). With SBO, some anomalies are observed, such as spinal defects, tufts of hair on the skin, and nevi, but the sac is not observed. In contrast, in SBA, there is a sac that includes only the meninges or nerve tissues with meninges; these are called meningocele and myelomeningocele, respectively. Etiopathogenesis The neural plate, which develops between the 2nd and 6th weeks of the embryonic period, closes on both sides and forms a neural tube. While the brain develops from the cranial part of the neural tube, the spinal cord develops from the caudal part. Problems that occur when closing the neural tube, Statistics: Spina bifida occurs between 1 and 2,875 births in the United States each year. The caudal part of the neural tube at this stage causes SB and other spinal cord disorders.The frequency of SAT varies from 0.2 to 10 per 1,000 people in different regions of the world.; However, in the United States, it drops to 0.2 per 1,000 people. In a study conducted in Turkey between 2003 and 2004, 17 cases of SBO and SBA were identified out of 8,631 live births. Based on these data, it can be assumed that the incidence is 1.97 per 1,000 people. Genetic factors play an important role in the etiology of SB. The incidence of SB is often 50 times higher than in the general population if the patient has siblings with SB. Genetic

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predisposition has also been identified in studies of twins and families. Since it is believed that the genes involved in folic acid metabolism are responsible for this, research is focused on these genes.However, environmental factors are also important for the development of the SB. These include taking medications such as carbamazepine, which affect folic acid metabolism during pregnancy, poorly controlled diabetes, and inflammatory diseases suffered in the first trimester of pregnancy. Many studies have shown that taking folic acid before and during pregnancy reduces the frequency of About 85 percent of the defects are in the lower back, and 15 percent are in the neck and chest. The integrity of the spine can be surgically restored, but nerve damage cannot be repaired, and if they are obvious, patients may have paresis of the lower extremities of varying severity. The higher the spinal cord malformation, the more serious the nerve damage and motor disorders (paresis and paralysis). According to research, this developmental anomaly occurs in about 7 cases per 10,000 newborns [1].

TYPES AND SYMPTOMS OF SPINA BIFIDA: * Latent spina bifida occulta is the most moderate form without obvious signs of malformations and skin changes.With this form, at least one vertebra changes, but there may also be no changes in the nerves and spinal cord. At birth, the baby may have a spot or a cavity in the area of the anomaly. And, as a rule, the child has no symptoms. In this form of the anomaly (as in others), there may be an anomaly in the development of the spinal cord, characterized by the fact that the spinal cord attaches to the spine to the end of the lumbar region, usually the spine ends at the level of the first lumbar vertebrae and hangs freely without attaching to the spine. * Meningocele (meningocele) is an anomaly of moderate severity (and the most common) in which the spinal canal does not close properly, and the soft meninges (membranes lining the spinal cord) extend beyond the bone structures of the spinal canal, but the spinal cord itself remains intact. The cystic mass is covered with skin. Most children with meningocele continue to have normal limb function, but may have partial paresis or bladder or bowel dysfunction. With this anomaly, underdevelopment of the spinal cord is often observed. Almost all patients with this anomaly require surgical treatment to close the defect and release the spinal cord [2].

*Myelomeningocele (myelomeningocele): the most severe common form associated with the concept of spina bifida. The spinal canal is not closed, and the swollen mass consists of soft meninges, pathologically altered spinal cord and nerves. In addition, the skin in this area is also underdeveloped. Children with this form of spina bifida have paresis in whole or in part under the defect and pelvic organ dysfunction. In addition, nerve damage and other pathologies are observed. Symptoms The symptoms of spina bifida vary significantly depending on the child's build and weight [3].

For example, at birth:

* There may be no obvious signs or symptoms of latent spina bifida occulta (spina bifida occulta)-just a small spot, animal fur, or a mole [spot].
* Meningocele (meningocele) has a sac-like protrusion located at the back of the spine.
* With myelomeningocele (myelomeningocele), protrusion also occurs, but with skin changes, the nerves and spinal cord separate.

Severe form of spina bifida with localization may have the following symptoms: paralysis of the lower extremities, dysfunction of the bladder and intestines.

In addition, such patients may usually have other developmental abnormalities:

* Hydrocephalus occurs in 75% of cases of myelomeningocele, and this condition requires surgical endoscopic treatment to restore normal outflow of cerebral fluid or the use of a shunt to remove excess fluid from the brain. Chiari's anomaly (displacement of the brain to the upper

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cervical spine) can exert pressure on the brain stem, which can manifest as motor disorders of speech, swallowing, and extremities.
* Underdevelopment of the spine, orthopedic problems including scoliosis, kyphosis, hip dysplasia (congenital dislocation), additional deformities of the foot, etc.
* Early sexual development (especially in girls with spina bifida and hydrocephalus).
* Depression and other neurotic conditions, obesity, dermatological problems abnormalities in the development of the urinary tract [4].
Heart disease vision problems Diagnostics During pregnancy, diagnostic measures can be taken to assess the presence of spina bifida in the embryo. These include: Amniocentesis (amniocentesis): A procedure in which a long, thin needle is inserted into the amniotic sac through the mother's abdominal cavity to extract a small amount of amniotic fluid for ultrasound examination. The fluid is analyzed to determine if there is an open neural tube defect. Although the analysis is very reliable, it does not reveal minor or closed defects. Diagnosis and treatment at the stage of intrauterine development SB can be detected at the stage of intrauterine development. A high level of alpha-fetoprotein indicates SB (1). With the help of ultrasound examination (ultrasound) performed at this stage, it is possible to diagnose SB; It is possible to accurately determine the lesion level, especially with the help of three-dimensional ultrasound (11,12). In a study conducted in In Turkey, it was noted that the diagnosis is established at the prenatal stage in 72% of cases (13). Magnetic resonance imaging (MRI) can provide accurate data in situations where it is impossible to obtain an adequate ultrasound image, for example, with maternal obesity, lack of water, fetal position, or advanced bone structure in an older fetus (14). At the prenatal stage, it is possible to assess the possible clinical course of the disease by determining the lesion level and concomitant malformations. Despite the fact that in some medical centers surgical interventions are performed for infants with Down syndrome in the prenatal stage, it is reported that these interventions pose a risk to mothers and infants, and full recovery is not achieved. Therefore, such operations should be performed in medical centers where there are multidisciplinary teams consisting of specialists in this field (15,16). Although this is a controversial issue, the family should be advised to seek advice on termination of pregnancy, especially in severe cases (11,14). There is no clear evidence that children who have been diagnosed with Down syndrome during pregnancy should be born naturally or by caesarean section. One study reported that children born by caesarean section have a two-segment lower neurological level; in clinical practice, cesarean section is often preferred. At the same time, there are studies that report that the method of delivery does not affect the neurological level (17-20). Closure of the lesions during the first 72 hours after delivery reduces the risk of infection of the central nervous system minimizes the likelihood of neurological disorders. In some patients with Hydrocephalus is present at the same time, and early intervention may be required. Therefore, it is recommended that children with SAT receive this method, which is absolutely harmless and allows noninvasively assessing the condition and visualizing internal organs, blood vessels, and fetal tissues. Sometimes you can find not only a spina bifida, but also other anomalies. in the center, where there are appropriate specialists, and the necessary interventions can be carried out as soon as possible in a planned manner (18, 20, 21). Clinical data and evaluation.Signs and Symptoms of spina bifida In general, infants with spina bifida in the form of a cyst have the following symptoms: Lethargy Poor nutrition Irritability Stridor Impaired coordination of oculomotor movements Delayed development Older children may be presented with the following: Cognitive or behavioral changes Reduced strength Increased spasticity Changes in bowel or bladder function Lower cranial nerve dysfunction Back pain Worsening of

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orthopedic deformities of the spine or lower extremities Charles Bonnet syndrome usually shows secondary signs of motor neuron damage, such as decreased deep tendon reflexes, muscle weakness, and hypoesthesia/anesthesia. This clinical picture may combine diseases of the spine, such as kyphosis and scoliosis, dislocation of the hip, contractures of the hip and knee joints, as well as deformities of the ankle and foot. The physical examination of a patient with Charles Bonnet syndrome begins with an examination. The presence of an open, operated, or skin-covered pouch on the back, hirsutism, and skin lesions such as discoloration; spinal abnormalities, misalignment of joints, and deformities, if any, are assessed. A child with Beckwith-Wiedemann syndrome has been monitored for some time and the nature of spontaneous movements of the lower extremities is studied. It has been reported that the frequency of spontaneous movements of the lower extremities in children with Beckwith-Wiedemann syndrome is lower than in healthy children. Spinous processes are examined by descending palpation; defects or abnormal formations are detected. The assessment ends with an examination of the neurological and musculoskeletal systems, which are carried out taking into account the patient's age [5].

During the examination, it is necessary to assess the strength of the muscles of the lower extremities, which is very important for determining the level of gait, using a manual blood test.; it is necessary to conduct a comprehensive sensory examination; and based on the results obtained, determine the neurological level of the lesion. Despite the fact that there are various classification systems for detecting motor disorders in SAT, the criteria of the International Research Group on Myelodysplasia best demonstrate the motor level. In these criteria, each motor level for lesions is T10 and below It is determined in detail (for example, for L2, the strength of the muscles of the ilio-lumbar muscle, the sartorial muscle and all adductor muscles of the thigh should be 3 or higher). Thus, the use of a common language for assessment and a clearer demonstration of the clinical condition during subsequent examinations became possible. Bifida's back and rehabilitation Patients with Budd-Chiari syndrome have clinical manifestations that vary depending on the lesion of motor neurons. While in mild cases with sacral segment lesions, only bladder and intestinal dysfunction and/or denervation of the internal muscles of the foot are observed, with lumbar segments in the lower extremities, varying degrees of muscle weakness are observed. If the L2–L4 roots are strong, then the strength of the knee extensor muscles, which play an important role in functional walking, is good. The flexors and adductor muscles of the thigh are innervated by the roots L1–L2. If these roots are protected, the patient can flex the hip. On the contrary, when the thoracic region is affected, all the roots innervating the muscles of the lower extremities are affected, and the legs are completely paralyzed [1, 27]. Spinal deformities and contractures of the lower extremities also negatively affect functional mobility. Therefore, these disorders should be detected during a physical examination, and then treated and monitored. In cases where an assessment cannot be performed using a manual muscle test, the affected muscles can be examined using electromyography (EMG). In a study involving 50 children with Burt-Johnson syndrome and 50 children with Landau-Kleffner syndrome, EMG showed that the most commonly affected muscle in both groups was the anterior tibialis muscle. In the same study, while children with SBO had no clinical manifestations or mild to moderate neurological deficits , children with SBA had severe neurological damage. with many affected roots . Imaging techniques should be used to determine the type and prevalence of lesions in patients with SB. Although conventional radiography can only show bone abnormalities, more detailed information about the lesions can be obtained using MRI. In suspicious cases, when only skin manifestations are observed, the diagnosis is SBO can be delivered using an MRI scan.

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Blood tests: Between 15 and 20 weeks of pregnancy, blood tests are recommended for all women who have previously had a neural tube defect and who have no family history of having a child with this disease. A blood test for alpha-fetoprotein and other biochemical parameters allows you to determine how high the risk of developing spinal abnormalities is. At birth, severe cases of spina bifida are manifested by the appearance of a fluid-filled pouch on the newborn's back. Visual indicators of non-severe forms (spina bifida occulta) may include a hairy spot on the skin or a cavity along the spine. Unusual weakness or lack of coordination of movements in the lower extremities also indicate the presence of a spina bifida. In children and adults, this anomaly is often diagnosed during regular checkups or, if necessary, neurological symptoms are differentiated using instrumental research methods (MRI, CT, radiography). Reasons During pregnancy, the human brain and spine begin to form as a flat plate of cells enclosed in a tube called the neural tube. If all or part of the neural tube does not close, it is called an open neural tube defect. An open neural tube is open in 80 percent of cases and covered with bone or skin in 20 percent of cases.

The cause of spina bifida (spina bifida and other defects) remains unknown, but it is caused by a combination of genetic, nutritional, and environmental factors such as:

*Lack of folic acid (vitamin B) in the mother's diet during pregnancy (consuming sufficient amounts of folic acid during pregnancy may reduce the risk of developing this disorder).

*Uncontrolled maternal diabetes * Some medications (antibiotics, anticonvulsants).

*The genetic factor is usually important only in 10% of cases.

Mother's age, How to be born according to the bill (first-born children are at greater risk). Socio-economic status (children born into low-income socio-economic families are at high risk). Ethnicity obesity or excessive alcohol consumption by a pregnant woman. In case of hyperthermia of a pregnant woman in the early stages (sauna, Jacuzzi).

Treatment :The prenatal period Women of childbearing age can reduce the risk of having a child with spina bifida by taking 400 micrograms (mcg) of folic acid every day before conception. Since folic acid is soluble in water, it does not stay in the body for long, and it must be taken every day to protect against birth defects of the nervous system. Since half of all pregnancies in the United States are unplanned, folic acid must be taken regardless of whether a woman is planning a pregnancy or not. Studies have shown that if all women of childbearing age took multivitamins with vitamin B folic acid, the risk of neural tube defects could be reduced by up to 70%. Fetal Surgery A randomized Trial compared the elimination of a defect during fetal development with the elimination of a defect after birth and yielded encouraging but mixed results. In patients who had the defect repaired during intrauterine development, the probability of needing ventriculoperitoneal bypass surgery was lower (44% vs. 84%), and the probability that they would be able to walk was higher (44% vs. 24%). In the group where the defect was eliminated during intrauterine development, there were also much more complications in the mother and child. Given the higher risk of complications, as well as the lack of long-term follow-up, the American College of Obstetricians and Gynecologists (ACOG) currently recommends performing fetal surgery only in specialized centers with experience in performing such operations [6].

Treatment of spina bifida can begin immediately after the birth of a child. If this defect is diagnosed prenatally, a cesarean section is recommended to reduce spinal cord damage when the fetus passes through the birth canal. Newborns with meningocele or myelomenigocele are recommended to undergo surgical treatment within 24 hours after birth. With such an operation, the bone defect is closed, and the function of the intact part of the spinal cord can be preserved.

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Unfortunately, surgical treatment cannot irreversibly restore the functions of damaged nerves. Currently, there are clinics that perform prenatal surgery to close the defect, but the methods have not yet become widespread. The main objective of treatment, both in uncomplicated form and in the postoperative period, is to preserve the functions of the musculoskeletal system, as well as the functions of the bladder and intestines. If necessary, orthopedics, therapeutic gymnastics, and physiotherapy are used. In cases where a spina bifida has been accidentally detected by X-ray examination (MRI, CT), measures should be taken to reduce the risk of spinal cord injury in the part of the spine where this defect exists. There is no cure for spina bifida because the nerve tissue cannot be replaced or repaired. Treatment of the various effects of spina bifida may include surgery, medication, and physical therapy. Most children will need assistive devices such as crutches or wheelchairs. Ongoing therapy, medical care, and/or surgical treatment may be necessary to prevent and treat complications throughout a person's life. Surgery to close the hole in a newborn's spinal cord is usually performed within 24 hours of birth to reduce the risk of infection and preserve existing spinal cord function. Surgical treatment in adults is used only in the presence of complications. Basically, treatment in adults is aimed only at preventing possible complications (physical therapy, physiotherapy, wearing a corset).

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