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A COMPREHENSIVE REVIEW OF SPINA BIFIDA: PATHOPHYSIOLOGY, ETIOLOGY, SYMPTOMS, DIAGNOSIS AND MANAGEMENT

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ABSTRACT

Inadequate closure of the neural tube during embryonic development results in spina bifida, a congenital neural tube defect. It can cause neurological and physical impairments by affecting the spinal cord. Meningocele, spina bifida occulta, and the most severe variant, myelomeningocele, are the primary forms. Its development is influenced by environmental, dietary, and genetic factors, particularly folic acid deficiency. Developmental delay, bowel and bladder problems, and muscle weakness are typical symptoms. Alpha-fetoprotein tests and ultrasonography are used for prenatal diagnosis. Antibiotics, hydrocephalus therapy, foetal surgery, and orthopaedic care to enhance mobility and quality of life are all part of management.

KEYWORDS: Spina bifida, Neural tube defect, Myelomeningocele, Congenital disorder, Hydrocephalus, Folic acid deficiency, Prenatal diagnosis, Orthopaedic management.

1. INTRODUCTION

The most prevalent congenital abnormality of the central nervous system is spina bifida (SB).

Although it is treatable, the condition foretells permanent handicap for everyone who has it.[1] This neurological disorder occurs when one or more vertebrae do not fully form, creating a gap in the spine. This causes lifelong harm to the spinal cord and nervous system.[2] Myelomeningocele (open Spinabifida), the most clinically relevant subtype, is a disease in which the lumbosacral spinal neural tube fails to complete during embryonic development.[3] The second most prevalent congenital abnormalities in humans that impact the development of the central nervous system are neuraltube defects (NTDs).[4]

1.1 TYPES OF SPINABIFIDA

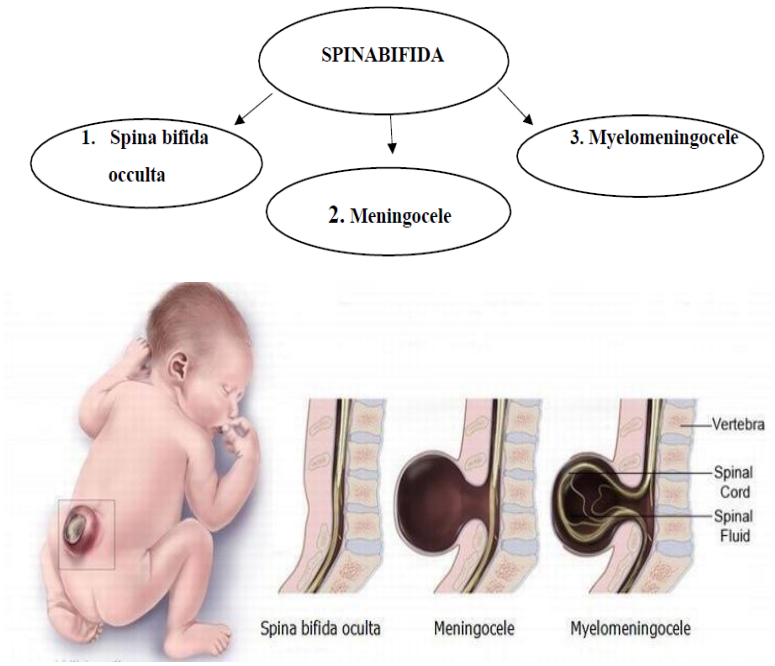

Fig.1: Types of Spina bifida.[19]

1. SpinaBifidaOcculta

Occulta" means "hidden." The most common and least severe type is spina bifida occulta. This type of spina bifida causes one or more of the vertebrae, the bones that make up the spine, to have a little gap or separation. Many people with spina bifida occulta are not even aware that they have the condition. It can be discovered by an imaging test, such as an X-ray, that is carried out for a different reason.[5]

2. Meningocele

This kind of spina bifida is uncommon. This kind causes a bag of spinal fluid to protrude via a spinal aperture. No nerves are affected and the spinal cord isn't in the fluid sac. Babies with meningocele may spinal abnormalities have some minor trouble with functioning, including with the bladder and bowels.[5] Additional are linked to a complicated meningocele.[6]

3. Myelomeningocele

Myelomeningocele is the most serious type. It also is known as open spina bifida. The spinal canal is open along several vertebrae in the lower or middle back. Part of the spinal cord, including the spinal cord's protective covering and spinal nerves, push through this openingatbirth, form ingasacon the baby's back. Tissuesandnervesusuallyareexposed. This makes the baby prone to dangerous infections. This type also may cause loss of movement in the legs, and bladder and bowel dysfunction.[5] Before the 12th postmenstrual week, abnormalities of the bony spine on foetal ultrasonography can identify myelomeningocele.[7] Myelomeningocele(MMC)isacongenitalabnormalityof the central nervous system that cannot be cured.[8]

1.2 ETIOLOGY

Neural tube development defects are believed to be complex, involving both hereditary, multiple genes and environmental factors.[9] It is widely acknowledged that insufficient consumption of folic acid, a synthetic version of folate, or natural folate. A child's risk of developing myelomeningocele may also be increased by maternal exposure to fumonisins, electromagnetic fields, hazardous waste sites, drinking water disinfection byproducts, and pesticides.[10]

1. Hereditary Multiple Genes

The likelihood of these outcomes is greatly increased by a family history of spina bifida. Research from the 1970s and 1980s revealed that family members of people with spina bifida were at an increased risk: 3–8% of siblings (first-degree relatives), 1-2% of second-degree relatives, and 0.5% in third-degree relatives[13–17], as opposed to 0.1% (or 1 per 1,000) in the US. According to these research, there is only one person with spina bifida in 95% of families, a non-linear fall in risk with decreasing degree of link, and a higher risk of NTDs in families with multiple NTDs.[11]

2. Multiple Genes

Genes involved in folate homocysteine metabolism, including MTHFR, MTR, MTRR, and MTHFD1, are commonly attributed. It is unknown if the risk of SB is linked to gene interactions, their presence in the maternal or foetal genome, or their interaction with environmental factors. Gene panels cannot accurately identify the likelihood of foetal SB pre-conception due to its complex etiology.[12]

1.3 PATHOPHYSIOLOGY

Gastrulation and primary and secondary neurulation are stages of normal spinal cord development that occur during the second and sixth weeks of pregnancy. In the initial stage of gastrulation, the endoderm and ectoderm form a bilaminar embryonic disk. Cell division and migration result in the creation of a mesoderm, which then forms a trilaminar disk. A neuroectoderm is created when the notochord and ectoderm connect. The neural plate spreads proximally and caudally from the midline. Spinal dysraphism may arise from pathological effects during first neurulation. The creation of nerve folds, also known as nerve grooves, is part of primary neurulation. By connecting the neuronal folds, then erveplate transforms into a neural tube.[13] Initially, the exposed neuroepithelium will differentiate and develop normally, but the" second hits" of amniotic fluid exposure and mechanical trauma of the exposed cord in utero eventually cause neuronal cell death and loss. This concept is backed by Stiefel et al.'s work in mutant mouse models, as well as clinical reports of rapid neonat al loss of motor function, despite the presence of normal-appearing movement in utero.[1]

1.4 SIGNSAND SYMPTOMS

In general, new borns with spinabifida cystic exhibit the following.[14]

- Lethargy
- Poor feeding
- Irritability
- Stridor
- Ocular-motorincoordination.
- Development Delay

❖ Older children may exhibit the following[14]

- Cognitive or behavioural changes
- Decreased strength
- Increased spasticity.
- Bowelor bladder function changes.
- Lower Cranial Nerve Dysfunction.
- Backache.

1.5 DIAGNOSIS

Prenatal diagnosis of spina bifida is made either by ultrasound at 18–20 weeks of gestation or by measuring alpha foetal protein in maternal serum at 16 weeks. The accuracy of test results ranges from 85 to 90%.4 Prenatal genetic counselling can assist families in choosing appropriate tests, scheduling them, and interpreting the findings.[15]

1. Test for Alpha-Feto Protein(AFP)

2. Ultrasound Findings

1. Test for Alpha-Feto Protein(AFP)

- Maternal blood test
- Amniocentesis

2. Ultrasound Findings

Myelomeningocele can be discovered before the 12th postmenstrual week through anomalies in the bony spine. A bulge on the foetal back's posterior shape. After the 12th postmenstrual week, sonographic foetal indicators for open NTDs include the "banana" sign. Thelemon sign indicates a concave shape of the frontal calvarium. The banana sign indicates the posterior convexity of the cerebellum in spina bifida.[16]

1.6 MANAGEMENT

- Foetal Surgery
- Antibiotics
- Management of Hydrocephalus by Ventricular Shunt
- Orthopaedic Care

- **FoetalSurgery**

Foetal surgery for my elomeningocele can block flow of spinal fluid from the back, andso may prevent or reverse the Chiari II malformation and hydrocephalus.[16]

- **Antibiotics**

To limit the danger of central nervous system infection, prophylaxis with broad-spectrum antibiotics is typically given until the back wound is closed. Early CNS infection can be avoided with proper wound care and precautions.[16]

- **Management of Hydrocephalus by Ventricular Shunt**

Hydrocephalus caused by myelomeningocele is commonly treated with a CSF shunt. The

shunt consists of three parts: aproximal tube in the ventricle, a valve to control the volume of CSF drained, and a distal tube that relocates the CSF to another portion of the body.[17]

❖ **Orthopaedic Care**

Orthopaedic management for patients with spinabifida focuses on preventing or repairing deformities to promote mobility and independence while adhering to realistic functional neuro segmental levels. Treatment for spina bifida depends on the patient's functional neuro segmental level, stable neurological status, and awareness of the condition's natural history.[18]

2. CONCLUSION

A congenital disorder called spina bifida results in abnormal bone formation in the spinal cord and spine. The least severe kind of neural tube defect is closed spinal dysraphism, which has limited neural involvement and a hidden vertebral lesion. Meningocele and myelomeningocele are examples of open spinal dysraphism, a condition in which neural tissues interact with the external environment. Neural tube anomalies vary by country, gender, race, and geography. The etiology and pathophysiology of spina bifida are influenced by a number of maternal, environmental, and dietary variables. However, it's unclear how these elements affect things, thus further study is needed in this field.

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