SPINA BIFIDA – ETIOLOGY, CLINICAL FEATURES AND MODERN MANAGEMENT

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Abstract: Spina bifida is a complex congenital neural tube defect characterized by incomplete closure of the spinal column during early embryonic development. It is associated with significant neurological, orthopedic, and urological complications. This article reviews the etiology, classification, clinical manifestations, diagnostic methods, and modern management of spina bifida.

Keywords: spina bifida, myelomeningocele, neural tube defects, folic acid, hydrocephalus, fetal surgery.

Introduction

Spina bifida occurs in approximately 1–2 per 1,000 live births worldwide and represents one of the most common congenital anomalies of the central nervous system. The condition results from a failure of neural tube closure between the 3rd and 4th weeks of gestation. Improvements in prenatal screening and surgical techniques have significantly increased survival rates and quality of life.

Etiology and Risk Factors

The development of spina bifida is influenced by genetic and environmental factors. The most important modifiable risk factor is maternal folic acid deficiency before and during early pregnancy. Other risk factors include maternal diabetes, obesity, hyperthermia in early pregnancy, and exposure to certain antiepileptic drugs.

Classification

Spina bifida is classified into several types:

- **Spina bifida occulta**
- **Meningocele**
- **Myelomeningocele** (the most severe and common form)

Each type differs in the degree of neural tissue involvement and severity of neurological deficit.

Clinical Manifestations

Clinical features depend on the level and severity of the defect and may include:

- Lower limb weakness or paralysis
- Sensory loss
- Neurogenic bladder and bowel dysfunction
- Orthopedic deformities such as scoliosis and clubfoot
- Hydrocephalus associated with Chiari II malformation

Diagnosis

Prenatal diagnosis is possible through maternal serum alpha-fetoprotein (AFP) screening and fetal ultrasound. Postnatal diagnosis relies on physical examination and neuroimaging, including spinal ultrasound in neonates and MRI.

Management

Treatment requires a multidisciplinary approach:

- Early surgical closure of the spinal defect
- Management of hydrocephalus with ventriculoperitoneal shunting
- Long-term urological and orthopedic care
- Rehabilitation and psychosocial support

Fetal surgery has emerged as a promising option for selected cases, reducing the severity of neurological deficits.

Prognosis and Quality of Life

With modern medical care, many individuals with spina bifida survive into adulthood and achieve good quality of life. However, lifelong medical follow-up is essential.

References:

1. Copp AJ, Greene NDE. Neural tube defects—recent advances. Nature Reviews Neuroscience. 2010;11(10):713–725.
2. Mitchell LE, Adzick NS, Melchionne J, et al. Spina bifida. Lancet. 2004;364(9448):1885–1895.
3. Adzick NS. Fetal surgery for spina bifida. New England Journal of Medicine. 2011;364(11):993–1004.
4. Bowman RM, McLone DG, Grant JA, et al. Spina bifida outcome studies. Journal of Neurosurgery Pediatrics. 2001;95(3):475–480.
5. Dias MS, Partington M. Embryology of myelomeningocele. Neurosurgical Focus. 2004;16(2):E1.