

## SPINA BIFIDA

### Definition and prevalence

Spina bifida is an inborn anomaly of the spinal column, which consists of incomplete closure of the posterior wall of the spinal column, most frequently in the lumbo-sacral region. However, it can occur anywhere along the spinal axis. As a result the dura, and/or the arachnoid and/or the spinal cord may protrude out of the bony defect of the vertebral arch. If the defect is open it is called spina bifida aperta, and if it is covered by skin the term is spina bifida occulta. This is one of the most common birth defects with an average prevalence worldwide of 1-2 cases per 1000 births.

### Etiology

A multifactorial inheritance has been proposed to be responsible, coupled with environmental factors, of which nutrition and folic acid are most important. Cytoplasmic factors, polygenic inheritance, chromosomal aberrations, and environmental influences (eg, teratogens) have been considered as possible causes. Fetus with an open neural tube defect has an elevation of alpha-fetoprotein in the amniotic fluid. Ultrasound confirmation with amniocentesis generally is possible at 15-18 weeks to rule out a spina bifida.

### Prevention

There is neither a single cause of spina bifida nor any known way to prevent it entirely. However, dietary supplementation with folic acid has been shown to be helpful in reducing the incidence of spina bifida. Sources of folic acid include whole grains, fortified breakfast cereals, dried beans, leaf vegetables and fruits. Folate fortification of enriched grain products has been mandatory in the United States since 1998. The US Food and Drug Administration, Public Health Agency of Canada and UK recommended amount of folic acid for women of childbearing age and women planning to become pregnant is at least 0.4 mg/day of folic acid from at least three months before conception, and continued for the first 12 weeks of pregnancy.

### Clinical picture

Patients with spina bifida have variable neurologic deficits. Examination of neurologic deficit helps determine the functional level at which the spina bifida cystica lesion has interrupted function. Patients are categorized into general groups according to the level of deficit. Generally, the neurological level is grouped as thoracic, upper lumbar (L1, L2, L3), lower lumbar (L4, L5), or sacral. The neurologic deficits characteristically cause deformity by muscle imbalance forces. Unopposed muscle pull can cause spinal deformity, progressive lower extremity contractures, hip dislocations, and, less commonly, dislocations in other joints (foot and ankle). 22% of patients have a significant fracture in their lifetime and may have a fracture associated with a significant surgical procedure, such as reduction of the hip or spine surgery. Osteoporosis is a frequent finding, complicating their already present osteopenia. Ulcers from bracing are prominent in the lower extremities in the pelvis and, particularly, over the bony prominences as a result of sitting. Infection is common, particularly with a neurogenic bladder.

### Treatment

A child born with myelomeningocele is transferred to a center, where neonatal surgery and closure can be performed. Surgery involves freeing lateral muscles and skin for coverage and attempting to form a closure of the neural elements with minimal scarring as late complication of a tethered cord has frequent and severe consequences. Further follow-up is carried out by a multidisciplinary team. Neonatal neurosurgery is followed by serial examination of muscle strength and joint range of motion, orthopedic evaluation to detect any early changes that may require intervention. Patients are followed-up for appropriate development and provided with prolonged physical therapy and adaptive training while in school. Neurosurgical follow-up is required to recognise the complications of hydrocephalus or a possible tethered cord and to monitor any potential causes of seizure activity.

Urologic evaluation is necessary to establish a bladder regimen to prevent frequent urologic infections and to recognize and treat early potential hydronephrosis or other causes of renal damage that can limit life expectancy. Pediatric evaluation is appropriate for any child and, specifically, should include efforts to maintain a reasonable weight, as children without ambulation tend to gain excessive weight and develop associated morbidity. Endocrinologically, a growth hormone deficiency may be present, which could cause patients to be about a foot shorter than their peers.

### Rehabilitation and follow-up care

Rehabilitation in children with spina bifida varies according to age and degree of sensory-motor impairment due to malformations. If spinal cord is not directly affected (as in the case of spina bifida occulta), there is normal motor development of the patient. However, there is a tendency these patients to develop chronic lower back pain. Rehabilitation for spina bifida includes physical therapy, occupational therapy, and recreational therapy. Speech therapy may be indicated for patients with speech and/or swallowing difficulties.

In managing the cases of newborns with myelomeningocele, the physical therapist establishes a baseline of muscle function. As the child develops, the physical therapist monitors joint alignment, muscle imbalances, contractures, posture, and signs of progressive neurologic dysfunction. The physical therapist also provides caregivers with instruction in handling and positioning techniques and recommends orthotic positioning devices to prevent soft tissue contractures. Near the end of the first year of life, it is recommended to provide the child with an effective means of independent mobility in conjunction with therapeutic exercises that promote trunk control and balance. Children with spina bifida often have impairment in fine motor skills and conducting activities of daily living. Early training is expected to compensate for these deficits and it should progress along the developmental sequence as closely as possible. For the school-aged child, recreational therapy provides opportunities for participation in adapted sports and exercise programs, which can result in long-term interest in personal fitness and health.

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