CHAPTER **25**

SPINA BIFIDA

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Spina bifida is a neural tube defect caused by failure of the fetus' spine to close during the first month of pregnancy. The neural tube begins to fuse on about day 27 and completely closes on day 28 of gestation.[1] The primordium of the vertebrae forms from mesoderm, which separates from the neural tube.[1] Failure of closure of the neural tube and associated primitive mesodermal and ectodermal elements account for the appearance of congenital midline defects known as dysraphism, the most common of which is spina bifida.[1]

There are three general types of spina bifida, with increasing degrees of severity: spina bifida occulta, meningocele, and myelomeningocele. Spina bifida occulta refers to an opening in one or more vertebrae of the spinal column without any apparent damage to the spinal cord. Meningocele occurs when the meninges herniates through the opening in the vertebrae into a sac called the "meningocele." The spinal cord remains intact, thus allowing for repair with little or no damage to the nerve pathways. The most severe form, myelomeningocele, describes a portion of the spinal cord protruding through the opening in the spine. In some cases, the sac is covered with skin; in others, tissues and nerves may be exposed.

RISK FACTORS

Non-genetic risk factors of spina bifida include maternal diabetes mellitus, obesity, heat exposure, and febrile illness in early pregnancy. Other factors include geographic location, ethnicity, teratogen exposure, folic acid deficiency, and exposure to toxins or anticonvulsant drugs.[2] Maternal use of aminopterin, salicylates, insulin, clomiphene, diuretics, antihistamines, or sulfas increases risk of spina bifida.[3] An increase in prevalence is noted with exposure to valproate and/or carbamazepine.[4,5] From 1991-1996, approximately 50% of cases of spina bifida were related to folic acid deficiency.[6] The US Centers for Disease Control and Prevention and the US Public Health Service Advisory in 1992 have recommended the administration of folic acid at a dosage of 0.4 mg/day for all women anticipating pregnancy;[1,6] however, it is estimated that one half of all pregnancies are unplanned.[6] Furthermore, it is estimated that 80% of women of reproductive age have no folic acid in their normal diet.[6] The dose of folic acid should not exceed 1.0 mg/day to avoid the risk of secondary vitamin B12 deficiency.[6]

Genetic causes of spina bifida are rare, as only 5% of neural tube defects occur in patients with a positive family history, even though chances of having a child with a neural tube defect are increased if genetic predisposition exists. If a woman delivers a child with spina bifida, the probability of another affected pregnancy is about 3-4%. Overall, incidence of spina bifida is approximately 1 per 1000 live births.[6,7]

DIAGNOSIS

The easiest and most common form of detection is via ultrasound. With an ultrasound, features such as an open spine or malformations at the head region are easily seen.[8] Also, elevated serum levels of alpha-fetoprotein (AFP) are noted in approximately 75-80% of women having fetuses with spina bifida.[8] With an open neural tube, the exposed fetal membranes and blood vessel surfaces increase the AFP levels in both maternal serum and amniotic fluid.[1] If results are inconclusive, measurement of AFP and acetyl-cholinesterase by amniocentesis is warranted.[1] Measurement of amniotic acetylcholinesterase activity may help detect an open neural tube in utero because AFP levels may be elevated in other conditions, such as gastroschisis, omphalocele, and nephrosis.[1] Increased levels of amniotic AFP and acetylcholinesterase detect at least 90% of spina bifida fetuses, whereas maternal serum AFP levels detect 60-80%.[1]

CLINICAL PRESENTATION

Spina bifida deformities are often seen with other nervous system abnormalities, such as tethering of the spinal cord, diastematomyelia, hydromyelia, and hydrocephalus usually associated with Arnold-Chiari

type II malformation.[1] Other defects of ectodermal and mesodermal origin may be seen, such as pelvic meningoceles, hamartomas, lipomas, and dermoid tumors.[1] Visible skin markers may be seen, including skin tags, hair tufts, abnormal dimpling, or aplasia cutis congenita.[1] Most patients with spina bifida have impairment of bowel and bladder function, making them more susceptible to urinary tract infections and stones. Patients may also experience reduced sexual function due to low genital sensation. Of the children born with spina bifida 80% have normal IQs, and 60% have learning disabilities including difficulties paying attention, problems with expression and comprehension of language, difficulties with reading, and demonstration of basic mathematic skills.[2] Another common problem associated with spina bifida is the development of a latex allergy, presumably due to numerous operations.[9]

The neurosegmental level of the lesion is the most important factor determining foot deformity in spina bifida patients.[10] In those patients with thoracic lesions, there is no motor activity below the knee.[10] When evaluating foot deformity in these patients, it is imperative to exclude possibly progressive neurological disorders such as a tethered cord, hydromyelia, or spasticity secondary to brain damage.[10] The most common foot deformity in these patients is equinus (55%).[10] Patients with mid-lumbar lesions predominantly have clubfoot deformities (87%), with the peroneal muscles being weak or inactive.[10] The anterior and posterior tibial muscles are unopposed in acting as deforming forces, causing a valgus, equinus, and adduction deformity of the foot.[10] A calcaneal deformity is most commonly seen in lower lumbar and sacral lesions (34%); however, a clubfoot deformity may develop in cases with muscle imbalance secondary to weak peroneal muscles.[10] In the majority of sacral lesions, no foot deformity is present at birth, but can develop as a result of weight bearing.[10] Digital contractures and hallux abductovalgus deformities are likely to develop during childhood or adolescence due to weakness of intrinsic muscles of the foot.[10]

TREATMENT

Prior to initiation of treatment of lower extremity deformities in spina bifida patients, it is necessary to accurately analyze several factors. First, a detailed sensory examination should be performed in each extremity, including proprioception, protective, and tactile sensation.[11] Motor function must also be assessed, including a determination of any degree of spasticity present. Review of any prior physical exams would prove beneficial to determine stability of neurologic function.[11] Particular note should be made of the gait cycle. Active and passive range of motion of the hip, knee, and ankle should be assessed.[11] The motion needed at the foot and ankle needs to compensate for any contractures elsewhere in the limb.[11] It is also important to consider the patient's mental capacity, psychological state, and any increased potential for non-compliance.

Treatment goals are individualized to the functional status of each patient. Patients with thoracic lesions have minimal potential for independent ambulation; thus, the primary goal in these patients is to correct deformities to allow for plantigrade foot positioning.[11] This facilitates fitting into shoegear, reduction of pressure ulcerations, and ease of wheelchair positioning. In patients with upper lumbar level lesions, active hip flexion and quadriceps power may allow some ambulatory potential for several years.[11] Treatment for these individuals should be aimed at maintaining the feet free of contracture and deformity, allowing for appropriate orthosis wear and prevention of pressure ulcerations.[11] Lower lumbar level lesions typically signify good long term ambulatory potential.[11] This may be limited by medial hamstring function and possible unopposed ankle joint dorsiflexion.[11] In these patients, restoration of muscle imbalance and correction of deformities are the goals, once again to minimize the potential of pressure ulcerations.[11]

Patients with sacral level lesions maintain some ankle joint plantarflexion, yet intrinsic paralysis may lead to foot deformities involving both forefoot and rearfoot malalignment.[11] These patients have the best long term prognosis for sustained independent ambulation, so treatment goals are wide-ranging and include stability of the limb during ambulation at the knee, hip, ankle, and foot, maintenance of a plantigrade foot with proper muscle balancing, restoration of joint motion where feasible to allow for maximal functional capacity of the foot, and avoidance of high pressure areas or bony prominences on the weight bearing surfaces of the foot.[11] Whereas stability may be obtained in some patients with ankle foot orthoses,[12] others may require more aggressive and definitive surgical treatment, including soft tissue releases designed to restore muscle balancing, and/or triple arthrodesis.[13-15]

CONCLUSION

Spina bifida is one cause of complex foot and ankle deformity with widely variable degrees of severity. Thus, the podiatric physician must evaluate the patient for precise sensory and motor function in the lower extremity prior to implementation of any treatment plan. It is imperative to assess the presence of any progressive neurological conditions as well as the presence of any spasticity. Additionally, careful consideration must be given to individually patient goals and needs. It is important to account for the patient's psychological and emotional state to enhance any likelihood of a successful outcome of any treatment plan. Spina bifida is a challenging disease to effectively treat in many aspects, ranging from the complexity of the physical examination to the ultimate determination of treatment goals individualized to each patient.

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