BAHAN AJAR IV
SPINA BIFIDA

Nama Mata Kuliah/Bobot SKS : Sistem Neuropsikiatri / 8 SKS
Standar Kompetensi : area kompetensi 5: landasan ilmiah kedokteran
Kompetensi Dasar : menerapkan ilmu kedokteran klinik pada sistem neuropsikiatri
Indikator : menegakkan diagnosis dan melakukan penatalaksanaan awal sebelum dirujuk sebagai kasus emergensi
Level Kompetensi : 2
Alokasi Waktu : 1 x 50 menit

1. Tujuan Instruksional Umum (TIU) :
Mampu mengenali dan mendiagnosis penyakit-penyakit genetik dan kongenital serta melakukan penangan sesuai dengan tingkat kompetensi yang ditentukan, dan melakukan rujukan bila perlu.

2. Tujuan Instruksional Khusus (TIK) :
   a. Mampu menyebutkan patogenesis terjadinya spina bifida
   b. Mampu melakukan promosi kesehatan dan pencegahan spina bifida
   c. Mampu melakukan penapisan / diagnosis penyakit spina bifida

Isi Materi;

Introduction

Spina bifida ("split spine") is a birth defect where there is incomplete closing of the backbone and membranes around the spinal
cord. There are three main types: spina bifida occulta, meningocele, and myelomeningocele. The most common location is the lower back, but in rare cases it may be the middle back or neck. Spina bifida is a disease categorized as dysraphism, or rachischisis, which is disorders of fusion of dorsal midlins structures of the primitive neural tube, a process that takes place during the first 3 weeks of postconceptual life. Other diseases in the same category is meningocele and encephaloceles. Exogenous factors are presumed to be operative in most cases but there are genetic forms. The most extreme form is anencephaly; it is characterized by the absence of the entire cranium at birth, and the undeveloped brain lies in the base of the skull, a small vascular mass without recognizable nervous structures.

Abnormalities of closure of the vertebral arches frequently found. These take the form of a spina bifida occulta, meningocele, and meningo(myelo)cele of the lumbosacral or other regions. In spina bifida occulta, the cord remains inside the canal and there is no external sac, although a subcutaneous lipoma or a dimple or wisp of hair on the overlying skin may mark the site of the lesion. In meningocele, there is a protrusion of only the dura and arachnoid through the defect in the vertebral laminae, forming a cystic swelling usually in the lumbosacral region; the cord remains in the canal, however. In meningomyelocele, which is 10 times as frequent as meningocele, the cord (more often the cauda equina) is extruded also and is closely applied to the fundus of the cystic swelling. An eventration of brain tissue and its coverings through an unfused midline defect in the skull is called an encephalocele.

The incidence of spinal dysraphism (myeloschisis), like that of anencephaly, varies widely from one locale to another, and the disorder is more likely to occur in a second child if one child has already been affected (the incidence then rises from 1 per 1,000 to 40 to 50 per 1,000).
Etiology
Exogenous factors were many times implicated in an increased rate of both myeloschisis and anencephaly, but the effects of starvation and vitamin deficiency could never be separated from the potential effect of a toxic factor. Inadequate intake of folate in early pregnancy is associated with an increased risk of these malformations. Folic acid, given before the 28th day of pregnancy is protective; vitamin A may also have slight protective benefit. Similar associations have been found with less certainty with exposure during pregnancy to certain antiepileptic drugs, particularly valproic acid and carbamazepine. Maternal diabetes and possibly obesity have been risk factors in some epidemiologic studies. The greatest risk, however, almost 30-fold higher, attaches to a previous pregnancy affected with spina bifida in particular.

**Diagnosis**

Presence of alpha-fetoprotein in the amniotic fluid (sampled at 15 to 16 weeks of pregnancy) and the deformity confirmed by ultrasonography in utero. Blood contamination is a source of error in the fetoprotein test (Milunsky). Acetylcholinesterase immunoassay, done on amniotic fluid, is another reliable means of confirming the presence of neural tube defects. Alterations to cerebral nervous tissue were histologically and cytologically characterised by immaturity and block of normal development during the early stages. For this reason the alteration of primary neuro-ectodermal tissue appears to be the initial factor with secondary involvement of anatomically and functionally contiguous fields and tissues, such as cranial bones and the eyes. The alteration of the nervous system resulted in the incapacity of the fetus to make fetal movements, making it vulnerable to extrinsic compression, above all uterine pressure. This led to the adoption of fixed postures with articular blockage most evident in the lower limbs (congenital clubfoot).
There is severe dysfunction of the cauda equina roots or conus medullaris contained in the sac. Stroking of the sac may elicit involuntary movements of the legs. As a rule the legs are motionless, urine dribbles, keeping the patient constantly wet, there is no response to pinprick over the lumbosacral dermatomal zones, and the tendon reflexes are absent. In contrast, craniocervical structures are normal. Differences are noted in the neurologic picture depending on the level of the lesion. If the lesion is entirely sacral, bladder and bowel sphincters are affected but legs escape; if lower lumbar and sacral, the buttocks, legs, and feet are more impaired than hip flexors and quadriceps; if upper lumbar, the feet and legs are sometimes spared and ankle reflexes retained, and there may be Babinski signs. The two common complications of these severe spinal defects are meningitis and progressive hydrocephalus from a Chiari malformation.

Treatment

Treatment for spina bifida is determined by the severity of the condition. Some conditions do not require any treatment at all. Postnatal surgery is the treatment for meningocele and myelomeningocele, usually performed 24 to 48 hours after delivery of the baby. Surgery so early can help reduce the risk of infection that could occur when the spinal nerves are exposed. Also, this helps minimize additional damage to the spinal cord. During this postnatal procedure, a neurosurgeon places the spinal cord back into the baby’s body and covers them with muscle and skin. During a prenatal surgery, which is done before the 26th week of pregnancy, surgeons expose the mother’s uterus, open the uterus, and repair the neural defect. Prenatal surgery may repair spina bifida defects while still pregnant, reducing further complications after birth. Although the after birth defects are reduced, the risk opposed to the mother are greatly increased, such as premature delivery.
Spina bifida treatment does not end with the initial surgery performed. Shunts may also be placed later to reduce complications from babies born with hydrocephalus. The forms of spina bifida with physical manifestations have irreparable nerve damage that has already occurred. Ongoing care from a team of specialists are usually needed. Paralysis, bladder, and bowel problems normally remain, so treatment of these conditions begins as soon as birth. Babies with myelomeningocele start physical therapy to prepare their bodies for appliances to help them eventually walk. As the child grows, additional surgeries may be needed to help properly close and grow the spinal column.

Prevention by the administration of folate during pregnancy is obviously paramount. Opinions as to proper management of the established lesion vary considerably. Excision and closure of the coverings of the meningomyelocele in the first few days of life are advised if the objective is to prevent fatal meningitis. After a few weeks or months, as hydrocephalus reveals its presence by a rapid increase in head size and enlargement of the ventricles on the CT scan, a ventriculoperitoneal shunt is required.

BIBLIOGRAPHY


3. Peter S. 2013. Spina bifida resource center

4. Rossi T. 2012. Spina bifida. Pathology project