Fetal neural tube defect

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Neural tube defects are a group of congenital malformations in which the spinal column is bifid as a result of failed closure of the embryonic neural tube.

They originate at various times during gestation.

Epidemiology

Neural tube defects (NTDs) are the most common congenital anomalies.

The most common NTDs are anencephaly and meningomyelocele, which arise during the process of neurulation, a well-defined period between the 17th and 30th day after ovulation.

Myelomeningocele (MMC) is the most common neural tube defect.

The incidence of NTDs is declining in several areas of the world, including the United States. While the reasons for this are unclear, there seems to be some effect from prenatal diagnosis, genetic counseling, and possibly nutritional supplementation [1)].

Classification

These NTDs are clinically apparent by being open, ie, leaving nervous tissue exposed, in contrast to postneurulation NTDs that are skin covered.

Open neural tube defect, which are more common.

Closed neural tube defect rare type.

Lemire classification

A system adapted from Lemire.

1. neurulation defects: non-closure of the neural tube results in open lesions

a) craniorachischisis:total dysraphism.Many die as spontaneous abortion

b) anencephaly: AKAexencephaly.Due to failure of fusion of the the anterior neuropore.Neither cranial vault nor scalp covers the partially destroyed brain. Uniformly fatal. Risk of recurrence in future pregnancies: 3%

c) meningomyelocele: most common in the lumbar region

● myelomeningocele (MM)

● myelocele

2. postneurulation defects: produces skin-covered (AKA closed) lesions (some may also be considered “migration abnormalities”)

a) cranial

● microcephaly

● hydranencephaly: loss of most of the cerebral hemispheres, replaced by CSF. Must distinguish from severe hydrocephalus

● holoprosencephaly

● lissencephaly

● porencephaly distinguish from schizencephaly

● agenesis of corpus callosum

● cerebellar hypoplasia/Dandy-Walker syndrome

● macroencephaly AKA megalencephaly:

b) spinal

● diastematomyelia, diplomyelia: see Split cord malformation

● hydromyelia/syringomyelia

Etiology

see Neural tube defect etiology.

Diagnosis

Maternal Serum Ischemia Modified Albumin (IMA) values were significantly higher in the study group. The IMA was proven to be a predictor with a sensitivity of 77.4% and specificity of 100% for Neural tube defects at a cut-off value of 1.32 [2)].

Complications

They can be associated with hydrocephalus.

Bronberg et al. found a significant decrease in the risk of all fetal deaths due to NTDs, particularly anencephaly, in Argentina over the study period, with the most reduction observed during the mandatory flour fortification era (introduced in Argentina in 2002). The inclusion of fetal deaths in NTD surveillance, coupled or uncoupled with other pregnancy outcomes, is essential for monitoring preventive supplementation measures [3)].

Prevention

There are several measures that can significantly reduce the risk of NTDs. Here are some key strategies for prevention:

Folic Acid Supplementation: Taking folic acid before and during early pregnancy is one of the most effective ways to reduce the risk of neural tube defects. Folic acid is a B vitamin that helps in the formation of the neural tube. Women of childbearing age should take 400 to 800 micrograms (mcg) of folic acid daily, even if they are not planning to become pregnant. If a woman has had a previous pregnancy affected by an NTD or is at higher risk, her healthcare provider may recommend a higher dose.

Dietary Sources of Folate: In addition to folic acid supplements, it's essential to include folate-rich foods in the diet. These include leafy greens, legumes (beans, lentils), fortified cereals, and citrus fruits.

Preconception Planning: Planning a pregnancy in advance allows women to start taking folic acid supplements and make other necessary lifestyle changes before conception. This includes addressing any chronic health conditions and discussing medications with a healthcare provider.

Managing Diabetes: Women with diabetes should work closely with their healthcare team to control their blood sugar levels before and during pregnancy. Poorly controlled diabetes can increase the risk of NTDs.

Avoiding Certain Medications: Some medications, such as certain antiseizure drugs and medications containing valproic acid, can increase the risk of NTDs. Women who are taking these medications should consult their healthcare provider before becoming pregnant to explore alternative treatments.

Avoiding Alcohol and Smoking: Alcohol and smoking during pregnancy have been associated with an

increased risk of NTDs. Avoiding these substances is crucial for fetal health.

Managing Obesity: Obesity in pregnancy is associated with an increased risk of NTDs. Maintaining a healthy weight before and during pregnancy can help reduce this risk.

Genetic Counseling: In some cases, neural tube defects may have a genetic component. Couples with a family history of NTDs or who have had a previous child with an NTD may benefit from genetic counseling to assess their risk and discuss potential preventive measures.

Prenatal Care: Receiving regular prenatal care is essential for monitoring the health of both the mother and the developing fetus. Early detection and management of any potential issues can help reduce the risk of complications.

It's important to note that while these measures can significantly reduce the risk of neural tube defects, they do not guarantee prevention in all cases. Some NTDs can occur due to genetic factors or other unknown causes. Therefore, early prenatal care and genetic counseling can be valuable for assessing individual risk and ensuring the best possible outcomes for both mother and baby.

Case series

There is a huge burden of neural tube defects (NTD) in Ethiopia, and surgical management is not readily available. Mengiste et al. aimed to assess the clinical profile and hospital outcomes of children with NTD who were operated on in Hawassa University Comprehensive Specialized Hospital, Hawassa, Ethiopia.

A retrospective cross-sectional study on 250 children with NTD who were treated in a tertiary hospital from March 2016 to May 2020 was conducted to describe the clinical profile and treatment outcome at discharge. Logistic regression analysis was carried out to evaluate factors that determine mortality.

Out of the 250 children, 50.4% were male. Myelomeningocele was the most common type of NTD (77.2%) followed by meningocele (10.4%). Only 3 mothers (1.2%) received periconceptional folic acid. Prenatal diagnosis of NTD was made in only 22 (8.8%) cases. 52.8% of the NTDs were ruptured at presentation and 50.8% had associated sepsis. At presentation, 42.4% were ≤72 hours of age and only 18 neonates (7.2%) were operated on within 72 hours of admission. 54% had associated hydrocephalus, 31.6% had Chiari II malformation and 19.6% had club foot. Surgical site infection, post-MMC repair hydrocephalus, and meningitis were seen in 8%, 14% and 16.8% of the participants, respectively. The mean duration of hospitalization was 24 ± 14.4 days. Twenty patients (8%) died before discharge from hospital. Prematurity [AOR: 26 (95% CI: 8.01, 86.04), P < 0.001] and the presence of meningitis [AOR: 3.8 (95% CI: 1.12,12.9), P = 0.03]were determinants of mortality.

NTDs are a substantial health problem in this part of the country. Periconceptional folic acid supplementation is almost non-existent. Prenatal detection of NTDs is very low and management is delayed in the majority of cases. Myelomeningocele is the most common type of NTD. There is high in-hospital mortality and prematurity and the presence of meningitis are its determinants [4].

Retrospective study of the association of hydrocephalus with neural tube defect (spina bifida or cephalocele) managed over a period of 7 years at the Department of Neurosurgery, University Hospital Yalgado Ouedraogo.

Thirty-eight cases were included. The mean age was 8.1 months, and the sex ratio was 0.81. There were 27 cases of spina bifida and 11 cases of cephalocele associated with hydrocephalus. A cerebral CT scan was performed in all patients. In 30 cases, the operative management of these pathologies was performed at the same operative time. Eight cases were operated in 2 separate operative stages with a mean time of 30 days between the 2 operations. The course was favorable in 22 patients operated by the simultaneous approach and in 3 patients operated by the separate approach (p = 0.07).

Surgical management of the association of hydrocephalus with neural tube defect in 1 or 2 operative stages gave similar clinical results. However, the treatment in 1 surgical stage would considerably reduce the charges [5)].

1)
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2)
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3)
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