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CASE REPORT

E.P.C.M.S.

A Rare Case of Anterior Encephalocele in a Hispanic Patient

M. Traci Groening, D.O.
Jose L. Gonzalez, M.D.
Zuber D. Mulla, MSPH, PhD
Texas Tech University H.S.C.

Background Information

Neural tube defects are severe congenital malformations that lead to severe handicap, major health costs and devastating social implications. Neural tube defects have an overall U.S. incidence of 1.5 to 2.0 pregnancies per 1000. The incidence is lower for patients of African or Asian descent (1). Screening for neural tube defects is done with maternal serum alpha fetal protein (MSAFP) levels between 14 and 20 weeks gestation. The detection of neural tube defects is done either by ultrasound and or the presence of acetylcholinesterase in amniotic fluid obtained through amniocentesis. The sensitivity for the detection of neural tube defects generally increases with gestational age, but detection of a neural tube defect has been made as early as 13 weeks.

Neural tube defects can include an array of defects resulting from the failure of the embryonic neural tube to close within 28 days from conception. Neural tube defects can include anencephaly (an absence of the bony calvarium), encephalocele (a herniation of the meninges or cerebral tissue through a defect in the calvarium), iniencephaly (herniation involving the high cervical spine), meningomyelocele or spina bifida cystica (herniation involving the lower thoracic and lumbar spine) and spina bifida. A direct relationship with low dietary folic acid has been linked as an environmental cause of neural tube defects, although most of the cases are multifactorial (1).

Encephaloceles can either be classified as posterior (occipital), anterior (frontoethmoidal), parietal or sphenoidal. This malformation is the result of failure of the surface ectoderm to separate from the neuroectoderm. This bony defect in the skull allows for herniation of the meninges or brain tissue; herniation of brain tissue is a poor prognostic sign. Unfortunately, only about 3% of patients with an affected fetus have an elevated MSAFP because most encephaloceles are covered by skin. Of patients with encephalocele, 21% are born live but only half survive, with a high incidence of mental impairment. Even after surgical repair, small encephaloceles may reoccur. Anterior encephaloceles, especially if there is no brain tissue involved, have a better prognosis than posterior encephaloceles.

The occiput (posterior) is the most common site for encephaloceles in the US and Western Europe. Frontoethmoidal encephaloceles are most common in Russia and Asia and account for 13-15% of the encephaloceles seen. Other anomalies are commonly seen with encephaloceles including chromosomal abnormalities (13-44%). Karyotyping should be offered to these patients. Associated anomalies include genetic syndromes such as Meckel-Gruber, von Voss, Chemke, Roberts, and Knobloch syndromes. Maternal rubella infection and maternal diabetes, especially insulin dependent diabetes, have been associated with increases in encephalocele. Alterations in glycemic control, in fact, have been linked to neural tube defects in general (2).

Case Presentation

This patient is a 21 year old primigravida that came to our care at 34 weeks gestation. She had a late entry into prenatal care, so early serum screening with MSAFP was not offered. Her history and physical exam were unremarkable. The routine prenatal laboratories were unremarkable. On a routine ultrasound, a defect was seen in the fetus, and this was originally diagnosed as an abdominal wall defect. Upon consultation with a Maternal Fetal Medicine specialist, the patient was correctly diagnosed with a large anterior encephalocele. The size of the lesion was judged not to be compatible with life. The patient was counseled about all options including pregnancy termination versus continuation of the pregnancy. She chose pregnancy termination. She delivered a single liveborn male with Apgars of 2/4/4 (1 heart rate/1 respiratory effort, 1 heart rate/1 respiratory effort/1 color/ 1 tone) at 1, 5, and 10 minutes, respectively. The neonate had an anterior encephalocele (Fig 1,2). The neonate shortly expired with comfort care provided. The placental pathology revealed a mature placenta with mild villous edema, focal microvilli formation, recent and remote infarcts and occasional calcifications. The fetal membranes had no evidence of inflammation.

Discussion

The prevalence of encephalocele has been reported to be 0.8 to 5.6 per 10,000 births. In Texas, encephaloceles were more common among the offspring of Hispanic women, rather than Caucasian women (1). Anterior encephaloceles are not as prevalent in Texas as posterior lesions. This finding follows general encephalocele trends seen in the United States.

Folic acid helps in the synthesis of DNA and RNA by mediating transfer of one carbon units. Without adequate stores of folic acid, abnormal cell division will ensue.

Folic acid supplementation has been shown to decrease the incidence of all neural tube defects, including encephaloceles. It has been estimated that 50 to 70% of all neural tube defects

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could be prevented if the women had been on folic acid supplements (3). The March of Dimes has recommended that all women of child bearing age be on folic acid, 0.4 mg daily one month before conception and during the first trimester of pregnancy. If a woman has had a previous child with a suspected neural tube defect, she should be taking 4 mg of folic acid daily. A minimum erythrocyte folate level of 900 nmol is needed to protect against neural tube defects (3). Since January 1998, the FDA has made folic acid fortification of grains, cereals and pasta mandatory in the United States. There has been an estimated 19% decrease in neural tube defects since then.

As providers, we should be informing our patients to eat a healthy, balanced diet with a daily folic acid supplement. A prenatal vitamin usually contains between 0.8 to 1.1 mg of folic acid. However, daily compliance with taking a prenatal vitamin has been found to be only about 53-58% among pregnant women (3). Some studies now suggest 5 mg of folic acid should be the new standard for prenatal vitamins. It's been hypothesized that 5 mg daily would prevent almost 90% of women from having a baby with a neural tube defect, which is an increase from current recommendations. Together with information provided by organizations such as the American College of Obstetricians and Gynecologists, the American Academy of Pediatrics and the March of Dimes, we can educate our patients about the need for folic acid supplementation.

REFERENCES

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3. Koren G. 2007. Increasing Folic Acid Supplementation. OBGYN News 42:9. 2007.

Figure 1

Figure 2

M. Traci Goening, D.O., Texas Tech University Health Sciences Center, El Paso, Texas.

Jose L. Gonzalez, M.D., Texas Tech University Health Sciences Center, El Paso, Texas.

Zuber D. Mulla, MSPH, PhD, Texas Tech University Health Sciences Center, El Paso, Texas.