Obstetric Ultrasound Quiz

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DOI: http://dx.doi.org/10.5915/21-1-5521

Case Report

Mrs. J. J. is a 25-year-old, G3P2002, white female. Mrs. J. J. had stopped birth control pills about one year previously and since then had regular menstrual cycles. Her menstrual dates are known with certainty. At 16 weeks gestational age (menstrual dates), she had a maternal serum alpha fetoprotein (MSAFP) screening test. The result was 41 ng/ml (10.8 MOM). The test was repeated the following week and the result was 470 ng/ml (11.2 MOM).

She was referred for an ultrasound examination at 18 weeks gestation. Additional history obtained at this time indicated no prior malformed offspring, no genetic diseases in the patient's or her hushand's families, no exposure to drugs or radiation, and a family history of diabetes.

Realtime ultrasound examination was performed. Representative ultrasound pictures are shown. (Figures 1 and 2)

What is your diagnosis?

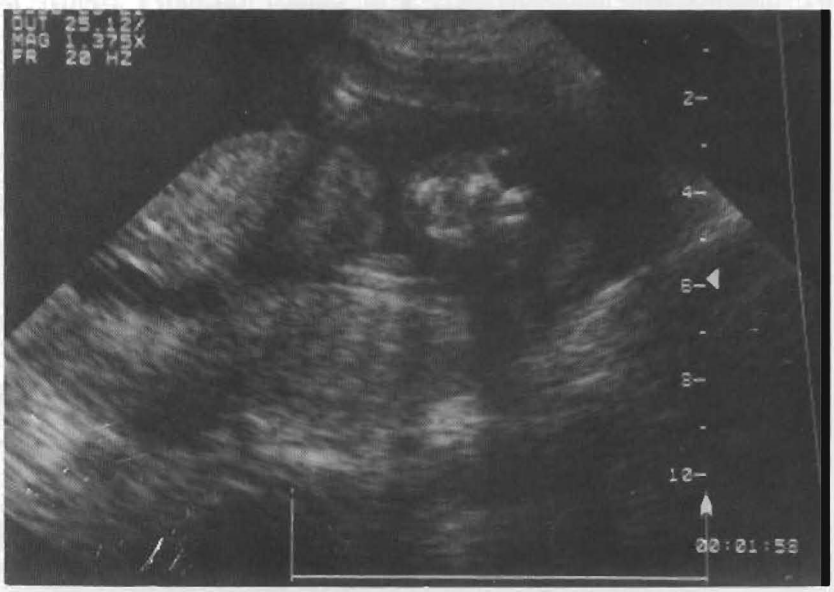

Figure 1. A longitudal midline scan. Pregnancy at 16.5 weeks. Posterior placenta.

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For the correct diagnosis and a review of the condition, refer to page number 42.

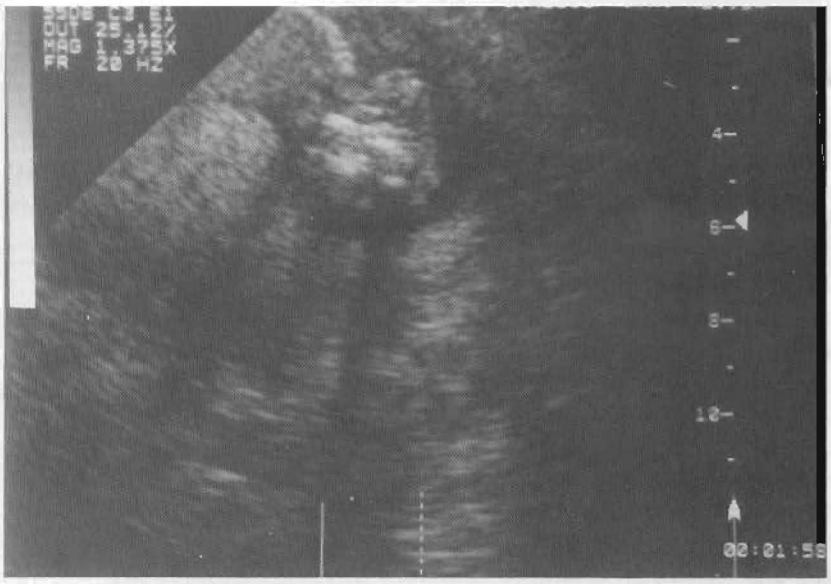

Figure 2. A longitudal parasagittal scan showing the fetal head in the lower uterine segment.

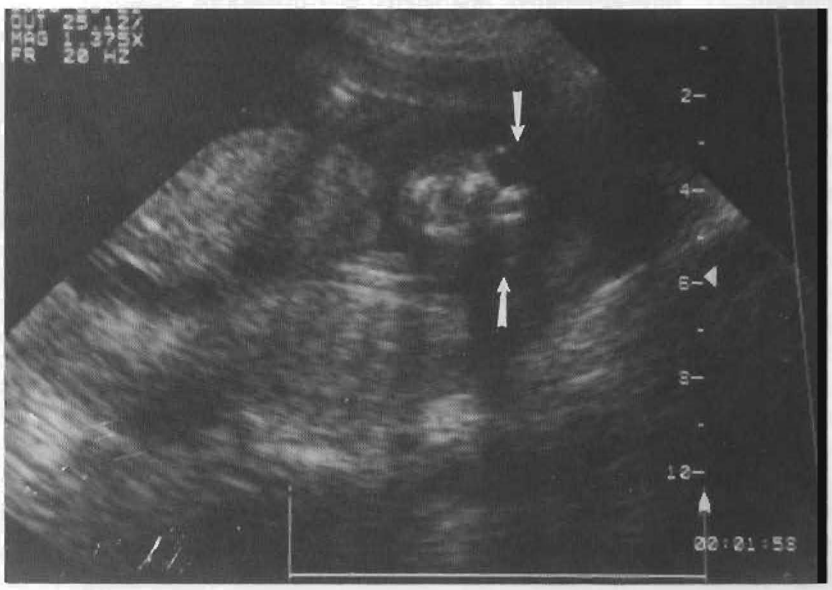

Figure 3. Same as Figure 1. The arrows point to the orbits. The nose and mouth are intact.

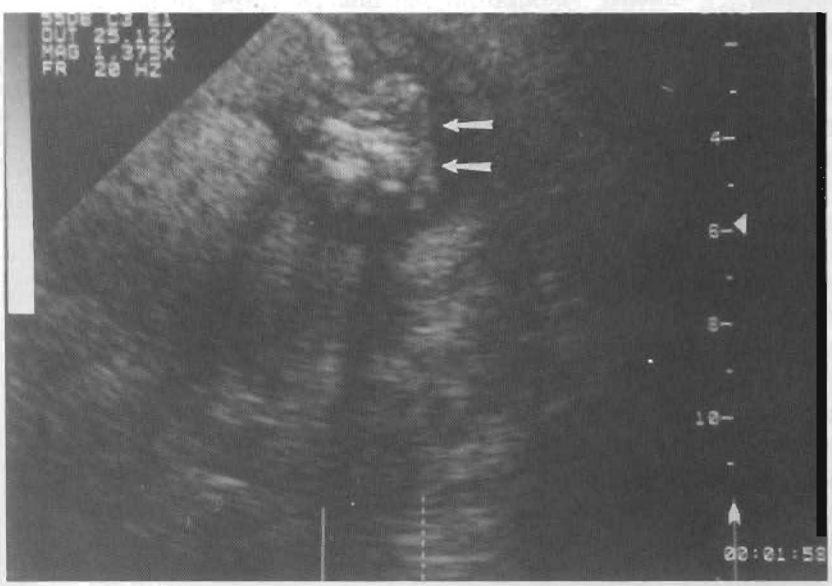

Figure 4. Same as Figure 2. The arrows point to the top of the fetal head. Only the base of the skull is well formed. The cranial vault is absent. No cerebral hemispheres [anencephaly].

Anencephaly

The ultrasound examination revealed a single fetus. The mean ultrasonic gestational age was 16.5 weeks. The amniotic fluid volume was normal.

A cranial vault could not be identified and there was no evidence of cerebral hemispheres. The face could be seen and was normally developed. (Figure 3) The spine was intact as seen in the longitudianl; sagittal and coronal, sections as well as in the transverse scans. Examination of the heart and abdoninal viscera revealed no additional abnormalities.

Diagnosis: Anencephaly

Discussion:

Anencephaly is characterized by the absence of both cerebral hemispheres as well as the cranial vault. It is to be distinguished from acrania, a condition in which the cranial vault is partially or completely absent while the cerebral hemispheres are present but are usually abnormally developed. Acrania is much rarer than anencephaly.

MSAFP testing is a recommended strategy for the diagnosis of neural tube defects (NTDs) including anencephaly and spina bifida.[1] This patient had two elevated MSAFPs. Ultrasound examination is indicated in such situations to rule out other causes of elevated MSAFP for example, a pregnancy that is more advanced than suspected, unrecognized twin pregnancy, or fetal death. Sonography will also confirm or rule out the diagnosis of anencephaly or other NTDs.

Anencephaly is easily diagnosed by sonography (Figures 3) as early as the 12th-13th weeks, based on the failure to demonstrate the cranial vault. The orbits are present but there is no normal bony structure or brain tissue cephalad to the orbits, the neck is short, and the fetus has a characteristic frog like appearance.[2] Polyhydramnios is represented in approximately 50% of the cases but is not usually apparent until the 26th week. The differential diagnosis includes amniotic band syndrome involving the head with loss of large portioins of the cranial vault, and severe mirocephaly. The latter is especially important to rule out in the third trimester, particularly at term when the fetal head is already engaged (or low) in the pelvis. If the fetus is not anencephalic, the next common cause (for increased MSAFP) is spina bifida. The diagnosis of spina bidia (meningocele/meningomyelocele) can be missed unless the examiner is an experienced sonographer/sonologist (Level II ultrasound examination). Occasionally a small defect involving only one or two vertebrae, especially if it is in the sacral region, may still be missed.[2,3] Therefore, if no explanation for the elevated MSAFP is found on sonography, amniocentesis and determination of amniotic fluid alpha fetoprotein and acetylcholinesterase is indicated. MSAFP screening is successful in identifying more than 85% of cases of anencephaly and 70% to 80% of cases of meningmyelocele.[1]

The incidence of anencephaly ranges between 0.2/1,000 births of U.S. blacks, 0.6/1,000 births in Japan, 1.2/1,000 U.S. whites to 3.5/1,000 births in the British Isles. The incidence in abortion material is approximately five times higher than at birth.[2] The incidence of NTD is increased 3-4 fold in diabetic pregnancies. It is noteworthy that this patient has a family history of diabetes but has not been tested during this pregnanty.

Anencephaly results from failure of closure of the anterior neuropore. An alternative explanation is that excess cerebrospinal fluid early on in embryonic development causes disrupture of the normally formed cerebral hemispheres.[2]

Anencephaly is commonly associated with other fetal anomalies. Associated malformations included: spina bifida (17% of cases), cleft lip/palate (2% of cases), and club feet (1.7% of cases).2

Only 1/3 of these fetuses are born alive and the disease is unfortunately fatal within a few hours or days. Termination of pregnancy should be offered to the patient at any time the diagnosis is made.

Another option that has been considered, recently, is to continue such a pregnancy in the hope that a live birth results and might be used as an organ donor. This, however, raises serious ethical questions and concerns.[4,5]

The risks of recurrence of NTDs in a subsequent pregnancy is 2 to 3% after one affected pregnancy and 5 to 10% after two affected pregnancies. In subsequent pregnancies, therefore, a detailed ultrasound examination is recommended as early as 13-14 weeks; MSAFP/amniocentesis at 16 weeks is recommended if ultrasound does not show an abnormalty in the fetus.

References:

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