INTRODUCTION

Failure of proper closure of the neural groove to form an intact neural tube, normally completed in 28 days, causes anencephaly anteriorly and spina bifida in the mid- and caudal portion of the groove. The recurrence risk for defects of neural tube closure, about 5% for parents who have had one affected offspring, includes approximately an equal risk for either anencephaly or spina bifida. This indicates a common basis for both disorders \(^3\). The risk rises to 10% or more following the birth of a second affected child \(^2\). Also, a short interval between conceptions appears to increase the risk of recurrence \(^1\). In Britain anencephaly and other major malformations of the central nervous system account for a major proportion of perinatal deaths \(^3\). If suspected, anencephaly can be diagnosed easily in late pregnancy by a single roentgenogram. Russell \(^3\) correctly diagnosed all 88 patients referred for radiologic studies. Another 25 anencephalics were detected in a group of patients examined roentgenographically, without clinical suspicion of fetal anomalies. When suspected, due to the birth of previous defective infants, anencephaly can be detected by sonography, amniocentesis and amniography, early enough in pregnancy to allow for safe interruption.

Anencephaly was recently diagnosed in our Institutions during the second trimester in 3 cases to be reported here.

CASE REPORTS

Case no. 1

M.T., 37-year-old white female, para 0010, of Scottish and English extraction, came to us at 16 weeks of pregnancy for a diagnostic amniocentesis. Indication for the procedure was her advanced age, specifically to rule out fetal Down’s syndrome \(^4\). No birth defects were reported in the patient’s family history, nor in that of her husband’s, with the exception of a first cousin of the

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patient, who is 14-year-old and has achondroplasia. Following our laboratory's routine procedure, sonographic placental localization was obtained prior to amniocentesis. On the sonogram the fetal head could not be outlined (fig. 1). Amniotic fluid cells were set up in culture and their chromosome analysis, completed some 4 weeks later, revealed a normal 46,XX karyotype. Alpha-fetoprotein (AFP) in the amniotic fluid measured 16.5 mg%, which is approximately ten times the normal value for that stage of pregnancy. At 20 weeks, sonar was repeated and confirmed the absence of a fetal head (fig. 2). Amniocentesis was repeated to perform an amniogram and a second control of the alpha-fetoprotein concentration. The AFP value was 5.4 mg%, much lower than the previous one, but still more than double the normal level for that gestational period and consistent with the diminishing AFP concentration found as pregnancy progresses.

An amniogram was performed with injection of 20 ml of methylglucamine diatrizoate (Renografin 60) and the radiopaque material dispersed uniformly throughout the amniotic fluid outlining the soft tissues of the fetus. The normal soft tissues of the back and buttocks were well outlined with no malformations. The cranial vault was absent with evidence of a few facial bones, confirming the diagnosis of anencephaly (fig. 3). A 4-h delayed film revealed slight opacification of the gastrointestinal tract.

At 21 weeks, pregnancy was interrupted by intra-amniotic instillation of hypertonic saline solution. Within 36 hrs labor ensued and 2 hrs and 40 min later the patient was delivered of a 280 g female anencephalic fetus (figs 4 and 5), measuring 16 cm crown to rump. A roentgenogram of the skeleton revealed flattened facial bones with no skull (fig. 6). Shortly thereafter the placenta was expelled. At autopsy nothing resembling cerebrum or cerebellum was found. The exposed membrane over the base of the skull consisted of thin-walled blood vessels, mixed with glial and fibrous tissue. Some disorganized masses of ependymal cells were present close to the mid-line.

Case no. 2

G.N., 26-year-old white female, class B diabetic primigravida. No birth defects revealed by the family history. The patient developed hydramnios at 24 weeks. The sonogram, promptly obtained, failed to outline the fetal head (fig. 7). Four weeks later another sonogram confirmed, and an abdominal X-ray corroborated the diagnosis of anencephaly. Pregnancy was interrupted by hysterotomy and a 500 g male anencephalic fetus delivered. The parents refused permission for an autopsy. At the time of this writing the patient is pregnant again. At 18 weeks a sonogram revealed a normal fetal head.

Case no. 3

A.Z., 25-year-old white female, para 1001, noted some vaginal bleeding at 17 weeks of gestation. There was no history of birth defects in her family. A sonogram was obtained to rule out placenta previa. No fetal head could be visualized (fig. 8). The diagnosis of anencephaly was confirmed at 21 weeks by another sonogram, and at 24 weeks by X-ray. Mild hydramnios developed. At 28 weeks, pregnancy was interrupted by intra-amniotic instillation of hypertonic saline solution and oxytocin infusion. Within 30 hrs the patient was delivered of a 720 g female still-born anencephalic fetus (fig. 9). Placenta was removed manually. Permission for autopsy was refused.

COMMENTS

In 2 of the 3 cases reported here the early antenatal diagnosis of anencephaly was serendipitously made by sonography employed to localize the placenta prior to amniocentesis in case no. 1 and to rule out placenta previa in case no. 3. In case no. 2 the association of hydramnios and diabetes indicated the strong possibility of fetal malformation.

Hydramnios is associated with an increased perinatal mortality and a high incidence of fetal anomalies. Jacoby and Charles have reviewed the pertinent literature and found that anencephaly varied in frequency from a minimum of 6.3 % to a maximum of 39 % in the analysis of a total of 3,331 infants from 15 series reported, including 168 of their own cases. According to Scott and Wilson, hydramnios accompanies nearly half of the cases of anencephaly. Hydramnios, therefore, when evident in the second trimester, strongly indicates fetal evaluation studies * Measurements done in Dr. A. Milunsky's Laboratory, at the Eunice Kennedy Shriver Center, in Waltham, Massachusetts.