

Prenatal diagnosis of anencephaly and acrania in pregnant females – Report series of eight cases

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ABSTRACT


Acrania and anencephaly are characterized by the partial or complete absence of skull and brain tissue. Due to this, the neural tissue is exposed and it leads to non-function of few parts of the hemispheres. A total of eight cases of acrania and anencephaly were included in the report. Five cases were acrania and three cases were anencephaly. Both cases were diagnosed between 15 and 24 weeks of gestation period. In the present report, all the pregnant women carry fetuses with neural tube defects. Out of five acrania cases, there is a partial or complete absence of cranium in all the cases, and in one case, there is a twin intrauterine gestational sac that was noted. However, out of twin sac, the first one is having a good heart rate (155 bpm) and the second fetus is having no cardiac activity, i.e., early fetal demise. On the other hand, out of three anencephaly cases, two anencephaly cases were diagnosed with partial absence of the fetal brain and the complete absence of the cranium, and in one case, there is a partial absence of cerebral parenchymal tissue above the orbit with the absence of cranial vault. By the use of ultrasonography, we can diagnose the anencephaly and acrania at a very early stage without any side effects. The cause of anencephaly and acrania is dependent on the number of factors one of which is a folic acid deficiency. Sentence is reviewed and corrected): It is always better to consume the folic acid supplements that are advised by the physician during the planning of pregnancy to avoid the congenital anomalies of the fetus like anencephaly and acrania. The ultimate focus of the study is to evaluate the morphology of the fetus in case of anencephaly and acrania which could lead to the early detection of abnormalities and also to create awareness among people to take folic acid supplements to eschew such abnormalities.

KEY WORDS: Anencephaly; Alpha-fetoprotein Test; Acrania; Ultrasonography

INTRODUCTION

Anencephaly is a congenital birth anomaly in which the maximum portion of the brain, scalp, and vault of the skull is absent and acrania is also a connate abnormality, in which there is an absence of fetal skull with freely exposed brain tissue to the amniotic fluid. Neural tube defects are the most common connatural malformation which will affect the

natural development of the fetus. These neural tube defects are categorized into spinal and cranial defects. The cranial defects are classified into anencephaly, iniencephaly, and encephalocele. The anencephaly is further divided into holoanencephaly and meroanencephaly.^[1] Approximately three-fourth of the congenital cases are born and almost all infants die within the week after the birth.^[2] Neonate is born with a permanent unconscious stage due to the dearth of the functioning of the outer layer of cerebrum composed of gray matter which plays an important role in consciousness and varying degrees of brain stem function causing brain death of the infant. About 65% of the cases of anencephaly die in the uterus and few may be delivered in preterm labor.^[3] Ultrasonography is the modality of choice to diagnosis the anencephaly and also can be diagnosed by

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an elevated level of alpha-fetoprotein. The main focus of this report is to evaluate the morphology of the fetus in case of anencephaly and acrania which could lead to the early detection of abnormalities and also to create awareness among people to take folic acid supplements to eschew such abnormalities.

CASE REPORTS

The study included eight unborn fetuses. In ultrasonography, out of eight fetuses, five were live fetuses with acrania and three were live fetuses with anencephaly. This case study was performed between March 2017 and November 2019. Fetal ultrasonography was performed on GE Logiq P5 with linear and convex high resolution and trans-vaginal probe. Before taking the data from the patient, the significance of the study was well explained and the confidentiality of the study participants was maintained throughout the study. Informed consent was obtained from each patient before the ultrasonography examination.

Cases Scenario 1: Acrania

Out of five live fetuses with acrania, four live fetuses were correctly identified at 15–24 weeks of gestation. All the four live fetus heart rates were within the range of 146–162 bpm and the weight range was from 147 to 585 g. In all the four cases, fetal long bone appeared normal, the placenta was anterior in position with normal in thickness, all four chambers of the heart were clearly visualized, both kidneys were seen with no evidence of hydronephrosis and no evidence of cord around the neck. In all four cases, interhemispheric fissure, brain convolutions, and sulci were clearly identified; however, ultrasound revealed a well-formed fetal brain with near-complete absence of the cranium (acrania) [Figure 1].

On the other hand, one live fetus was correctly identified at the 15th week of gestation with a heart rate of 155 bpm, and twin intrauterine gestational sac was visible. In this twin's gestational pregnancy, the first fetus has shown normal cardiac activity, but there was an absence of calvarium and cerebral hemispheres were surrounded by a thin membrane. There was evidence of brain parenchymal tissue floating within the amniotic fluid and the second fetus had been reduced to an amorphous soft tissue mass devoid of any cardiac activity, i.e., early fetal demise was diagnosed.

Cases Scenario 2: Anencephaly

Out of three live fetuses with anencephaly, two live fetuses were properly identified at 18–22 weeks of gestation. Two live fetuses were having a heart rate of 142 bpm with the weight 294 ± 47 g and 481 ± 77 g, respectively. On the observation, both orbits were symmetrical in size and shape,

fetal nose and nasolabial folds were recognized clearly. The same fetus's cervical, thoracic, and lumbar spine appeared normal with no morphology of the spinal canal. No mass was seen attached or arising from the spine but on the observation, both the fetuses showed absence of fetal brain with the complete absence of the cranium in the coronal scan [Figures 2 and 3].

On the other hand, one live fetus was correctly identified at 16 weeks of gestation. The heart rate of the fetus was 149 bpm with a weight of 154.67 g. On the observation of single live intrauterine pregnancy, the placenta was present anteriorly with no evidence of placenta previa and amniotic fluid was normal. Spine, kidneys, heart, four limbs with all long bones, fetal stomach, and bladder bubbles were seen normal. There was an absence of cerebral parenchymal tissue above the orbit with the absence of cranial vault which was diagnosed. On the observation, the fetus of bilateral prominent orbits showed a "Frog face sign" appearance in both prenatal and postnatal [Figures 4 and 5].



Figure 1: Antenatal ultrasonography reveals well-formed fetal brain with near-complete absence of the cranium (acrania)



Figure 2: Coronal scan of fetus showing deficient cranial vault – acrania

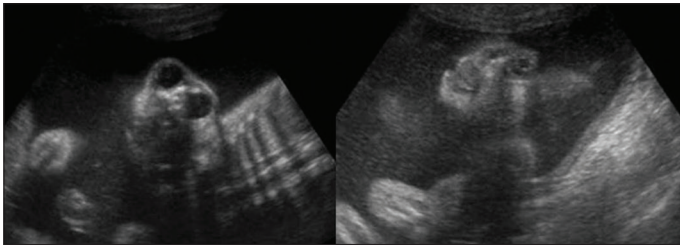


Figure 3: Coronal and parasagittal views of the fetal face with missing cranial vault



Figure 4: Anencephaly resulting in the “Frog Face sign” due to an absence of recognizable tissue superior to the level of the fetal orbits

Out of eight patients, one patient had a history of inter uterine pregnancy of an anencephalic fetus at the 24th gestational week 3 years before, and in the present cases, she is having the same diagnosis [Figure 6].

DISCUSSION

Anencephaly results in the failure of neural tube closure which lead to incomplete development of brain cell and calvaria. The prevalence of neural tube defects may vary from 0.5 to more than 10 per 1000 pregnancies. There are many risk factors that may influence birth defects such as diabetes, obesity, and change in nutritional status. Anencephaly is more prevalent among males than females.^[4] The couple who are planning to conceive should aware of the significance of folic acid supplements and give suggestions to take at least 400 µg/day of folic acid.^[5] Fortification of both wheat and maize flour is important to prevent the neural birth defect.^[6,7]

Mostly, the neural tube defects will start before the 4th week following the conception, so after conception alpha-fetoprotein test is very important for the early diagnosis of different neural tube defects like anencephaly. The elevated normal level of alpha-fetoprotein may indicate that the developing fetus has neural tube defects such as anencephaly and spina bifida. As the patient visited the hospital for ultrasound scan during their 18–23rd weeks gestation period, so the fetus could not be aborted, if she would go for abortion, then she may face serious health issues and her next pregnancy



Figure 5: Postnatal appearance of the baby missing forehead with characteristic anencephalic profile of the fetus

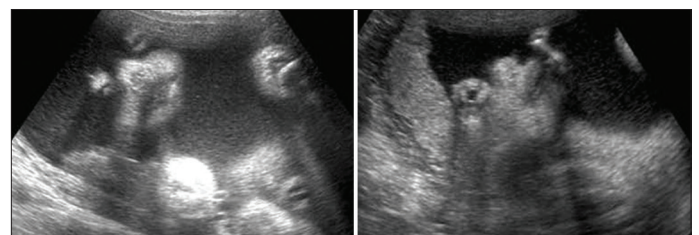


Figure 6: A 24-year-old G3 P2 was referred at 24 weeks of gestation. Ultrasound examination showed increased amount of the amniotic fluid and anencephaly

may also be affected.^[8] It is important that females should go for an ultrasound scan from the 1st trimester onwards.

Chatzipapas *et al.*, 1999, performed a case study based on anencephaly in early pregnancy in which six cases of anencephaly were diagnosed in the early first trimester. Out of six cases, five cases demonstrated the “Mickey Mouse Sign” in the coronal section of the head scan.^[9] In the present study, we diagnosed a few cases which demonstrated the “Frog Face sign” in the prenatal scan.

Alcantara *et al.*, 2016, published a case report based on anencephaly in Mexican Population which demonstrates that Mexico is one of the highest incidences of acrania and anencephaly with a rate of 13.34/10,000 births.^[10] Likewise, Kaur *et al.*, 2013, conducted a case report based on anencephaly in which they observed that the women who were going to get pregnant consumed the suggested amount of folic acid before and after pregnancy, the risk of anencephaly could be decreased by 70%.^[11,12] In the present study, most of the patients are from rural area and they are not aware of folic acid deficiency which might be the cause of such neural defects.

This study was conducted in a rural village of Anand district, Gujarat, India, where people are not aware of prenatal care, and the resultant cases were recorded to let the people aware of the importance of pre- and postnatal care during pregnancy.

CONCLUSION

Anencephaly and acrania can be diagnosed in the first trimester and the health effects of pregnant patients will be reduced. All anencephaly and acrania fetuses usually show an elevated level of serum alpha-fetoprotein. Physicians also advised pregnant females to consume folic acid supplements and good nutrition to avoid fetus birth defects; however, if the fetus is found abnormal in utero, associated anomalies of the particular fetus must be checked. The ultimate focus of the study is to create awareness among the rural population about the health risks with possible causes such as diabetes, nutritional deficiency, and lack of folic acid supplement intakes.

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