ANENCEPHALY AND ITS ASSOCIATED ANOMALIES IN ANTENATAL SCANS

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ABSTRACT

BACKGROUND

With modern real time ultrasonography, anencephaly, a fatal developmental defect of the brain and cranial vault can be detected as early as 11-12 weeks and the pregnant women can be saved from the trauma of unnecessary child bearing for longer periods.

This study is done to detect the incidence of anencephaly in rural pregnant women of this Rayalaseema region and to know how early can anencephaly be detected with ultrasonography.

MATERIALS AND METHODS

This study is done in a period of two years at Santhiram Medical College and General Hospital, Nandyal in 14000 pregnant women, out of which 10 cases of anencephaly were detected with associated anomalies.

RESULTS & CONCLUSIONS

Anencephaly, a fatal developmental cranial defect can be prevented by usage of folic acid in early conception and by genetic counselling in further pregnancies.

KEYWORDS

Anencephaly, Ultrasonography, Neural tube defects, Spina bifida, Maternal serum alpha-fetoprotein, Folic acid, Genetic counselling, Exencephaly (Acrania).

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INTRODUCTION: Anencephaly is a serious developmental defect of the central nervous system and is characterized by the absence of cranial vault and telencephalon.[1] Necrotic remnants of the brain stem and rhombencephalic structures are covered by a vascular membrane.[2] Associated malformations are common and include spina bifida, cleft lip/palate, club foot and omphalocoele.[3] Polyhydramnios is frequently found. Incidence of anencephaly is around 1-2 in 1000 with a female predilection of 4:1.

Ultrasonography is the easy method of detecting anencephaly. Although the foetal head can be identified by vaginal ultrasonography as early as the 8th - 9th week of gestation, the diagnosis may be difficult in the first trimester. Absent calvarial mineralization before 10-12 weeks' gestation makes the differentiation of normal and abnormal brain difficult. So anencephaly can be diagnosed in early second trimester i.e. after 12 weeks. In this study, anencephaly and its associated anomalies are evaluated.

AIMS AND OBJECTIVES: To know the incidence and the early detection of anencephaly in this Rayalaseema region pregnant women and try to know the cause and its preventive measures in future pregnancies.

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MATERIAL AND METHODS: This present prospective study of anencephaly and associated malformations was carried out in Department of Radiodiagnosis of Santhiram Medical College and General Hospital, Nandyal.

Total of 14000 pregnant women were scanned in the period of 2 years from August 2013 to January 2016. All the antenatal scans for ultrasonography were analysed and data was collected with anomalies. Ten cases with anencephaly were detected.

Ultrasound examination was performed using of 3D Esaote My Lab 50 - ultrasound machine.

Inclusion Criteria: All the antenatal cases who came for regular checkup and anomaly scans to departments of Obstetrics & Gynaecology and Radiodiagnosis were included in this study. There is no exclusion criteria. All the pregnant women aged between 18-35 years and with gestational age 10 weeks to 36 weeks were included.

Exclusion Criteria: Nil.

RESULTS: Total of 14000 pregnant women were studied and analysed according to their gestational age of foetus and anomalies noted. An attempt was made to classify them accordingly to the sonographic appearances from the literature available so as to know the associated anomalies. Out of 10 anencephaly cases, 1 case was associated with spina bifida, 3 with polyhydramnios, 1 with omphalocoele

and 1 with hydronephrosis, 2 Foetuses were in transverse lie.

Nuchal Translucency scan is done during 9-14 weeks. During this period, out of 10 anencephaly foetuses, 4 were detected, because these pregnant women attended during the early second trimester.

Before 12 weeks, no cases were detected, this is mostly due to these pregnant women coming for their antenatal checkup most often during the second trimester. Sometimes, even in the end of 3rd trimester for their antenatal checkups.

Out of 10 cases, 8 were female and 2 were male foetuses. These observations were not revealed to the parents and is only for statistical analysis. Most of the pregnant women were in the age group of 18-25 years most of them were primigravida.

In our study, the incidence of anencephaly is 1:1400 as that in the literature is 1:1000.

No lab studies as maternal serum alpha-fetoprotein and amniotic alpha-fetoprotein were conducted in these pregnant women or the foetuses.

DISCUSSION: Anencephaly is a serious developmental defect of the central nervous system in which the brain and cranial vault are grossly malformed. The cerebrum and cerebellum are reduced or absent, but the hindbrain is present. This defect results when the neural tube fails to close during the third to fourth weeks of development, leading to foetal loss, stillbirth, or neonatal death.

The neural tube is formed as closure of the neural groove progresses from the middle toward the ends in both directions, with completion between day 24 for the cranial end and day 26 for the caudal end. Disruptions of the normal closure process give rise to neural tube defects.

Causes: Multifactorial, inheritance in multiple genes,[4] environmental factors and absence of sufficient folates.[5] Folic acid substitution in next pregnancy is always a must.

Prognosis: Anencephaly is lethal in all cases because of the severe brain malformation that is present. A significant proportion of all anencephalic foetuses are stillborn or are aborted spontaneously, die after birth.

Lab Studies.

We did not do any lab Tests.

Imageology of Anencephaly: Though it is said that transvaginal ultrasound can allow detection of anencephaly at 8 to 9 weeks,[6] most of our cases are detected only after 12 weeks to 25 weeks' time. However, they were detected on 1st exam as most pregnant women seek this ultrasound in 2nd trimester.

The typical "frog eye" or "mickey mouse" appearance was seen in 9 of the 10 cases[7], one case it was slightly different in appearance and that foetus also had spina bifida. In three instances where we did ultrasound late after 24 weeks (for women who were first seen) polyhydramnios was present. In pregnancies where the anencephaly detection was early, polyhydramnios was not seen. Incidentally in these women, we also noticed transverse lie in two instances.

Sl. No. of foetus	Age of mother	weeks of foetus	Associated anomalies	Complic-ations
1.	21 yrs.	12 weeks		
2.	24 yrs.	20 weeks	Omphalocoele	
3.	20 yrs.	16 weeks	Spina bifida	
4.	26 yrs.	32 weeks		Polyhydr-amnios
5.	20 yrs.	24 weeks	Hydronephrosis	
6.	19 yrs.	14 weeks		
7.	22 yrs.	26 weeks		
8.	21 yrs.	14 weeks		
9.	25 yrs.	13 weeks		
10.	24 yrs.	22 weeks		Polyhydr-amnios

Table showing age of mother, gestational age of foetus, associated anomalies with anencephaly

	Meta-analysis/ 19 articles	Present study
Total number of pregnant women included in study	308307	14000
Incidence of anencephaly	1:1000	1: 1400

When we compared our studies with incidence chart

Dr. Hangkipe and Frank Gillad et al have stated the incidence of anencephaly 1 on 1000 pregnant scans. However, in our study, the incidence is 1:1400. The male-female ratio of anencephaly of 1: 4 is matching with our study. This author has noted many associated anomalies with anencephaly like other neural defects,

Congenital heart defects,
Cleft palate,
Diaphragmatic hernias,
Spinal dysraphism,
Skeletal anomalies,
GIT anomalies,
Hydronephrosis.

In our study, we noticed the following anomalies;

Total no of anencephalies = 10
Spina bifida: 1 = 10%
Omphalocoele: 1 = 10%
Hydronephrosis: 1 = 10%

Out of 10 foetuses, 3 foetuses have other anomaly;
Associated problems are;

Polyhydramnios:	3 = 30%.
Transverse lie:	2 = 20%.

Percentage of anomalies associated with anencephaly- Overall % is 30%, hydronephrosis is 10%, spina bifida 10% and omphalocoele is 10 %.

Once anencephaly is detected, usually the pregnancy is terminated. Some of the pregnancies were terminated in our hospital, were verified and found correct. Some rural women who do not agree for immediate termination, we could not do followup.

Since it is both retrospective and prospective study, lab investigations were not included. Out of these 10 pregnant women, only 3 said they had consanguineous marriages. To conclude, anencephaly is a foetal defect with grossly poor outcome. It can be detected as early as 11-12 weeks which helps pregnant women to avoid unproductive pregnancy troubles.

CONCLUSION: Anencephaly can be best diagnosed by ultrasonography as early as 11-12 weeks. It can be prevented by taking folic acid tablets as supplements in early conception period, in future pregnancies.

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Fig. 1: Foetus with frog shaped head

Fig. 2: Anencephaly foetus–spine, abdomen and skull

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