

# Frequency of Anencephaly in All Pregnant Population: An Observational Study

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# Abstract

The present study was conducted in the Department of Obstetrics and Gynaecology of Federal Government Services Hospital Islamabad, Pakistan, for a six months period, in order to calculate frequency of anencephaly and identify causative factors in association with births affected with neural tube defects. All pregnant ladies delivered at Federal Government Services Hospital and MCH centre in the six months were included in the study and frequency of anencephaly was computed to be 8.11 per 1,000 births. All first trimester abortions, ectopic pregnancy, septic induced abortions and molar pregnancy were excluded. Among the study group 15 (42.8%) patients were primiparas, mean maternal age was 27.8 years, and majority belonged to lower socioeconomic status. Folic acid had been used by 37.1% patients. Mean gestational age was 25.5 weeks and patients were diagnosed for anencephaly by ultrasound. Major complications recorded were polyhydraminos (97.14%). Females were predominant in anencephalic group. The frequency of anencephaly may be reduced by creating awareness about periconceptional use of folic acid, and early termination of pregnancy.

Keywords: Anencephaly; Neural tube defects; Congenital anomalies; Pakistan

**Abbreviations:** NTD: Neural Tube Defect; CNS: Central Nervous System; MAFP: Maternal Serum Alpha Fetoprotein.

# Introduction

Anencephaly is a neural tube defect (NTD), in which the brain and cranial vault are grossly malformed, the cerebrum and cerebellum are reduced or absent, but the hind brain is present. It occurs during early pregnancy between 24th to 26th days post-conception. Overall incidence of anencephaly is 1 per 1,000, with considerable variation throughout the world. In Pakistan about 6-9% perinatal deaths are attributed to congenital malformations and an earlier data from Faisalabad reported the incidence of anencephaly as 3.2 per 1000 [1,2].

NTDs can be diagnosed antenatally at a time when it is still possible to terminate the pregnancy without putting the life of the expecting mother at stake. Ultrasound screening identifies a growing number of central nervous system (CNS) abnormalities, which helps in the early termination of pregnancy, therefore, resulting in substantial changes in the neonatal presentation of neurological congenital abnormalities [3]. Maternal serum alpha fetoprotein (MAFP) screening has also been shown to be an effective method of screening for NTDs at 16<sup>th</sup> week of gestation, however ultrasound can be used to diagnose as early as the 12<sup>th</sup> week [4]. Diagnosis of 100% anencephalic and 80% spinabifida cases by ultrasound has been reported, however an interesting finding in that particular study was that female sex was predominant in anencephalic foetuses (61.50%) [5]. For anencephaly, known risk factors are maternal age and weight, previous history of NTDs, recurrent miscarriages and intake of antiepileptic drugs [6,7]. Maternal obesity and diabetes are also associated with increased risk of CNS malformations in the offspring, and have a common mechanism [8,9]. An additional risk factor is fever in first trimester for NTDs, and should be treated with antipyretic drugs [10]. Predisposing environmental and nutritional factors for NTDs include drinking water, minerals, intake of folic acid antagonists like Valproic acid, alcohol, smoking and several environmental pollutants [11-13]. In India, NTDs have been reported due to deficiency of vitamin B12, B6 and folate, which result in hyperhomocysteinemia, which is linked to NTDs [14]. In some parts of the world pregnant women are restricted to certain kind of foods, keeping in view that it could be harmful to pregnancy, which further adds to their increased stress or compromised nutrition [15]. Vegetarianism and poverty also predispose to deficiency in these vitamins [16]. The prevalence of NTD has been shown to vary by race / ethnicity, with highest rates among women of Hispanic ethnicity and lowest rates among black and Asian women. A high consanguinity marriage and 100% lack of periconceptional folic acid intake has been attributed to be cause behind NTDs in an Iraqi study [17].

There are certain complications with an encephaly during pregnancy, like polyhydraminos, prematurity, placental abruption and post-dates. Therefore, early prenatal diagnosis on ultrasound and early termination of pregnancy is beneficial for patient so that complications could be prevented. Use of periconception folic acid supplementation has reduced the incidence of anencephalic and spinabifida cases by 50 - 60%, with a significant reduction of infant mortality and disability [18,19]. Frequency of anencephaly can be reduced by realizing the importance of periconceptional the use of folic acid and preventing the associated risk factors with NTDs. Early prenatal diagnosis is helpful in decreasing the indirect prevalence of perinatal morality by offering termination of pregnancy. Paulik have emphasized upon the importance to increase awareness of women in child bearing age towards folic acid use [20]. Oakley has concluded that only 10% folic acid preventable spina bifida is actually being prevented [21]. This study was conducted to determine the frequency and associated causative factors responsible for anencephaly. Thus factors responsible for NTDs should be explored which will help us to manage our patients according to our own social circumstances, diet, social taboos and socioeconomic status.

# **Patients and Methods**

## **Data Collection Procedure**

Data was collected by recording details of the patients, including addresses. It was also checked that whether patients were already registered or not. Then detailed obstetrical history was taken which included menstrual history, gravity, parity, abortions, still births, total live births and age of last child. Patients were interviewed for past illness like diabetes, gestational diabetes, epilepsy and history of fever in first trimester. Family history of twin pregnancy, congenital heart diseases, NTDs, cleft lip, cleft palate and diabetes, was also recorded. In addition, personal history, including history of contraception, appetite, bowel habits, addiction, allergies, weight gain or weight loss and smoking was taken. Diagnosis of an encephaly was initially done by ultrasound during first, second and third trimester depending on the gestational age when the patient was presented. Place and mode of delivery along with associated anomaly, sex, weight, whether anencephalic baby was delivered dead or alive, and if mother had complications like polyhydraminos and postpartum haemorrhage, were noted.

#### **Data Analysis Plan**

Data was recorded on a proforma by collecting observational facts and was entered and analyzed on SPSS version 10. Categorical data of causative factors like history of diabetes, gestational diabetes, history of epilepsy, and history of fever were recorded in the form of frequency and percentage, while numerical data of causative factors like age, monthly income, parity and gestational age were recorded in the form of mean + SD (standard deviation).

#### **Results**

During the study period of six months, 4,313 pregnant patients delivered in Federal Government Services Hospital and MCH centre. Out of these, 35 neonates had anencephaly, giving incidence of 8.11 per 1000 births.

Among the study group 15 (42.86%) patients were primiparas, and 13 (37.14%) patients were grand multipara so frequency was highest in primiparas followed by grand multipara. Only seven (20%) patients fell between parity 1 to 2. Mean maternal age recorded in our study was 27.8 years, ranging between 25-35 years. Among the studied group, 3 belonged to the high risk group of teenage pregnancy, while only one pregnant lady was over 40. A major chunk of the patients were residing in the capital, Islamabad (62.86%), while 28.57% were residents of the adjoining city of Rawalpindi. The remaining three patients were residing in the peripheral area of Islamabad. Regular follow-up was seen in case of 14 patients, while 21 were brought to the hospital only because their medical condition warranted immediate care. Folic acid ingestion in first trimester was only 37.1%.

Although the FGSH mainly caters the public sector employees and their families, yet spouses of 24 patients were employed in the public sector. Analysis from the socio economic angle exhibited that the spouse of one patient was job less, while only two patients out of 35 belonged to the upper class. Majority of the expecting mothers had not completed secondary education, as 16 (45.7%) were uneducated, 14 (40%) had completed eight years of schooling and only 5 (14.3%) had completed secondary school (ten years of formal education).

Even though mean gestational age was 25.5 weeks, 21 patients were diagnosed by ultrasound in second trimester, 12 the third trimester and only 2 were diagnosed in first trimester. No history of miscarriages was present in 25 patients, while 4 patients each reported either one or two abortions. Two patients informed about past occurrence of 4 and 6 recurrent miscarriages, respectively. Gestational diabetes was recorded in case of two patients, and only one patient had history of fever in the first trimester. Details of anencephalic cases are given in Table 1.

		Frequency (%)
Mode of Delivery	Spontaneous Vaginal Delivery	32(91.43)
	Lower Segment Caesarean Section	3(8.57)
Anomalies	Anencephaly	33(94.29)
	Iniencephaly	1(2.86)
	Spinabifida + Anencephaly	1(2.86)
Gender of child	Male	11(31.43)
	Female	23(65.71)
	Ambiguous	1(2.86)

**Table 1:** Details of the anencephalic cases (n=35).

## Discussion

The loss of a wanted pregnancy is always distressing to the couples, especially if this is a case of congenitally abnormal baby. Determination of the associated factors for congenital anomalies continues to be a major concern for obstetricians and other health care professionals concerned with the management of these patients.

Majority of patients in the study group were non booked, the main reasons for late booking were unawareness, poverty and non-availability of medical facilities. Although incidence of anencephaly is 1 per 1000 births in the world, because of multifactor etiology, general prevalence of anencephaly varies considerably in different parts of world. In a study at Faisalabad, incidence of anencephaly was recorded as 3.2 per 1000 births and in India incidence has been reported as 6.57-8.21 per 1000 live births [2]. In northern Iran the prevalence of anencephaly was reported as 1.31 per 1000 births, while it was 0.748 per 1000 newborns in Afyonkarahisar, Turkey, and in Texas it has been reported as 0.281 per 1000 live births [22-24].

In the study group frequency of an encephaly was highest in primiparas and grand multipara as compared to multipara, and it also appeared to decrease with increasing maternal age, mean being 27.8 years (range 25-35) which correlates with the local study in which maternal age was 21 to 30 years and none was above 40 years [1]. However, in our study one patient was more than 40 years old.

Most of the patients belonged to lower SES. Nearly half the women in the study were uneducated and only 14.3% had completed secondary education, again lower educational level depicts direct relation with increased frequency of NTDs which coincides with association between SES (as measured by maternal education, maternal occupation, monthly family income) and anencephaly in a case control study of Mexican population. Lower SES is thought to contribute to the risk of NTDs and rate of anencephaly and spinabifida are usually high [25-28].

Antenatal diagnosis for anencephaly was 100% as compared with 80% detection of spinabifida. In the study group, due to unawareness, 34.3% and 60% of the patients were diagnosed by ultrasound in third and second trimester respectively, and only 5.7% were diagnosed in first trimester, which is the optimal gestational age for early diagnosis.

In our study 91.4% neonates were delivered vaginally after induction of labour with prostaglandin tablet and three

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were delivered by lower segment caesarean section in which two were twins (one anencephaly and second normal baby) at term pregnancy and third was at term pregnancy with polyhydraminos and previous scar.

Bad obstetric history was observed in only 28.57% patients, and 71.43% patients had no history of miscarriages which does not correlate to a study carried in 2006 where 95% patients had history of miscarriages, a known risk indicator for an encephaly in future gestations [24,29]. Due to unawareness, unplanned pregnancies and poverty in our part of the world, majority of the pregnant women do not have access for antenatal checkup because of unavailability of medical facilities, therefore, they are deprived of folic acid ingestion during first trimester [19,21,30]. In our study 37.1% patients had taken folic acid and 62.9% were without folic acid ingestion. Polyhydraminos was present in 97.14%, and 2.9% patients were post-dates. In an Iranian study, polyhydraminos was observed in 90% patients, which matches our study group [31]. In our study group females children were predominant over males and only one child was born with ambiguous genitalia, which coincides with a study carried in China [5]. We did not observe any addiction in our study group, though cigarette smoking and alcohol have been positively correlated with NTDs. Similarly we did not find any patient with epilepsy who is taking antiepileptic drugs. Only one patient with history of fever and two with gestational diabetes delivered affected babies in this study group. Though fever and gestational diabetes are associated risk factors for NTDs in many studies, but in this study it was not predominant.

#### Conclusion

From our study, it was concluded that majority of patients affected with anencephaly belonged to lower SES, were undernourished, lacked education and did not have access to medical facilities. These patients required counselling for proper spacing in subsequent pregnancies, peri conceptional use of folic acid, early antenatal check and ultrasound to avoid maternal and foetal complications. Improving the knowledge and changing the habits of women and medical professionals will be critical in efforts to realize the full preventive potential of folic acid. The global health community must take a concert effort to meet this challenge.

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