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Original Research Article

FREQUENCY OF ANENCEPHALY AND ITS ASSOCIATED ANOMALIES

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ABSTRACT

Introduction: Anencephaly accounts for one of the most common birth defects & is associated with a high mortality & morbidity. The objective was to determine frequency of anencephaly and its associated anomalies, risk factors, clinical presentations & assess maternal awareness on folic acid supplementation and its preventive role in occurrence of anencephaly.

Materials and Methods: The study includes 60 anencephalic fetuses (23 males & 37 females) of 20-30 weeks. The fetuses were examined for external abnormalities & dissected. Measurements of crown heel, crown rump, head circumference, foot length, & weight were taken. All the major organs were weighed & data recorded. The samples were fixed with 10% buffered formaldehyde. The internal anomalies were noted.

Results: The number of deliveries was 57429 deliveries. 23(38.4%) were males & 37(61.6%) were females. The frequency of anencephaly in 2014 was 15134/9 (0.06%), in 2015 16361/21 (0.13%). Weight of 40% were <500gms, 35% were between 500-1000gms & 25% <1000. Maternal age in 83.4% were 21-35 years, in <20 years & >40 years were 1.6% & in 36-40 years were 13.4%. Associated anomalies were present in 42 (70%) fetuses. Out of 42 fetuses, those who had associated anomalies were 17 (40.4%) males and 25 (59.6%) were females. All the fetuses had Acrania (100%) & 19 (45.3%) fetuses had spina bifida; there were no anomalies found in reproductive system.

Conclusion: 4mg of folic acid should be taken before 6 months of conceiving & during the entire pregnancy. In order to prevent the anencephaly and associated anomalies certain maternal risk factors should be avoided as prescribed by gynecologist.

KEY WORDS: Anencephaly, Crown Heel, Crown Rump, Head Circumference, Foot Length, Weight.

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INTRODUCTION

Anencephaly is an embryological malformation of central nervous system; invariably lethal, characterized by the absence of brain & cranial chambers & other defect of the cranial structure. Till now no treatment is available for Anencephaly but can be diagnosed by ultrasound scanning [1]. The neural defects are generally distinguished through the failure of neural tube to close during the early stage development of embryonic development [2]. It is a very rare congenital disorder in which malformation of brain occur during embryonic development at third post-fertilization week & characterized by a hyper-retroflexion of cephalic pole & may lead to spontaneous abortion, stillbirth & death in

early infancy or lifetime disability [3,4]. Much of the brain is underdeveloped or absent altogether. Typically, the brainstem & portion of midbrain is present; the portion of brain that is present controls only basic breathing, cardiovascular, suckling & elimination reflexes; therefore the infants never gain consciousness or feel pain [5]. This study was designed to estimate frequency of anencephaly & its associated anomalies of fetal autopsies in Victoria Hospital and Vani Vilas Hospital, Bangalore, Karnataka, India.

MATERIALS AND METHODS

The present study of congenital anomalies in anencephalic fetuses was done at Bangalore Medical College & Research Institute, Bangalore. Study was conducted over a time period of 3 years from August 2014 to June 2017. The total number of deliveries during this period was 57429. Of these deliveries the number of Fetuses with Anencephaly was 60 (23 males & 37 females); the age of fetuses were 20 to 30 weeks. Consent for autopsy was requested compassionately, respectfully & fully informed. The dissecting instruments required for fetal autopsy are small scissors & forceps & scalpels. Measurements of crown heel, crown rump, head circumference, foot length, & weight are taken for comparison with standard chart. Dissection was performed by positioning of the body. The body was kept in supine position with a wooden block under the shoulder to keep the neck in extended position.

A curved incision was made bilaterally from the acromion process through the medial border of shoulder joint to mid-axillary line laterally, this continued to the iliac crest over the inguinal ligament to meet pubic symphysis. The skin with the superficial tissue flap was reflected up the root of the neck, then to the inferior margin of mandible bilaterally, taking care not to injure the neck structures and rectus sheath. This way, whole of the front of the neck, chest and abdomen was exposed [6].

Opening the abdominal cavity: A paramedian incision was made on rectus, near pubic symphysis, up to xiphoid process.

Opening thoracic cavity: Sternum was removed by cutting at costochondral junction and then separating sternoclavicular joint. All the major organs (lungs, heart, spleen, kidney, adrenal gland) were weighed & data recorded in autopsy protocol along with expected values. Photographs were taken. The samples were fixed with 10% buffered formaldehyde approximately to 24 hours. All the procedures were approved by research board & ethical committee of BMCRI, Bangalore. The associated abnormalities were grouped according to main organ system to which they belonged. The observed data were subjected to Fisher's exact tests and the significance was determined with $P< 0.05$ for statistical significance. The statistical tests were performed using software SPSS 15 (Statistical Package for Social Sciences)

RESULTS

The analysis of data revealed that incidence of anencephaly in Victoria and Vani Vilas hospital was 1.04 in 1000 births, over a period of 3 years from August 2014 to June 2017. The total number of deliveries during this period was 57429 deliveries. In the present study 23(38.4%) were males & 37(61.6%) were females. The majority were females in the present study. The frequency of anencephaly in 2014 was 15134/9 (0.06%) while in 2015, it was 16361/21 (0.13%) (Table 1).

Table 1: No of deliveries from August 2014 to June 2017.

Year	Number of deliveries	Anencephaly	Percent
Aug-Dec 2014	15134	9	0.06
Jan-Dec 2015	16361	21	0.13
Jan -Dec2016	17289	22	0.13
Jan -June 2017	8645	8	0.09
Total	57429	60	

Gestational age of fetuses was 20 to 30 weeks, in which 20 weeks was 26.6%, 21weeks cases were 15% & 30 weeks were 10% (Table 2).

Of total 60 fetuses the weight of 40% were <500gms, 35% were between 500-1000gms & 25% <1000.

Classification of mothers according to age showed that maternal age in 83.4% were 21-35 years, in <20 years & >40 years were 1.6% & in 36-40 years were 13.4%. The mean of maternal age is 24.4 ±4.

When mothers were classified according to their

level of education, it was noticed that 91.6% were found to be <high school (illiteracy), 5% were high school & >high school were 3.4%. The study showed that majority of mothers were 76.7% unemployed (housewives) & 46% of the respondents were found to have regular visits to antenatal care centers.

Associated anomalies were present in 42 (70%) fetuses (Table 3). Out of 42 fetuses, those who had associated anomalies were 17 (40.4%) males and 25 (59.6%) were females. All the fetuses had Acrania (100%) & 19 (45.3%) fetuses had spina bifida; there were no anomalies found in reproductive system.

Table 2: Age of fetuses.

Age of fetuses (weeks)	Frequency of Age	Percent
20	16	26.6
21	9	15
22	6	10
23	4	6.7
24	7	11.6
25	3	5
26	1	1.6
27	4	6.7
28	1	1.6
29	3	5
30	6	10
Total	60	100

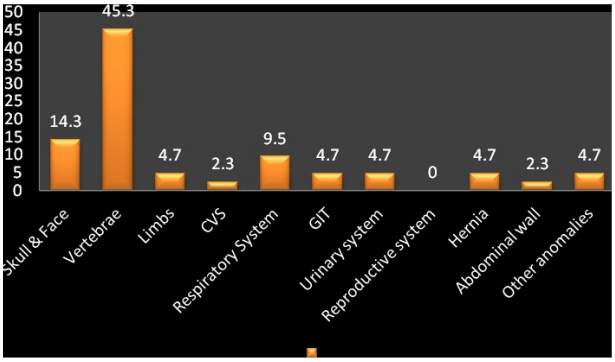
Table 3: Percentage distribution of associated anomalies (n=42).

System	Anomalies Present			
	M	F	Total	Percentage (%)
Skull & Face	2	4	6	14.3
Vertebrae	10	9	19	45.3
Limbs	1	1	2	4.7
CVS	1	0	1	2.3
Respiratory System	0	4	4	9.5
GIT	0	2	2	4.7
Urinary system	1	1	2	4.7
Reproductive system	0	0	0	0
Hernia	2	0	2	4.7
Abdominal wall	0	1	1	2.3
Other anomalies	0	2	2	4.7
Total	17	25	42	100

M- Male, F-Female

Male fetuses had 40.4% of overall associated anomalies & female fetuses 59.6% of associated anomalies. The female anencephalic fetuses were found to have more number of associated anomalies. (Chart 1)

Chart 1: Percentage of system wise Associated anomalies.



System wise anomalies: The 6 cases (14.3%) had Facial defects (4.7% were males & 9.5% were females). The most common defect was cleft palate & cleft lip (9.5% cases), it was found to be statistically significant. In 19 (45.3%) cases spina bifida was very common, in 1 (2.3%) along with spina bifida cleft lip & palate was also found. The spina bifida upto sacral region was seen in 4.7% of cases & it was found to be statistically significant (Table 4).

Table 4: Classification of spina bifida.

Spinabifida	No of cases			Percent
	M	F	Total	
Cervical	2	3	5	11.9
Thoracolumbar	5	1	6	14.3
Lumbosacral	3	3	6	14.3
Sacral	0	2*	2	4.7

*statistically significant

Anomalies in limbs: 1 case (2.3%) had Amelia of right lower limb, congenital dislocation of right elbow joint, malrotation of gut towards left, which was found to be statistically significant (Fig 1).

CVS: in 1 male fetus (2.3%), there was a single ventricle (mono ventricle) on the left side, with two outlets (aorta & pulmonary trunk), which was found to be statistically significant (Fig 2 a,b).

Lungs: in 1 case (2.3%), left lung had 2 fissures & 3 lobes, in 1 (2.3%) right lung had 3 fissures with 4 lobes. In 2 cases (4.7%) both lungs had single oblique fissure with 2 lobes (Fig 3). In 1 case (2.3%), there was malrotation of gut towards left side & also absence of pancreas (Fig 4).

Urinary system: 1 case (2.3%) had unilateral

biureter on left side (Fig 5) & 1 case (2.3%) had horse shoe shaped kidney; both are statistically significant (Fig 6 a,b).

There were no anomalies found in reproductive system. In 2 male cases (4.7%), there was umbilical hernia (Fig 7). Diaphragmatic hernia was not seen. Amniotic band syndrome (fig 8), encephalocele (fig 9) & omphalocele (fig 10) were seen in 2.3% of cases.

Fig. 1: Amelia,cleft lip,umbilical hernia.

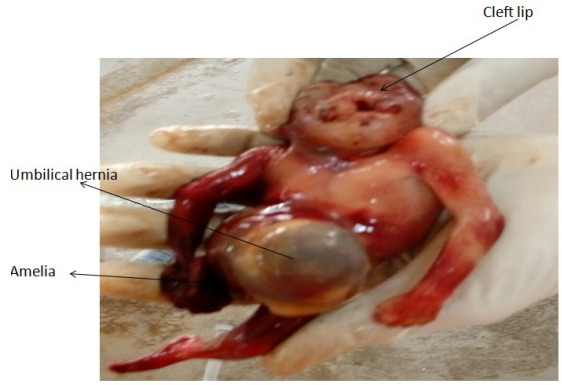


Fig. 2: double outlet, monoventricle.

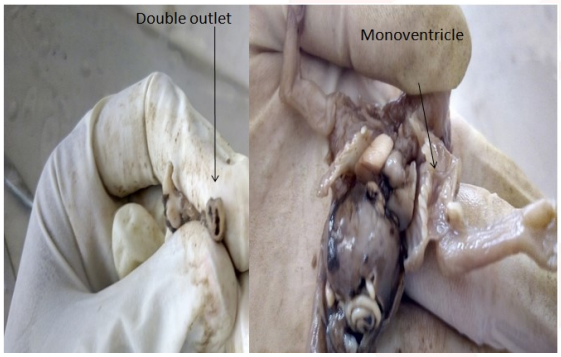


Fig. 3: bilobed right lung.

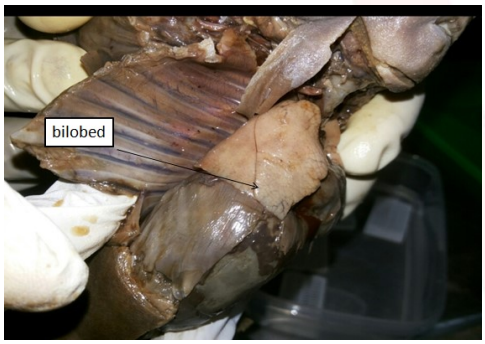


Fig. 4: absence of pancreas.

Fig. 5: biureter left side.

Fig. 6 a,b: Horse shoe shaped kidney.

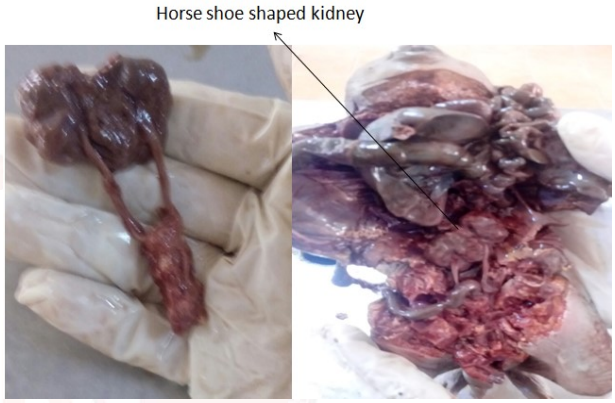


Fig. 7: Umbilical hernia.

Fig. 8: Amniotic band syndrome.

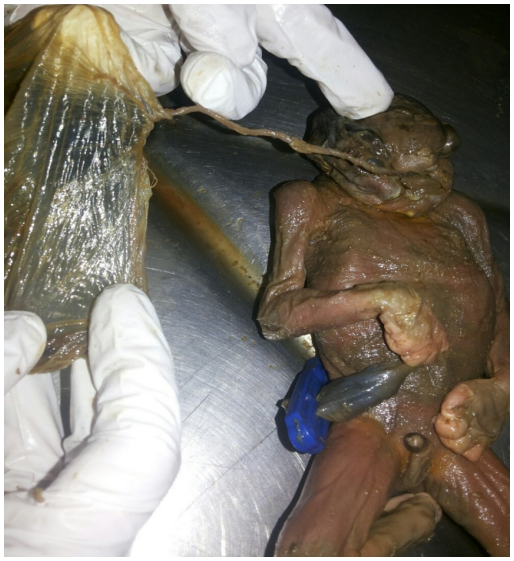


Fig. 9: Encepholocele.

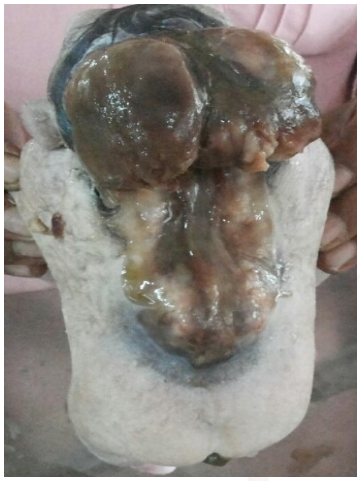
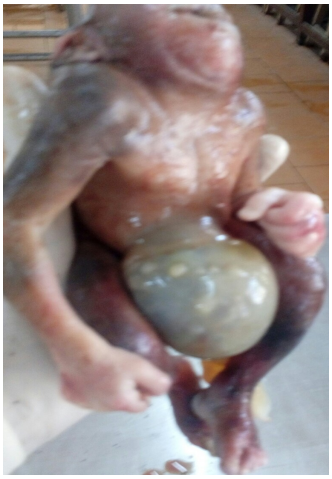


Fig. 10: Omphalocele.



DISCUSSION

Table 5: Comparison of associated anomalies with Anencephaly.

System	Tan et al (1984) [12]	Vare et al (1971) [13]	Neilson et al (2006) [14]	Pandurang et al (2012) [15]	Singh et al (2015) [16]	Present study (%)
Total incidences	9.4	-	43	73	58	70
Head & Neck	NAD	7.5	14	2.5	13	15
CVS	3	7.5	4.75	14.5	13	2.3
Respiratory system	3	NAD	NAD	2.5	17.3	9.5
GIT	29	32	NAD	14.5	13	4.7
Renal	3	27	12	NAD	11.5	4.7
Musculoskeleton	20	14.5	16.5	14.5	10.1	2.3
Genital	NAD	5	NAD	12	1.4	NAD
Diaphragmatic hernia	NAD	5	2.3	1	5.7	NAD
Umbilical hernia	1	NAD	NAD	NAD	NAD	4.7

Incidence of anencephaly is reported to be 1:1000 to 1:20000. In the present study the incidence of anencephaly in Victoria and Vani Vilas hospital was 1.04 in 1000 births.

In the present study 23 (38.4%) were males & 37(61.6%) were females. The majority were females in the present study, the literature also showed that anencephaly is more common in females [7,8].

The rate of anencephaly could be higher in underdeveloped countries due to possible misdiagnosis, most probably due to poor diet; improper maternal health care, inappropriate treatment & environmental factors also contribute to it [9]. The ratio of anencephaly is higher in Iranian population than compared to European population. It has been calculated that in Iranian population 13.1/10,000 newborns had anencephaly whose mothers aged >35 years & consanguineous marriage contributes 36 % to anencephaly which was found to be similar in present study [10].

Congenital anomalies vary in different studies. The study of malformation is greatly helpful in genetic counseling & prenatal diagnosis in successive pregnancies [11]. Anomalies were found in 42 cases (70%) in the present study & it is compared with the other studies.

In present study, out of 42 cases, 25 females (41.7%) & 17 males (28.3%) showed associated anomalies; majority of anomalies were in female anencephalic fetuses which was similar to some studies. Cleft lip & palate were the most common defect (14.3%) & 19 (45.3%) had spina bifida. There were no reproductive system anomalies in the present study.

Most of NTDs are associated with omphalocele, diaphragmatic hernia & cleft lip. It was first described in 1981 by Czeizel & named “schisis association (SA)” [17]. Polydactyly, conotruncal septation defects, limb abnormalities &

extremely rarely, oligodactyly were minor manifestations of SA [17,18].

Another common association is amniotic band syndrome (ABS). ABS is a group of congenital anomalies that includes the extremities, cranio facial, vertebral regions & body wall defects [19,20].

Its pathogenesis includes germ disc disruption, genetic disruption, vascular disruption & amniotic disruption [21]. We determined one case of ABS, amelia, omphalocele & encephalocele in the present study.

Most of the organs in the present study were anatomically normal when it was compared with the control group & some studies [22].

Literature showed that anencephalic fetuses were successful donors of hearts & kidneys for transplantation [23].

The intake of 4mg per day of folic acid intake is recommended in mothers with history of neural tube defect [23,24].

CONCLUSION

Present study concludes that since there is a chance of occurrence of anencephaly, 4mg of folic acid should be taken before 6 months of conceiving & entire pregnancy.

Maternal factors, like age, alcohol consumption, smoking, febrile illness, consumption of certain drugs and chemicals, family history, season of birth, environmental factors, are risk factors of conceiving an anencephalic child and its associated anomalies; hence these factors should be avoided

Consanguineous marriages could be avoided and they could be told about the risks of the same in successive generations too.

All pregnant mothers have to go in for triple marker tests; that is, beta HCG, alphafetoprotein and estradiol.

Amniocentesis could be made compulsory for mothers with a history of an anencephalic child. The mother has to be counseled regarding the risks of another such fetus. The family has to be told about pedigree charts, incidence and occurrence of anencephaly in the population.

They should be told about the importance of consuming folic acid before and during pregnancy. Majority of organs were normal anatomically & histologically, hence anencephalic fetal organs can be transplanted.

Conflicts of Interests: None

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