Prevalence of anencephaly associated anomalies

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Abstract

Introduction: Defective closure of rostral pore of neural tube leads to anencephaly. Folic acid deficiency is most common cause anencephaly. In most of the cases of Anencephaly is associated with other systemic anomalies.
Objectives: Aim of present study was to find out the incidence of anencephaly associated systemic anomalies and their correlation with maternal age, sex of the foetus.
Materials and Methods: 32 anencephaly foetuses obtained from the Department of obstetrics and gynaecology Navodaya Medical College. The period of study was from January 2013 to December 2016.
Results: Out of 32 cases, 25(78.12%) cases showed presence of systemic anomalies associated with anencephaly, 53.12% of the cases were observed in primigravida. Most common associated anomaly was spinabifida 28.12% followed by gastrointestinal anomalies 6.25%.
Conclusion: Incidence of anencephaly associated anomalies is not uncommon; hence gross systemic examination of the anencephalic abortus is required to find out incidence of anencephaly associated anomalies.

Keywords: Anencephaly, Craniospinal rachinoschisis, Exomphalos, Spina bifida.

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Introduction

Anencephaly is a neural tube defect which is due to the defective closure of cranial neuropore. Closure of the cranial neuropore takes place between the third and fourth week of gestation at 18-20 somite stage.[1] Anencephaly is most common congenital malformation during foetal development. The prevalence of anencephaly in India is 1 to 5 per 1000 live births.[2]

Present study was undertaken to determine the prevalence of anencephaly among congenital malformations. Organs from infants with anencephaly can be used for organ transplantation. David A et al have discussed anencephaly infant and their use in organ transplant, but most frequently anencephaly is associated with other anomalies. In present study we have correlated anencephaly with associated systemic anomalies, maternal age, birth order and sex of the foetus. Our study will be helpful in selecting anencephaly infant for organ transplantation.[3]

The Anencephaly shows a heterogeneous aetiology, ranging from environmental to genetic causes. Genetic disorders related with folic acid metabolism, glucose metabolism and inositol metabolism are associated with neural tube defects.[4]

Aetiology of Meroanencephaly, holoanencephaly, craniorachischisis and spinabifida was described by multisite closure model of neural tube of Van Allen et al. Anencephaly is commonly associated with other systemic anomalies; only few studies are available on anencephaly associated disorders in India.[5]

Materials and Methods

32 foetuses with neural tube defect were obtained from department of obstetrics and gynaecology. This study was done for three years from January 2013 to February 2016.

During this period, the foetuses which were still born with neural defect were collected after getting the consent of the parents. Foetuses died or terminated due to other causes were excluded from the study. They were embalmed and stored in 10% formalin solution.

The foetuses were observed in detail externally for the sex, type of neural defect and other associated anomalies and the findings were documented. The approximate crown rump length was measured and the probable gestational age of the foetuses was calculated. The foetuses were dissected to find other internal anomalies. The statistical analysis was performed by using frequency and percentage.

Results

Table 1: Incidence of anencephaly and its correlation with maternal age, parity, gestational age and sex of the foetus.

Age of mother	Parity		Gestational age	Sex of foetus	
20 years to 30 years	Primigravida	17 (53.12%)	20 weeks to full term	Male	7 (21.87%)
	Gravid two	11 (34.37%)		Female	23 (71.87%)
	Gravid three	3 (9.37%)		Ambiguous genitalia	2(6.23%)
	Gravid four	1 (3.12%)			

Table 2: Number of cases of anencephaly associated systemic anomalies

S. No	System	Associated anomalies	No of cases
1	Gastrointestinal system	Gastroschisis	01 (3.12%)
		Exomphalos	01(3.12%)
2	Respiratory system	Cyst in lung	01(3.12%)
3	Musculoskeletal system	Achondroplasia	01(3.12%)
		Absence vertebral column	01(3.12%)
4	Cardiovascular system	Single umbilical artery	01(3.12%)
5	Nervous system	Meroanencephalic	04(12.5%)
		Holoanencephalic	03 (9.37%)
		Spinabifida	09(28.12%)
		Craniospinal rachinoschisis	03 (9.37%)
		Myelocele	05(15.62%)
6	Head face neck	Cleft palate	01(3.12%)

Fig. 1: Encephalocele

Fig. 2: Craniospinal rachischisis

Fig. 3: Gastroschisis

Discussion

The prevalence of anencephaly shows great variation in time and space. Among the 194 World Health Organization member states, the percent reporting within each region is as follows: African (8/47;17%), Eastern Mediterranean (12/21;57%), European (26/53;49%), Americas (15/35;43%), South-East Asian (4/11;36%) and Western Pacific (9/27;33%).[6]

According to Mahadevan et al the prevalence of anencephaly in India is 1.8 to 7 per 1000 live births.[7] In present study maternal age studied was between 20 years to 30 years as we have not observed anencephaly foetus with maternal age above 30 years. In a study by Isabela, et al there was no association between maternal age and anencephaly.[8] A study by Dwight et al there was declining risk in young mother and older mothers, they observed a 'U' shape pattern while correlating anencephaly and maternal age.[9] Vieira et al observed higher risk of spina bifida than anencephaly in younger and older mothers. Thus more detailed studies are required to correlate maternal age and anencephaly.[10]

In present study anencephaly associated disorders were more common in primigravida (53.12%). In a study by Vare et al Anencephaly were observed more common in primigravida.[11]

Prevalence of anencephaly in female foetus (71.87%) was more than in male. Isabela et al observed 64% of female anencephalic foetuses in their study (y) The female preponderance also reported by Jaquier et al[12] and James et al,[13] but study by Obeidi et al did not observed female preponderance.[14]

Anencephaly associated anomalies ranges from 9.4 to 43%.[11, 16-18, 20] In present study it was higher (78.12%) than previous studies but similar to study by C. Panduranga et al 73%.[15]

The gastrointestinal anomalies associated with the anencephaly in present study we have observed one gastroschisis and one omphalocele (6.25%). In previous studies Golipure et al 5.3%[16] and David T et al 5.7%[17] gastrointestinal anomalies were similar to present study, but Vare et al mentioned (32%)[11] Gastrointestinal anomalies.

Respiratory anomalies in present study and study by Tan et al[18] were same (3%), but majority of previous studies have not mentioned about respiratory anomalies associated with anencephaly.

Musculoskeletal disorders in present study we have observed two cases (6.25%) achondroplasia and absence of vertebral column, but Vare et al mentioned 27%[11] and according to David T et al 16.3%.[17]

Eslavat et al[19] mentioned 33.3% cardiovascular disorders associated with neural tube defects, in present study 3.12%, Tan et al 3%[18] and Nielson et al 4.75%[20] have mentioned observations similar to present study.

Central nervous system disorders like spinabifida (28.12%), Craniospinal rachinoschisis (9.37%) and Myelocele (15.62%) were observed in present study, in previous studies only C. Panduranga et al mentioned Central nervous system disorders spinabifida (26.82%), Craniospinal rachinoschisis (12.19%) were close to present study.

3.12% of cleft palate was present in present study; previous studies mentioned similar observation Golipure et al 3.5%[16] and David et al 3.5%,[17] except Nielsen et al mentioned 14%[20] of head neck disorders.

Disorders like Diaphragmatic hernia and Genital defect were mentioned by very few previous studies; Nielsen et al 2.3%,[20] David et al 2.7%[17] and Vare et al 5%,[11] in present study we have not seen these disorders.

Conclusion

Anencephaly is common in foetus of primigravid mother and most common in female foetus. Central nervous system anomalies like spinabifida (28.12%), Craniospinal rachinoschisis (9.37%) and Myelocele (5%) are commonly associated with anencephaly. Ultrasound screening in first trimester can diagnose Anencephaly and other associated systemic disorders, but macroscopic systemic examination of the abortus is required, as in most cases anencephaly is also associated with other systemic anomalies. Present study provide information of prevalence of anencephaly associated disorders, which will help in counselling of eligible couple and for management in reducing incidence of anencephaly associated disorders.

Importance of present study – Organs are required in organ transplant. Anencephalic foetuses survival is not possible, hence organs of anencephalic foetuses can be used for organ transplantation, but anencephaly is associated with other systemic anomalies. Our study will provide data of anencephaly associated disorders for counselling of eligible couple, in use of anencephalic foetuses in organ transplantation and for management in reducing incidence of anencephaly associated disorders.

Limitations of present study – We have not studied aetiology of anencephaly. We have

studied only anencephaly foetuses from hospital attached to our medical college. Duration of present study was 3years.

Conflict of Interest

No financial and other conflicting interests.

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