Research Article

Congenital malformations in still born fetuses

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Abstract

Introduction: The study of congenital malformations in fetuses is essential to reduce not only the incidence of still born fetuses but also infant mortality. The birth of a child with congenital malformations has a very traumatic outcome on the family. The study of congenital malformations on live born neonates cannot provide a proper outlook. The present study emphasized the importance of thorough study on the still born fetuses. 100 still born fetuses were dissected and examined to record the incidence of congenital anomalies in major systems. Congenital malformations were seen in 36% of cases. Central nervous system was the commonest system involved followed by the musculoskeletal system. The above findings were discussed in comparison with available literature.
Keywords: Anencephaly, Fetuses, Polycystic kidney, Spina bifida.

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INTRODUCTION

Congenital malformations are a major cause of mortality and morbidity in developing as well as developed countries. With the advent of new antibiotics and vaccines the infant mortality due infectious causes has decreased. This has led to emphasis on the fetal loss due to congenital malformations. The worldwide incidence of congenital disorder is estimated at 3-7%, but actual numbers vary widely due to under reporting of cases in developing countries[1]. The presence of congenital abnormalities in a baby has an emotional effect on only on the mother but also on the family[2]. A congenital anomaly is defined in terms of physical structure as a malformation, an abnormality of physical structure or form usually found at birth or during the first few weeks of life[3]. The congenital malformations can be identified prenatally by ultra-sound and maternal serum analysis and by autopsy after the fetal death. Majority of studies have concentrated on the congenital malformations in live born neonates but very few studies have done it in fetus. There is a marked difference in the percentage of different congenital malformations in newborns and fetuses. Fetuses afflicted with major malformations result in spontaneous or induced abortions. Therefore a large number of malformations do not get counted in studies done at birth[4]. The present study was undertaken to find the percentage of congenital malformations in still born fetuses. This would help to identify the cause of specific disorder and correlation of malformations to each other.

MATERIAL AND METHODS

100 fetuses, whose relatives gave consent were included in the study. The fetuses were still born fetuses obtained from obstetrics department. All fetuses above 20 weeks were included in the study. The placenta and umbilical cord were examined for any gross abnormality. Prior to autopsy a thorough external examination of the fetus was done. Crown rump length was considered as an indicator of the gestational age of the fetus. The autopsies were performed as per guidelines provided by fetal autopsy protocol[5,6]. A longitudinal incision was taken from the suprasternal notch up to the pubic symphisis, followed by a horizontal incision to retract the skin flap. The thoracic and abdominal cavities were opened and any deviation from the normal anatomy was photographed and noted.

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The congenital malformations were classified by organ system according to the 10th version of the World Health Organization International Classification of Diseases (ICD-10).

OBSERVATION AND RESULTS

The 100 still born fetuses were dissected during the period of 18 months. Obtaining the consent was the main hinderence which compelled us to restrict the sample size. Of the total cases, 54 were males and 46 were females. Congenital anomalies were obtained in 36 cases, of which 17 were males and 19 females.

Table 1: Classification of congenital anomalies according to the major systems

System	Malformation	No of cases
Central nervous system	Anencephaly	6
	Spina bifida	2
	Anencephaly + spina bifida	3
	Kyphosis/ scoliosis	2
Genitourinary system	Polycystic kidney	4
	Agenesis of kidney	0
	Horseshoe kidney	1
	Undifferentiated gonads	1
Musculoskeletal system	Diaphragmatic hernia	4
	Cleft lip/ cleft palate	3
	Club foot	3
Gastrointestinal system	Omphalocele	2
	Intestinal malformations (malrotation)	2
	Absence of organ	0
Cardiovascular system	Atrial septal defect	2
	Single ventricle	1

On classification of the congenital anomalies (Table1), central nervous system (CNS) was the commonest, with 36% of all the anomalies. Anencephaly was the commonest of the CNS anomalies at 46%. Anencephaly was common in female fetuses. Spina bifida was the second commonest CNS anomaly. Two cases had anencephaly associated with spina bifida.

Genitourinary defects were seen in 17% of cases with polycystic kidneys being the commonest. One case of horseshoe kidney and one case of undifferentiated gonads were found. Diaphragmatic hernia, cleft palate, cleft lip and club foot which were included in musculoskeletal defects. They were found in 28% of cases. But two cases of cleft palate were associated with anencephaly. Diaphragmatic hernia was detected in 40% of the musculoskeletal defects. Malrotation of the intestine was found in 2 cases (6%) and omphalocele in 2 cases (6%). Absence of any organ was not found in any case. Malformations of the cardiovascular system were found in 3 cases (8%). Atrial septal defect was found in 2 cases and single ventricle in one case.

DISCUSSION

During the most sensitive period of embryogenesis i.e. the 3rd - 8th weeks of gestation factors like genetic, environmental, teratogenic and infectious agents play important role for the origin of malformations[7,8]. The congenital malformation accounts for the leading cause of neonatal deaths and stillbirths. However, detailed information about the incidence, type, anatomical parts or organs involved in congenital malformation are not available. The incidence of congenital malformations in the present study was 36%. Studies done by Mohan *et al*. (2004)[9] in north India and Sunethri Padma[10] (2011) in Hyderabad had incidence of 38% and 27% respectively which was similar to present study. In another study by Puri *et al*., (2009)[11] the incidence of fetal anomalies was 63% and by K kapoor *et al* (2013)[4] it was 69% which was more than the present study. On comparing the sex ratio, we found the congenital malformations were slightly more in females. There were 19 female fetuses and 17 male fetuses because of which the study failed to reach a statistically significant consensus. Deviations in sex ratio have been reported by many studies but a significant outcome has not been reported. Some studies have found the incidence to be more in males[12] but many studies have reported almost same incidence, which was similar to the present study. The significance of sex predominance is important as the dominance of a particular malformation helps to predict the occurrence and outcome. In the present study, significant sex predominance was found in only anencephaly which was common in females. All the previous studies have found anencephaly to be common in females[13,14]. The most common congenital malformation in present study was of the central nervous system. Anencephaly was the commonest followed by spina bifida. Though some studies have reported musculoskeletal[15,16] and gastrointestinal system[17] to be commonest, they were all conducted on live born babies. Studies conducted on still born fetuses have found central nervous system anomalies to be commonest[4,10]. Musculoskeletal system was the second commonest anomaly in the present study. Various studies have reported the incidence of musculoskeletal anomalies to be between 10% to 19% [4,18,19]. In the present study the incidence was 28%, this was because the present study included clubbed foot in the results. Study conducted by K kapoor[4] which had included similar malformations in the musculoskeletal system reported incidence of 21%. Urogenital malformations incidence varied from 3.8% by Grover (2000)[2] to 26.2% by Dutta (2010)[16]. In the present study the incidence was 17% with polycystic kidneys as the commonest. Renal agenesis was not found in any case. Cardiovascular anomalies was the rarest in the preset study, as found by previous authors[14,20].

CONCLUSION

The present study was carried out in a tertiary care hospital and the incidence of congenital malformation at 36% signifies the importance on recording the findings in still born fetuses. It is also of significance in future planning and counselling to affected families. The availability of genetic services should be emphasized so as to find the cause of the malformations. Even though the prenatal ultra sonogram reasonably predicts the malformations, a thorough study of the fetus is essential to look for additional malformations.

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