

RESEARCH ARTICLE

PATHOLOGY

PATTERN OF DISTRIBUTION OF CONGENITAL ANOMALIES IN STILLBORN: A HOSPITAL BASED PROSPECTIVE STUDY

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ABSTRACT

Background: Congenital anomalies (CAs) are one of the most common causes of disability in developed and developing countries. **Aims:** To determine frequency, pattern of distribution of CAs in still born with a focus on sex ratio. **Methods:** Maternal history, associated risk factors, socio economic status, sex and type of CAs in babies were recorded. Diagnosis of CAs was based on findings of autopsies conducted. **Results:** Autopsy was conducted on 102 still born babies, of which 28 were found to have CAs. The occurrence of CAs was more in males than females. The pattern CAs included CNS, Digestive system, urinary system and circulatory system. In CNS group, Anencephaly was the most common malformation followed by Spinabifida. **Conclusion:** Frequency of CAs were more common in males than females, with CNS anomalies being the most common and also these anomalies were more common in gestational age of 29-32 weeks.



KEY WORDS

Congenital anomalies; autopsy; Anencephaly; Spinabifida; still born.

INTRODUCTION

Congenital anomalies (CAs) are important causes of mortality in developed and developing countries¹. The etiology of most congenital anomalies remains unknown, although there are few well established and avoidable external risk factors². Since the 1960s, a general surveillance has been carried out monitoring the appearance of congenital anomalies in various populations around the world³. Worldwide surveys have shown that the birth prevalence of congenital anomalies varies greatly from country to country⁴. These variations may be explained by social, racial, ecological, and economical influences^{5, 6}. Congenital anomalies, a leading cause of fetal loss, contribute significantly to preterm birth and childhood and adult morbidity⁷.

To decrease the incidence of various congenital anomalies and their prevalence in every society, it is important that the distribution and prevalence of congenital anomalies are identified for every country, and even for every region⁵. The objective of the present study was to determine the sex ratio, incidence and pattern of congenital anomalies in still born babies over a period of 30 months at Gandhi hospital and to contribute to the efforts in their screening, diagnosis and treatment. To the best of our knowledge, similar study was not done at Gandhi hospital till date.

MATERIALS & METHODS

This was a descriptive, cross-sectional study of stillborn babies delivered at Gandhi Hospital during a 30 months period, July 2007 to December 2009. Gandhi Hospital serves as a referral centre for obstetrics and gynecology and prenatal intensive care for all other hospitals and clinics in and around Hyderabad.

All singleton pregnancies with stillborn babies delivered in this hospital during the above mentioned period were included in the study. Variables like extensive maternal history age and sex of the fetus were recorded. Autopsy was done on fetuses for which consent was obtained. Prior to autopsy a thorough external examination of the fetus was done and also specific measurements and norms for particular gestational age were recorded. Femur length was considered as an indicator of the gestational age of the fetus. Autopsy was performed according to potter's procedure. Relevant tissues were processed for histopathological examination. These findings were correlated with antenatal scan report.

The types of birth defects were classified according to International statistical classification of diseases and related health problems, 10th revision (ICD-10) (Table 1). The data were analyzed by SPSS, version 10 software. Statistical analysis performed by the chi-square test, and mean and percentage age values was calculated. $P < 0.05$ was considered to be statistically significant.

The study was approved by the Institutional Ethics Committee of Nandamuri Taraka Ramarao University of Health Sciences (NTRUHS). Written consents were taken from all the participants of this study.

RESULTS

During the period of 30 months, the total number of births was 17953 out of which 1100 were still born. But consent for autopsy could be obtained only for 102 fetuses. Autopsy was conducted on those fetuses and findings were recorded according to the preformed proforma. Congenital anomalies were detected in 28 fetuses. The anomalies were classified

according to ICD -10 (Tables 1). More than one anomaly was found in two fetuses.

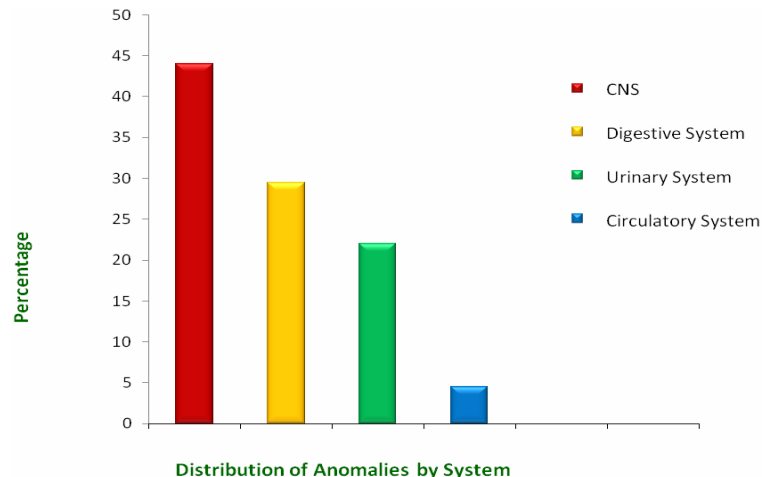
Table 1
congenital Anomalies by system according to ICD-10

S.No.	Malformation/System	Number of cases
1.	Central Nervous System	
	• Anencephaly alone	6
	• Anencephaly + Spinabifida	1
	• Spina Bifida alone	5
2.	Digestive system	
	• Omphalocele	4
	• Duodenal atresia+Omphalomesentric cyst	1
	• Astomia	1
	• Agnathia	1
	• Aglossia+Agnathia	1
3.	Urinary System	
	• Horse shoe kidney	3
	• Polycystic Kidney	2
	• Congenital posterior urethral valve	1
	• Congenital Bladder obstruction	1
4.	Circulatory System	
	Dextrocardia	1

There were 28 cases (1.5/1000) of single or multiple congenital anomalies among 17953 live births that occurred during the year of July 2007 to December 2009. Out of 102 autopsies, congenital anomalies were found in 28 fetuses (27%). Percentage of anomalies according to system wise diagrammatically represented in Fig 1. The most common anomalies were that of

central nervous system [12, 44%], with Anencephaly being the commonest one (50%). In one fetus we found Anencephaly associated with Spinabifida which was the only anomaly found in rest of the fetus with central nervous system anomalies.

Fig 1
Percentage distribution of anomalies in various systems



Digestive system was the next commonest one (9, 29.5%) and in this category Omphalocele was found to be the frequent one (50%). Astomia, Agnathia, Aglossia occurred as single anomalies in one fetus each. There was one case where we found duodenal atresia along with Omphalomesenteric cyst.

Third commonest system was Urinary system (6, 22%) with Horse shoe Kidney being the commonest one. Polycystic Kidney was seen in two fetuses which were confirmed by histopathological examination and they were of infantile type. Grossly distended bladder was seen in two cases and with meticulous examination we could diagnose Congenital posterior urethral valve in one and Congenital Bladder Obstruction in other. These two fetuses also had megaureters and hydronephrosis. Present study in histopathology findings were

relevant and of diagnostic significance only in polycystic kidneys and hydronephrosis and rest of tissues were normal.

The least common group was that of circulatory system (1, 4.5%) where only one case was recorded with Dextrocardia.

Maternal age was in range of 19 years to 28 years with a mean of 23.5 years associated risk factors like consanguinity, previous history of abortion, previous child with congenital anomalies and low socio economic group were considered and observed that none of them were relevant low socio economic group.

The incidence of these congenital anomalies was found to be more in male fetus on comparison to the females ($p < 0.01$). The sex ratio was 1.7:1 (Table 2).

Table 2
Sex Wise Distribution of Congenital Anomalies

Sex	Cases	Percentage
Male	18	64%
Female	10	36%

Male : female - 1.7:1

Antenatal Ultrasonography was done for all the fetuses and this was done just before the delivery of the babies. The gestational age varied from 16 week to 36 weeks, however majority of the fetuses with anomalies were in 29 to 32 weeks of gestation (Table 3). Accurate

assessment of the fetal age was done based on femur length and also correlated with BPD, head circumference and abdominal circumference. Antenatal scan reports correlated with autopsy findings in 59% of the cases.

Table 3
Gestational age of fetus

Age (weeks)	Male	Female
16- 20	1	-
21-24	2	-
25-28	3	2
29-32	7	4
33-36	5	4

DISCUSSION

Most children who are born with major congenital anomalies and survive infancy are affected physically, mentally or socially and can be at increased risk of morbidity due to various health disorders^{8, 8, 9}. While the prevalence of congenital anomalies at birth in developed countries is reported to be between 3-5%, those reported in India before the 1990s were in general lower than this figure¹⁰. However, a study that investigated the number of all infants born in one year with a congenital anomaly at 10 university hospitals in India found a birth prevalence of congenital anomalies of 3.65%¹¹. Antepartum deaths have declined in advanced countries but it is still a major problem in developing countries especially south Asia¹².

In the present study the incidence of congenital anomalies was 0.7%. Most of the studies where incidence for CAs was quoted, the study group included new borne along with still born. Whereas the present study included only still borne making it one of the rarest studies especially at Gandhi Hospital. Bhat BV et al¹³ had some similar studies where the incidence of CAs in still borne was 8 % and 15.7% respectively. In comparison with these studies, the incidence in our study may be low possibly because the number of cases in our study group was less. Obtaining consent for autopsy was the major hindrance for screening more number of fetuses. Apart from this, inadequate genetic screening of the fetus to rule out chromosomal anomalies was another factor due to nonavailability of a genetic laboratory at our institute. Worldwide surveys have shown that the birth prevalence of CA varies greatly from 1.07% in Japan as high as 4.3% in Taiwan⁴.

In our study we focused on sex ratio in fetuses having CAs and we found that the occurrence was more amongst male babies than female babies (1.7:1). Similar ratios were reported in other studies done in India¹³. In other study authors have found that the rate of malformations in males is nearly twice as that of females^{14, 15}. However gender of the fetus did

not affect the prevalence of CAs and both genders were equally distributed in an another study⁴. Mild deviations in sex ratio have been reported for many CAs, but so far no satisfactory explanation for these deviations has been found¹⁶. The significance of sex predominance can be substantiated by the fact that when there is a dominance of one sex for a particular malformation, this information can help predict the likelihood of the malformations in a patient and influence diagnostic approach¹⁷.

The most common CAs in our study was that of central nervous system which constituted to 44% of the total anomalies with Anencephaly being the commonest one followed by Spinabifida. Similar finding were observed in another studies^{4, 18}. In contradiction to our findings musculoskeletal system anomalies were commonly found in few studies¹³. In another study authors reported that cardiovascular anomalies were common³.

An additional observation in our study was that majority of the fetuses with CAs were in the gestational age of 29-32 weeks, which was similar to the observations of Michels and Khaskheli^{12, 19}.

Maternal age is an important parameter in the birth of a congenitally malformed fetus²⁰. For this reason, the risk of birth of a congenitally malformed fetus in mothers who are older than 35 years of age needs to be examined more carefully. In this study, mothers are not older than 35 years age.

It is noteworthy that our series of autopsies come from a government hospital where most of the patients belong to low socioeconomic group. A low Socio-occupational status will often correlate with environmental exposure such as in door pollution or living close to industries². Hence environmental factors can be implicated to the incidence of CAs in our patients and also male predominance.



In summary, the most common congenital anomalies in Hyderabad in the last 2 years are nervous system anomalies followed by digestive, urinary and cardiovascular system. The low birth prevalence of congenital anomalies (1.5/1000) in this study may be a result of, a small study group inadequate genetic counseling to rule out consanguineous marriages, chromosomal anomalies and as well as personal and institutional characteristics of the current documentation system. Present study showed that high frequency of abnormalities found in between 25-28 weeks of the fetus males and females.

CONCLUSION

Congenital anomalies are one of the most important causes of fetal deaths and hence it

becomes mandatory to keep on account of incidence and prevalence in the society. In our study we found the incidence of CAs among still born at our institute to be 0.7% with male predominance. Also most of the fetuses were in the gestational age of 29-32 weeks. This study was an effort to explore the pattern and distribution of CAs prevailing in the population of Hyderabad with an emphasis on male preponderance. We strongly recommend the formation of new strategies and studies to investigate causes of increased male predominance in CAs and also a good functioning registry of birth defects and surveillance system along with genetic service network in India.

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