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Original Research Article

Prevalence, pattern and outcome of congenital malformations in a tertiary care centre in South India

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ABSTRACT

Background: Congenital malformations are of major concern as they are cosmetically unacceptable, often associated with significant functional abnormalities and may sometimes even be life-threatening. This study was done to explore the prevalence of structural congenital malformation among hospital newborns, both live and stillborn in a tertiary care center in Southern India.
Methods: This hospital-based prospective study involving all inborn neonates and still births was conducted for the period from January 2014 to December 2014. A total of 2276 newborn (2217 live births and 59 still births) were clinically examined for detection of gross congenital malformations and relevant investigations including karyotyping was done. Risk factors that had probable associations with birth defects were estimated by calculating the Odd's Ratio. Statistical analysis was done using Chi-Square test.
Results: The prevalence of congenital malformations was 12%. Major malformations accounted for 53.28% and minor malformations 46.71%. The commonest structural malformation involved cardiovascular system. 58% of neonates did not require life style medications whereas 21.17% required surgical intervention. 20.8% of the cases succumbed to death.
Conclusions: Autopsy of stillborn babies should be done to evaluate the cause and incidence of malformations. High risk mothers should be identified in the antenatal period to detect malformations early and plan management accordingly.

Keywords: Autopsy, Genetics, Malformations

INTRODUCTION

Birth defects are a major health concern and are a leading cause of neonatal and early childhood mortality. They represent permanent structural, functional and/or biochemical-molecular changes produced by intrinsic abnormality of development in a body during perinatal life. Centre for Disease Control and Prevention (CDC) USA, had reported an incidence of about 3% of all live births in the USA during 2004-2006.[1] It is the most important cause of under-five mortality in developing nations like India and account for about 61 to 69.9/1000 of all live births.[2] This high prevalence warrants the need to take immediate steps to tackle the problem on a war footing, more so when 70% of these defects are preventable.[2] India ranks second in the world with regard to reported occurrence of congenital anomalies in neonates and children.[2] This fact highlights the urgency and importance of documenting all congenital malformations occurring in neonates born in hospital

setting, so as to focus and develop appropriate preventive and remedial strategies. Quantifying birth defects in a population is a felt need as it helps in appropriate allocation of health budget to tackle and reduce perinatal, neonatal and infant mortality rates.

Hence this study was conducted to assess the prevalence and spectrum of congenital malformations occurring among institutional births in Mahatma Gandhi Medical College and Research Institute (MGMC and RI), Puducherry and possibly identify the probable risk factors associated with them.

METHODS

This cross-sectional study was carried out on all neonates (both live and stillborn) born in the Mahatma Gandhi Medical College and Research Institute, a tertiary health care delivery centre in Pondicherry, over a period of 12 months from Jan 2014 to Dec 2014. Approval was taken from the Institutional Human Ethics Committee before commencement of the study. The study was conducted on 2276 subjects (55 stillborns and 2221 live borns) delivered in the labour room under the Department of Obstetrics and Gynaecology of the institute. All new-borns (both live births and still births) delivered during the study period (January 2014 to December 2014) in the institute were included. Any new-born born outside but treated here was excluded from the study.

Written expressed consent was taken from the parents. A predesigned proforma was used to obtain information on the degree of consanguinity, maternal age, parity, mode of conception, maternal complications, family history of congenital malformation, bad obstetric history, maternal drug intake, alcohol consumption, birth weight of new-born to identify the possible risk factors leading to those malformations. The babies were thoroughly evaluated to identify the nature, severity of structural congenital malformations and to classify them as single or multiple malformation syndromes or associations. Appropriate investigations and treatment was provided to neonates who required them. Chromosomal analysis was done as per standard Hungerford method.

Statistical analysis was done by using Epi-Info V6. Percentage for qualitative data was done using mean and proportion, standard deviation, Chi square test, Student t test and Odd's ratio. A p value of <0.05 was considered statistically significant.

RESULTS

During this one-year study period, 2276 deliveries were conducted in our institute out of which 274 babies were malformed. Thus, the incidence of congenital malformations was 12% of all deliveries and 10.13% (225/2221) among live births and 89.09% (49/55) among still births (Table 1).

Table 1: Incidence of congenital malformations.

	Total deliveries	Malformations	Malformations (%)
Total deliveries	2276	274	12
Live births	2221	225	10.13
Still births	55	49	89

53.28% of the cases had major malformations while 46.71% had minor malformations (Table 2).

Table 2: Distribution of congenital malformations as major and minor malformations.

Types of congenital malformations	Number of cases	Percentage of cases (%)
Major	146	53.28
Minor	128	46.71

The male: female ratio of the babies born with congenital malformations was 2:1 (183 males and 91 females) (Table 3).

Table 3: Distribution of recognizable congenital malformations in males and females.

Gender	Number of cases with congenital malformations
Male	183 (66.79%)
Female	91 (33.21%)

53.2% of cases with malformations weighed less than 2500 gm (Table 4) and 31% of new-borns were born before 37 completed weeks (Table 5).

Table 4: Prevalence of congenital malformations based on birth weight.

Birth weight	Number of cases	Percentage of cases
<2500 gm	146	53.2
>2500 gm	128	46.7

Table 5: Prevalence of congenital malformations by gestational age.

Gestational age	Number of cases	Percentage of cases
Term	189	68.9
Preterm	85	31

Congenital malformations were noted with highest frequency (72.63%) in babies born of mothers aged between 26 and 30 years of age (Table 6).

Table 6: Correlation of maternal age with congenital malformations.

Maternal age (years)	Number of babies with congenital malformations
< 20	28 (10.22%)
20-25	7 (2.55%)
26-30	199 (72.63%)
>30	40 (14.60%)

The maternal risk factors identified were consanguinity in 204 babies (74%), bad obstetric history in 98 babies (35.7%) and maternal systemic illness in 52 babies (21%). We observed no significant risk associated with the various modes of conception or delivery, antenatal immunization status, IFA supplementation and exposure to teratogens in the occurrence of recognizable congenital malformations.

Table 7: Systemic distribution of recognizable major congenital malformations.

Systems involved	Number of cases	Percentage of cases (%)
Cardiovascular	77	52.7
CNS	37	24.6
Renal	13	8.9
Gastrointestinal	11	7.5
Respiratory	5	3.4
Genito-urinary	2	1.3
Musculoskeletal	1	0.6
Total major malformations	146	100

Among the major congenital malformations, a greater number of cases (77 cases or 52.7%) had cardiovascular involvement (Table 7).

Table 8: Systemic distribution of recognizable minor congenital malformations.

Systems involved	Number of cases	Percentage of cases
Cutaneous	48	37.5
Musculoskeletal	29	22.6
Genito-urinary	17	13.5
Ear	14	11.1
Eye	9	7.1
Gastrointestinal	7	5.6
Cardiovascular	4	3.1
Total minor malformations	128	100

Minor congenital malformations had mostly manifested as cutaneous malformations (48 cases or 37.5%) (Table 8). Among Cardiovascular malformations, acyanotic lesions (VSD, ASD and PDA) were mostly present. (Table 9) Most cardiovascular lesions were acyanotic lesions (68 cases or 88%) which presented with murmur found incidentally on clinical examination. Cyanotic heart diseases included transposition of great arteries (2 cases), tetralogy of Fallot (2 cases) which required intensive care and referral to higher centre for surgery after stabilising with prostaglandin when required.

Table 9: Distribution of malformations affecting major systems.

System involved	Types of malformations	No. of cases
Cardiovascular	VSD	38
	PDA	25
	ASD	5
	CoA with MAPCAS	1
	Hypertrophic cardiomyopathy	1
	Myxomatous mitral valve	1
	TGA	2
	Ebsteins anomaly	1
	Double outlet right ventricle	2
	Hypoplastic left heart syndrome	2
	Pulmonary stenosis	1
	TOF	2
Central nervous system	Anencephaly	22
	Hydrocephalus	2
	Sacral myelomeningocele	1
	Choroid plexus cyst	1
	Myelomeningocele	7
	Microcephaly	4
Genitourinary	Hypospadiasis	6
	Hymenal tag	5
	Ambiguous genitalia	2
	Imperforate anus	2
	Bucket handle anus	2
	HUN	6
	PUV	3
	Dysplastic right kidney	1
	Pelvicalcyiectasis	1
	Polycystic kidney disease	2
Gastrointestinal	Cleft Palate	5
	Cleft Lip + Cleft Palate	3
	Omphalocele	1
	C. Absence of Rectus Abdominis	1
	Prune Belly Syndrome	1
	Umbilical Hernia	7
Respiratory	Tracheo Esophageal Fistula	2
	Eventration Of Diaphragm	1
	Congenital Diaphragmatic Hernia	1
	Congenital Tracheal atresia	1

Table 10: Chromosomal anomalies observed in the study.

Types of anomalies	Chromosomes involved
Trisomy 21 with mosaic turners	47xx +21/46 x 0 + 21
Down's syndrome	trisomy 21
Multiple aberrant numerical metaphase/ polyploidy	
Down's syndrome	trisomy 21
Mosaic male Turners	46xy / 45x0
True hermaphrodite(intersex)	46xy / 46xx
Sirenomelia	46 x, t (x; 16) denovo

Anencephaly (22 cases) and Myelomeningocele were the commonest CNS malformations observed. 1 case of sacral myelomeningocele was successfully operated. Among genitourinary malformations, Hydroureteronephrosis was detected antenatally in 6 cases which was confirmed by postnatal ultrasound. Of the 2 neonates with ambiguous genitalia, one was diagnosed as Sirenomelia (46 X, t (X; 16) denovo) and the other as mosaic male turners (46 XY / 45 X0) by karyotyping (Table 10).

7 cases of umbilical hernia and 5 cases of isolated cleft palate were the commonest Gastrointestinal malformations. Feeding through cleft palate was managed with obturator plate and Haberman's bottle feeder. Definitive surgery was planned later. Among respiratory malformations, 2 cases of tracheoesophageal fistula were operated. 1 case of tracheal atresia succumbed in spite of emergency tracheostomy being done.

Table 11: Outcome of congenital malformations.

Types of outcome	No. of cases	%
Malformations requiring no intervention	159	58.02
Malformations requiring surgical intervention	58	21.17
Death	57	20.8

Most of the malformations required no intervention (159 cases) (Table 11). However, 58 babies underwent surgical procedures and rest 57 babies succumbed to their illness.

DISCUSSION

Teratology and dysmorphology are terminologies used to describe the various embryological, structural, functional or bio-metabolic disorders in a developing foetus giving rise to congenital malformations.[3] Today, "birth defects" have emerged as a major health concern globally, more so in developed countries where they contribute significantly to neonatal and early childhood mortality. These malformations account for 3% of "major" structural defects, and 15% of "minor" anomalies.[3]

The incidence of structural congenital malformations in 2276 deliveries conducted in this hospital was found to be 274 accounting for 12.0 % of births. Similar results of 61 to 69.9 of birth defects per 1000 live births have been reported in recent studies from India, and South-East Asia.[2] Over this one-year period of this study, the congenital malformations contributed to 12.0% of total annual neonatal mortality noticed in this hospital. Other studies from India have also shown similar figures of malformations contributing to the overall neonatal mortality rate in India (9.6%), accounting thus to 8-15% of perinatal deaths and 13-16 % of neonatal deaths.[4]

Male new-borns (66.7%) had greater incidence of recognizable congenital birth defects than females (33.2%). Similar dominance in males had earlier been reported by Barua et al (60.67% males 37.37% females).[5] This could be due to X- linked recessive factors or Y linked genetic basis. Present study showed prevalence of malformations in 68.9% of term babies and 31% of preterm babies.

This finding is similar studies done by Malla BK (64% term, 36% preterm) and Dutta H (59.4% term, 40.6% preterm).[6,7] 53.2% of babies with malformations weighed less than <2500 gm in the present study. Similar observation (59.8% in babies <2500gm) has been seen in study by Patel ZM.[8] Congenital malformations were significantly more with advanced maternal age >26 years of age. Similar observations were reported by Ronya R et al in mothers older than 30 years of age.[9] Smith D observed that chromosomal abnormalities like Trisomies and Klinefelter's to occur with greater incidence in children born to elderly gravidarums.[10] The most common systems involved in this study were cardiovascular system (29.5%) and central nervous system (13.5%), followed by musculoskeletal system (10.9%) and genitourinary system (7.2%). However, study done by Taksande A showed more involvement of cardiovascular system (23%), musculoskeletal system (21.9%), gastrointestinal tract (14%), genitourinary (18.9%) and central nervous system (9.1%).[11]

This discrepancy could be due to the effect of racial, ethnic and social factors in different parts of the world. During karyotyping, 7 different types of chromosomal anomalies were detected, 2 cases of which had downs syndrome and one of which showed a rare anomaly Male Mosaic Turner's. Karyotyping done by Mohammed YA et al showed chromosomal anomalies in 28 cases (27%) of which Downs syndrome was found in 16 cases.[12]

The limitations of the present study are that it excludes all malformed neonates born outside but treated in our hospital, thus the description of the level of incidence from this region of the state is not covered.

Only recognizable major and minor malformations were considered in this study and karyotyping was done. However, autopsy was not done, and the structural malformations of the internal organs could not be revealed. Lastly, birth defects also include bio-metabolic changes. Present study targeted only structural malformations and other functional anomalies were not studied.

CONCLUSION

Study of malformations should include live and still born babies to get a realistic picture of the incidence of malformations. Autopsy should be included in routine investigation of birth defects as large proportion of defects is found at autopsy. A careful screening and premarital counselling for possible congenital malformation may be undertaken. Preventive genetics can be practised by recognition of individuals who are at an increased risk for producing offspring with a hereditary disorder or in carriers. Hence mothers with positive family history of malformations and bad obstetric history should be screened antenatally for the early detection of possible malformations thereby, reducing the mortality rates.

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