RESEARCH PAPER | Medical Science | Volume : 3 | Issue : 11 | Nov 2013 | ISSN - 2249-555X

# Congenital Anomalies Among Singleton Live Births in a Tertiary Care Teaching Hospital

KEYWORDS: Congenital anomalies, Birth defects, Malformations, Abnormalities

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**ABSTRACT** *Objective: To estimate the incidence and pattern of different Congenital Anomalies among the singleton newborns.*

*Methods: A hospital based study has been carried out in 3,538 newborns in the Department of Obstetrics & Gynaecology and Neonatology, Sri Ramachandra Medical College and Research Institute, Chennai. All subjects were examined thoroughly for anomalies at the time of birth and for the duration of their hospital stay. Gender, Gestational age at delivery, birth weight, APGAR score at 1 and 5 minutes were recorded. DATA has been recorded and analyzed in SPSS 16.0 software.*

*Results: Out of 3,538 consecutive births [Boy babies: 1,855 (52.5%); Girl babies: 1,683 (47.5%)], 73 newborns [2.06% (Boys: 1.13%; Girls: 0.93%)] were identified to have different Congenital anomalies. Major congenital anomalies occurred in higher rate [54(74%)] than Minor anomalies [19(26%)].Congenital anomalies of Cardio Vascular System were the most frequent and accounted in 18 (24.7%) cases, which is followed by the Urogenital 12(16.4%)] and Excretory systems.*

*Conclusion: Overall prevalence of Congenital anomalies is 2.06% among singleton newborns with higher prevalence of Major Congenital anomalies (74%). Cardio Vascular System anomalies (24.7%) were predominant followed by anomalies of Excretory and Urogenital system.*

**INTRODUCTION:**
The term Congenital Anomalies (CA) holds the same meaning of Birth Defects, Congenital Disorders and Congenital Malformations. Congenital Anomalies are defined as "Structural or Functional anomalies including Metabolic Disorders which are present at the time of birth"[1].

Prevalence of CA are quoted between 1.21[2] – 6.7%[3] in the existing literature, which are causing 3.2 million Birth Defect related disabilities every year[1] worldwide and accounts for 8 – 15% of perinatal deaths 13 – 16% of neonatal deaths in India[4].

More than 4,000 different Birth Defects are known, ranging from mild to severe[5]. Few of them are seen right after the baby is born and few can be identified with special tests/ symptoms with advancing age. Many Birth Defects can be prevented by controlling maternal infections, proper diet supplements, vaccines and with efficient antenatal care[6]. Even after all the above, Birth Defects are accounting for a considerable number. Approximately 50% of all the CA are idiopathic[1] even after finding Low Socio Economic status, Genetics, Infections, Maternal Malnutrition, Environmental factors Maternal Health related problems and certain Drugs as causatives[1].

All the CA can be classified into Major and Minor Anomalies depending on the medical/social implications. The CA which needs surgical/medical intervention are Major and which doesn't need are Minor anomalies[7]. Irrespective of Major/ Minor, all the anomalies has the great impact on better living and emotional disturbances in the society.

This present Hospital based study aimed to estimate the incidence and pattern of different CA among the singleton newborns.

**METHODS:**
A hospital based prospective study has been carried out in the Department of Obstetrics & Gynaecology and Neonatology, Sri Ramachandra Medical College and Research Institute (SRMC&RI), Chennai for the duration of 18 months (February 2012 – July 2013).

Institutional Ethical Committee approval has been acquired for the study.

A total of 3,538 subjects were enrolled in the study.

Newborns of all the singleton pregnancies were examined thoroughly for the anomalies. Gender, Gestational age at delivery, birth weight, APGAR score at 1 and 5 minutes were recorded. All the newborns were followed up for their postnatal hospital stay to rule out anomalies.

All the DATA has been recorded and analyzed in SPSS 16.0 software.

**RESULTS:**
Out of 3,538 consecutive births [Boy babies: 1,855 (52.5%); Girl babies: 1,683 (47.5%)], 73 newborns [2.06% (Boys: 1.13%; Girls: 0.93%)] were identified to have different Congenital anomalies. Amongst this group boys were 40(54.8%) and girls were 33(45.2%) which constitutes 2% of all boys and 1.96% of all girl baby population of this study.

Preterm and Term babies were 28(38.35%) and 45(61.6%) within all the anomalous neonates.

Among all the 73 anomalous babies 35(48%) were with normal birth weight (2,500 – 3,800gms), 2(2.7%) were Large for Gestational Age (LGA ≥ 3,800gms) and 36(49%) with Low Birth Weight (LBW ≤ 2,500gms). The mean birth weight of all the neonates was 2403.85±748.807gms (Range: 910 - 4200). (Table: 1)

Major congenital anomalies occurred in 54(74%) and Minor anomalies encountered in 19(26%) newborns. Within all the CA only 3(4.1%) were Genetical.

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In the present study, Gender of the baby Vs Minor / Major anomalies has no significance (P=0.184). Similarly, Maternal Risk Factor Vs Minor/Major anomalies also proven not significant (P=0.123).

Congenital anomalies of Cardio Vascular System (CVS) were the most frequent and accounted for 18 (24.7%) cases, which is followed by the Urogenital [12(16.4%)] and Excretory system[10(13.7%)] (Table: 2,3a - 3e).

**DISCUSSION:**

Prevalence of CA among newborns is between 1.21[2] – 6.7%[3] in the existing literature. The overall prevalence of CA in the present study is 2.06% among 3,538 newborns, which is almost similar to the findings of Emilio Antonio Luca Gianicolo et al[8] study, where the prevalence was 2.3% in his study in 8,503 newborns.

But a lower prevalence was reported by Arjun Singh et al[9] and Harry W et al[10] as 1.5 and 1.15% (Mean = 1.3%), whereas in most of the studies prevalence was higher than the current finding varying from 2.5 – 6.7% (Mean= 4.4%) [3, 11, 12].

In line with most of the previous studies[9, 11] the present study showing the male gender with higher CA (54.8%) than female (45.2%). Which is 1.13 and 0.93% of whole male and female population of the study.

Furthermore a higher rate of CA was reported in Low Birth Weight (LBW) neonates than Large for Gestational Age (LGA) babies and babies with normal birth weight similar to the findings of Mohamed A. El Koumi et al[13].

In contrast to the findings of previous studies, present study showing the higher prevalence of Major CA than Minor anomalies. And there was no significant difference between birth weight and risk status (Major/Minor CA) of anomaly (P=≥0.184) or maternal high risk condition and risk status (Major/Minor CA) of anomaly (P=≥0.123).

The pattern of anomalies also different from most of the studies from India and outside India where Central Nervous System (CNS) [14, 15] or Musculo Skeletal system[9, 11, 13] anomalies were reported predominant. In accordance to Emilio Antonio Luca Gianicolo et al[8] and ICMR Annual report 2002 – 03[4] Cardio Vascular System (CVS) anomalies were predominant [18 (24.7%)] in the present study followed by Urogenital system[12(16.4%)] and Excretory[10(13.7%)].

Only in 5 patients CA occurred in combination of two different systems (Table 2) i.e. Ear & Musculoskeletal system (1, 1.4%), Ear & Urogenital system (1, 1.4%), Respiratory system & CNS (1, 1.4%) and CVS and GIT (2, 2.7%).

The main outcome of our study i.e. prevalence and predominant system affected with anomalies is different from the existing studies holding high prevalence of total anomalies (2.06%), in particular Major CA (74%) and CVS as the predominantly affected system followed by Excretory and Urogenital system. It is representing the tertiary under SRMC&RI is in high risk for anomalies. Thus further research can be suggested in Chennai region on risk identification of different CA, which will reinforce the prevention programme of CA as prevention is the key concern of CA.

**CONCLUSION:**

Overall prevalence of Congenital anomalies is 2.06% among singleton newborns with higher prevalence of Major Congenital anomalies (74%).

Cardio Vascular System anomalies (24.7%) are predominant followed by anomalies of Excretory and Urogenital system.

**ACKNOWLEDGEMENTS**

We would like to thank INSPIRE Division, Department of Science and Technology, Government of India for funding. DR. Binu Ninan Professor of Neonatology, Dr. S. Melani Rajendran, Dr. Rameshkumar Subramanian, Professor of Anatomy, SRMC & RI, Dr. V. Sankar, Head I/C, Department of Anatomy, PGIBMS and Dr. Pushpalatha Baruva, Professor of Obstetrics and Gynaecology for their support.

**Table 1: Descriptive statistics of various variables**

| VARIABLE | MIN. | MAX. | Mean | Std. DEVIATION |
|---|---|---|---|---|
| AGE OF MOTHER | 18 | 42 | 25.77 | 3.890 |
| Hb (gms/dl) | 8 | 13 | 11.29 | 1.011 |
| GES. AGE (WEEKS) | 26+1 | 41+5 | 35+2 | 3.491 |
| APGAR AT (1 Min) | 3 | 8 | 7.31 | 1.148 |
| APGAR AT (5 Min) | 5 | 9 | 8.49 | .924 |
| WEIGHT OF BABY (gms) | 910 | 4200 | 2403.85 | 748.807 |
| NORMAL BIRTH Wt.(gms) | 2490 | 3500 | 2937.10 | 257.853 |
| LGA BIRTH Wt. (gms) | 4000 | 4200 | 4100.00 | 141.421 |
| LBW (gms) | 910 | 2480 | 1781.25 | 471.716 |

**Table 2: Frequency and Percentage of anomalies involved in various systems**

| SYSTEM | FREQUENCY | PERCENTAGE (%) |
|---|---|---|
| CENTRAL NERVOUS SYSTEM (CNS) | 6 | 8.2 |
| CARDIO VASCULAR SYSTEM (CVS) | 18 | 24.7 |
| EXCRETORY SYSTEM | 10 | 13.7 |
| GENETICAL | 3 | 4.1 |
| GASTRO INTESTINAL TRACT | 6 | 8.2 |
| MOUTH | 2 | 2.7 |
| MUSCULO SKELETAL | 4 | 5.5 |
| RESPIRATORY SYSTEM (RS) | 7 | 9.6 |
| UROGENITAL | 12 | 16.4 |
| CVS AND GIT | 2 | 2.7 |
| EAR AND MUSCULOSKELETAL | 1 | 1.4 |
| EAR AND UROGENTAL | 1 | 1.4 |
| RS AND CNS | 1 | 1.4 |
| Total | 73 | 100.0 |

**Table 3a - 3e: Specific Malformations according to systems Involved**

| Table 3a | | | |
|---|---|---|---|
| | MALFORMATION | NO OF CASES | PERCENTAGE (%) |
| CARDIO VASCULAR SYSTEM (CVS) [18 (24.7%)] | | | |
| MAJOR (10) | VSD, MALFORMATIONS OF PERIPHERAL VASCULAR SYSTEM | 1 | 1.4 |
| | VSD, ASD, DISCORDANT VENTRICULOARTERIAL CONNECTION, TRANSPOSITION OF GREAT VESSELS | 1 | 1.4 |
| | ASD | 3 | 4.1 |
| | VSD | 2 | 2.7 |
| | ISOLATED VENTRICULO MEGALI | 1 | 1.4 |
| | ANEURYSM OF RENAL ARTERY | 1 | 1.4 |
| | HIRSCHSPRUNG'S DISEASE | 1 | 1.4 |
| MINOR (8) | PATENT DUCTUS ARTERIOSUS | 8 | 11 |
| CVS & GASTRO INTESTINAL TRACT [2 (2.7%)] | | | |
| MAJOR (2) | MALFORMATION OF BILE DUCTS, ASD,VSD | 1 | 1.4 |
| | PIERRE ROBIN DEFORMITY OR SYNDROME | 1 | 1.4 |

Volume : 3 | Issue : 11 | Nov 2013 | ISSN - 2249-555X

**Table 3b**

| | MALFORMATION | NO OF CASES | PERCENTAGE (%) |
|---|---|---|---|
| RESPIRATORY SYSTEM (RS) [7 (9.6%)] | | | |
| MAJOR (7) | CHOANAL ATRESIA | 2 | 2.7 |
| | HYALINE MEMBRANE DISEASE | 4 | 5.5 |
| | DIAPHRAGMATIC HERNIA | 1 | 1.4 |
| RS & CENTRAL NERVOUS SYSTEM [1 (1.4%)] | | | |
| MAJOR (1) | CONGENITAL MALFORMATION OF NOSE AND CEREBRAL CYSTS | 1 | 1.4 |
| CENTRAL NERVOUS SYSTEM [6 (8.2%)] | | | |
| MAJOR (6) | HYPOPLASTIC CEREBELLUM | 1 | 1.4 |
| | SPINA BIFIDA | 2 | 2.7 |
| | HYDROCEPHALUS | 1 | 1.4 |
| | CONGENITAL CEREBRAL CYSTS | 1 | 1.4 |
| | MENINGOMYLOCOELE | 1 | 1.4 |

**Table 3c**

| | MALFORMATION | NO OF CASES | PERCENTAGE (%) |
|---|---|---|---|
| MOUTH [2 (2.7%)] | | | |
| MAJOR (2) | CLEFT PALATE (UNILATERAL) | 1 | 1.4 |
| | CLEFT LIP, CLEFT PALATE (UNILATERAL) | 1 | 1.4 |
| GASTRO INTESTINAL TRACT [6 (8.2%)] | | | |
| MAJOR (6) | ECTOPIC ANUS, IMPERFORATED ANUS | 1 | 1.4 |
| | STENOSIS OF ANUS AND RECTUM | 1 | 1.4 |
| | ATRESIA /STENOSIS OF DUODENUM | 1 | 1.4 |
| | ABSENCE OF ANUS | 1 | 1.4 |
| | ANTERIORLY PLACED ANUS | 1 | 1.4 |
| | CYSTIC FIBROSIS OF LIVER | 1 | 1.4 |
| EAR & UROGENITAL SYSTEM [1 (1.4%)] | | | |
| MAJOR (1) | ABSENT EAR PINNA Lt, HYPOSPEDIAS, PARAPHYMOSYS | 1 | 1.4 |
| EAR & MUSCULO-SKELETAL [1 (1.4%)] | | | |
| MINOR (1) | LONG EAR, POLYDACTYLY | 1 | 1.4 |

**Table 3d**

| | MALFORMATION | NO OF CASES | PERCENTAGE (%) |
|---|---|---|---|
| UROGENITAL SYSTEM [12 (16.4%)] | | | |
| MAJOR (12) | HYPOSPEDIAS | 5 | |
| | POSTERIOR URETHRAL VALVE | 4 | |
| | UNDESCENDED TESTIS | 3 | |
| EXCRETORY SYSTEM [10 (13.7%)] | | | |
| MAJOR (8) | BATTERS SYNDROME - ROMK TYPE | 1 | 1.4 |
| | LT DISPLASTIC KIDNEY | 1 | 1.4 |
| | HYDRONEPHROSIS | 6 | |
| MINOR (2) | POLYCYSTIC KIDNEY | 2 | 2.7 |

**Table 3e**

| | MALFORMATION | NO OF CASES | PERCENTAGE (%) |
|---|---|---|---|
| MUSCULOSKELETAL SYSTEM [4 (5.5%)] | | | |
| MAJOR (1) | NEOPLASM OF BONE AND ARTICULARCARTILAGE | 1 | 1.4 |
| MINOR (3) | TALIPES EQUINOVARUS | 3 | 4.1 |
| GENETICAL [3 (4.1%) of 73] | | | |
| MAJOR(3) | DOWN'S SYNDROME | 2 | 2.7 |
| | DISORDER OF PLASMA-PROTEIN METABOLISM | 1 | 1.4 |

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