IJPRP

Indian Journal of Pathology: Research and Practice 8(4) July - August 2019
DOI: http://dx.doi.org/10.21088/ijprp.2278.148X.8419.12

Original Research Article

A Study of Multiple Congenital Anomalies in Fetus and Neonates

Kasturi Kshitija[1], Seethamsetty Saritha[2], Sheshagiri Bhaskar[3]

[1]Assistant Professor, Department of Pathology, Bhaskar Medical College & General Hospital, Yenkapally, Telangana 500075, India. [2] Professor & Head, Department of Anatomy, Kamineni Academy of Medical Sciences & Research Center, Hyderabad, Telangana 500068, India. [3]Senior Consultant, Maternity Hospital, India.

Corresponding Author:

Kasturi Kshitija, Assistant Professor, Bhaskar Medical College & General Hospital, Yenkapally, Telangana 500075, India.

E-mail: kshiti80@yahoo.co.in

Received on 19.04.2019,

Accepted on 20.05.2019

Abstract

Background: Congenital Anomalies are one of the leading causes of paediatric mortality in developed & developing countries. Based on World Health Organization 3 million fetuses and infants are born each year with congenital anomalies. In India it is 6-7% which translates to around 1.7 million birth defects annually. This study was undertaken to present the spectrum of multiple congenital anomalies in fetus and neonates. *Materials and Methods:* This is a prospective based observational study done from March 2018 to March 2019. The data for the study was collected from a maternity hospital in Hyderabad. Autopsy was performed on dead fetuses and neonates with birth defects by a standard technique adopted by Edith L. Potter and was documented. *Results:* Of the total of 2043 pregnancies, 47 cases had multiple congenital anomalies with a prevalence of 2.3%. Of which 41 were fetal cases (87.23%) and 6 were neonatal cases (12.76%). Increase incidence of birth defects were noted in consanguinity, young primiparous and low socioeconomic group. Malformations of central nervous system (23.91%) was the commonest. *Conclusions:* Anomalous child has negative impact on mental status of the whole family. The outcome of our study is to initiate a national programme which focuses on prevention, care and surveillance for children born with birth defects.

Keywords: Congenital Anomalies; Birth defects; Fetus; Neonates.

How to cite this article:

Kasturi Kshitija, Seethamsetty Saritha, Sheshagiri Bhaskar. A Study of Multiple Congenital Anomalies in Fetus and Neonates. Indian J Pathol Res Pract. 2019;8(4):449–457.

Introduction

Congenital abnormalities, congenital anomalies (CAs) and birth defects are interchangeable terms used to describe developmental defects that are present at birth [1]. They represents defects in morphogenesis during early fetal life. According to the WHO document of 1972, the term congenital malformations should be confined to structural defects at birth [2]. According to March of Dimes

© Red Flower Publication Pvt. Ltd.

(MOD) Global Report on Birth Defects worldwide 7.9 million births occur annually with birth defects and 94% of these births occur in the low and middle income countries. According to joint WHO and MOD meeting report, birth defects account for 7% of all neonatal mortality [3]. The aetiology of congenital malformation is genetic (30% to 40%), environmental (5% to 10%) and 50% of cases, the cause is unknown. Among the genetic aetiology, chromosomal abnormality constitutes 6%, single gene disorders 25% and multifactorial 20% to 30% [4]. In India congenital anomalies account for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths [5]. This increase risk of birth defects is due to endogamy, consanguinity, large number of unplanned pregnancies, poor antenatal care and maternal nutritional status.

Materials and Methods

The present prospective study was done for a period of one year from March 2018 to March 2019. The data for the study was collected from a maternity hospital in Hyderabad. The study was designed to know about the spectrum of congenital malformations in fetal and neonatal deaths. Fetal malformations in pregnant women were identified prenatally by Ultrasound and Targeted Imaging for Fetal Anomalies scan. Medical termination of Pregnancy was done in these cases with due consent of women in confinement. Mothers who delivered babies with congenital anomalies were also documented. Autopsy protocol was followed to dissect fetuses and neonates showing congenital defects. The autopsy details documented were gender, weight, gestational age of the baby and the type of congenital anomaly involved. Maternal details included the age of the mother, consanguinity, past obstetric history, socio-economic status, past and present medical illness in antenatal period. All multiple pregnancies were excluded from the study. All the congenital malformations were grouped under different organ systems. Each anomaly was counted separately. The results were analyzed by simple statistical techniques. The study also obtained clearance from the ethical committee of the institution.

Results

In a total of 2043 cases, multiple congenital defects were observed in 47 cases (2.3%). From this they were 41 fetal cases (87.23%) and 6 were neonatal cases (12.96%). Out of 47 cases, females were 27 (57.44%), males were 19 (40.42%) and 1 (2.12%) was ambiguous (Fig. 4). The pregnant women were between 19 to 39 years of age groups. Consanguinity was noted in 31 cases (65.95%). Most of the pregnant women with congenital defect in their babies belonged to age group of 19 to 29 years which were 33 cases (70.21%) and in rest 14 cases (29.78%) they were in age group of 30 to 39 years. With regards to parity, in 29 cases (61.7%) primiparous was observed and rest 18 cases (38.29%) multiparous were seen. In 35 cases (74.46%) women belonged to low socioeconomic group and in 12 cases (25.53%) they belonged to middle class. There were 36 unregistered pregnant women (76.59%) who did not have prior antenatal checkup (Fig. 2). In 24 cases (51.06%) anomalies were detected in second trimester with fetuses weighing between 200 to 800 gms, 17 cases (36.17%) in third trimester with fetuses weighing between 800 to 1500 gms and 6 cases (12.76%) in neonatal period with babies weighing between 800 to 1500 gms. Multiple congenital defects were observed in each case. Anomalies identified in II trimester were 48 (52.17%) in 24 cases (Table 1, Fig. 3), in III trimester there were 34 (36.95%) in 17 cases (Table 2, Fig. 3), in neonatal period there were 10 (9.78%) in 6 cases (Table 3, Fig. 3). On a whole 92 anomalies were detected in 47 cases. Central nervous system (23.91%) malformations was commonest followed by genitourinary and limb defects (13.04% each), other defects (11.11%), musculoskeletal and face defects (10.86% each), abdominal and gastrointestinal defects (7.6%), cardiovascular system (7.6%) and respiratory system (2.17%) (Table 4, Fig. 1). Congenital anomalies detected in neonatal, II & III trimester were tabulated according to the organ system.

Table 1: Defects in II trimester according to the system involved

S. No	Organ System	Type of defect	No.	Total	%
1	Central Nervous System	Craniorachischisis	2	16	33.33
		Spinabifida (cystic & aperta)	6		
		Anencephaly	3		
		Arnold Chiari malformations type II	1		
		Dandy Walker Malformation	1		
		Meningocoele (lumbar & occipital)	3		
2	Respiratory System	Bilateral lung hypoplasia	1	1	2.08

3	Genitourinary System	Abdominal testis	1	4	8.3
		Dilated & tortuous ureter	1		
		Long dilated bladder	1		
		Polycystic kidney disease	1		
4	Musculoskeletal System	Scoliosis	1	5	10.41
		Thoracic dystropy	1		
		Rhizome dwarfism	1		
		Overriding of skull bones	2		
5	Limbs	Club feet	1	7	14.58
		Right thumb absent	1		
		Left thumb hyperextension	1		
		Bilateral macrodactyly of toes	1		
		Bilateral polydactyly of hands	1		
		Oligodactyly of hands	1		
		Ill-defined right leg & foot with single toe	1		
6	Face	Facial dysmorphism	2	3	6.25
		Orofacial anomaly	1		
7	Abdominal & Gastrointestinal defect	Omphalocele	3	5	10.41
		Prune belly syndrome	1		
		Lumbar hernia with intestinal contents	1		
8	Others	Sirenomelia	1	7	14.58
		Left diaphragm not developed	1		
		Bilateral cystic hygroma	2		
		Chest anomaly	1		
		Single umbilical artery	2		
		Total	48	48	100

Table 2: Defects in III Trimester according to organ system involved

S. No	Organ System	Type of defect	No.	Total	%
1	Central Nervous System	Spinabifida	2	4	11.76
		Hydrocephalus	1		
		Meningomyelocele	1		
2	Respiratory System	Bilateral lung hypoplasia with absence of interlobar fissure	1	1	2.94
3	Cardiovascular	Patent Interventricular foramen	1	1	2.94
4	Genitourinary System	Horse shoe kidney	1	7	20.58
		Renal agenesis	2		
		Absence of bladder	1		
		Bladder Exostropy	1		
		Penial scrotal Transposition	1		
		Ambiguous External Genitalia	1		
5	Musculoskeletal System	Craniosyntosis	2	5	14.7
		Macrocephalus	1		
		Bell shaped thorax	1		
		Kyphoscoliosis	1		
6	Limbs	Syndactyly of fingers & toes	1	5	14.7
		Severe limb shortening	1		
		Phocomelia	1		
		Merormelia of upper limbs	1		
		Talipes Equinovarus	1		
7	Face	Facial anomaly	2	7	20.58
		Cleft lip	1		
		Cleft palate	1		
		Microstomia	1		
		Synotia	1		
		Down slanting of palpebral fissure	1		
7	Abdominal & Gastrointestinal defect	Gastroschisis	1	1	2.94
8	Others	Sirenomelia	1	3	8.82
		Absent diaphragm	1		
		Single umbilical artery	1		
		Total	34	34	100

Table 3: Defects in Neonates according to organ system involved

S. No	Organ System	Type of defect	No.	Total	%
1	CNS	Meningomyelocele (sacral & lumbar)	2	2	20
2	Cardiovascular	Ectopic cardis	1	6	60
		Patent ductus arteriosus	2		
		Ventricular Septal defect	2		
		Atrial Septal defect	1		
3	Genitourinary System	Bifid Penis	1	1	10
4	Abdominal & Gastrointestinal defect	Omphalocele	1	1	10
		Total	10	10	100

Table 4: Total number of Congenital defects in Fetal & Neonatal Period according to organ system involved

S. No	Organ System	II Trimester	III Trimester	Neonatal	Total	%
1	CNS	16	4	2	22	23.91
2	Respiratory	1	1	0	2	2.17
3	Genitourinary	4	7	1	12	13.04
4	CVS	0	1	6	7	7.6
5	Musculoskeletal	5	5	0	10	10.86
6	Limbs	7	5	0	12	13.04
7	Face	3	7	0	10	10.86
8	Abdominal & GIT	5	1	1	7	7.6
9	Others	7	3	0	10	10.86
	Total	48	34	10	92	100

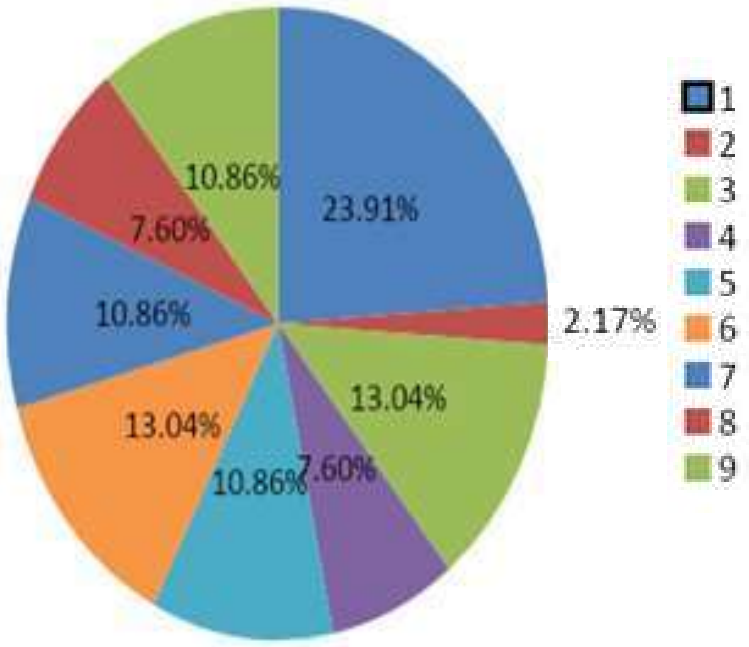

Fig. 1: System wise distribution of Congenital anomalies in percentages

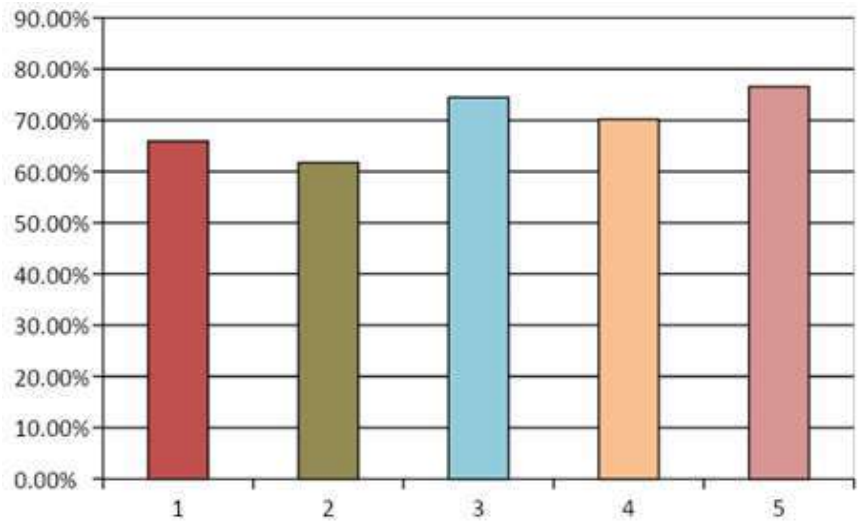

Fig. 2: Maternal details

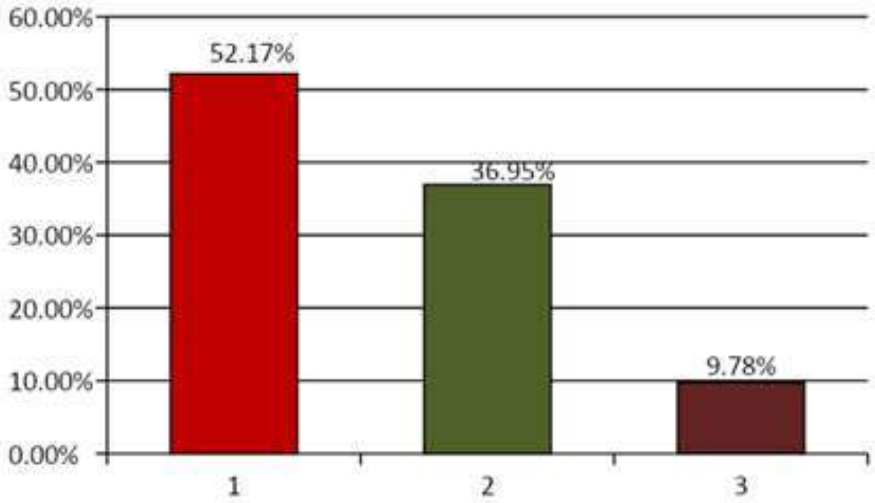

Fig. 3: Percentages of total number of defects detected in fetal and neonatal period

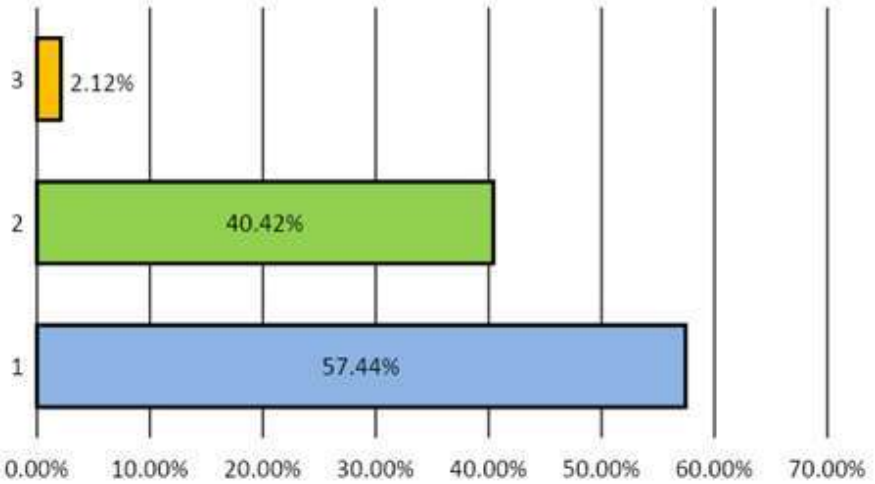

Fig. 4: Percentage of Gender Distribution

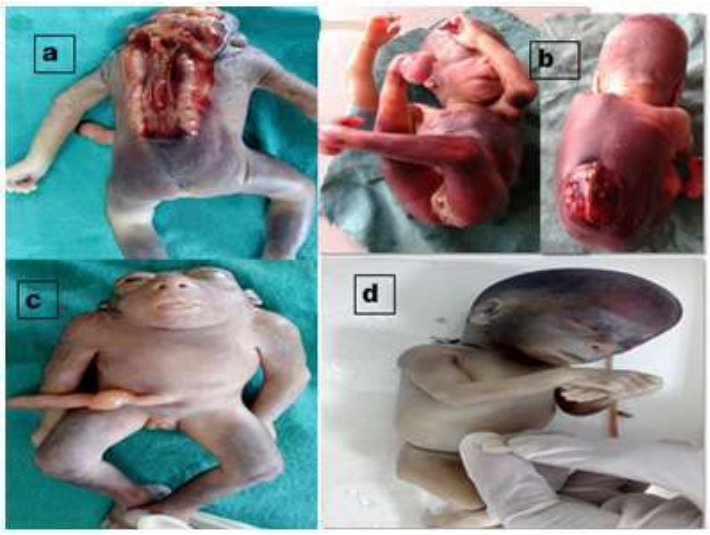

Fig. 5: (a) Craniorachischisis. (b) Anencephaly.(c) Arnold Chiari malformations type II with Spina bifida (d) Dandy Walker Malformation with single umbilical artery

Fig. 6: (a) (b) Potter sequence with renal & bladder agenesis & Penoscrotal Transposition.(c) Diphallus (d) Ambiguous External Genitalia

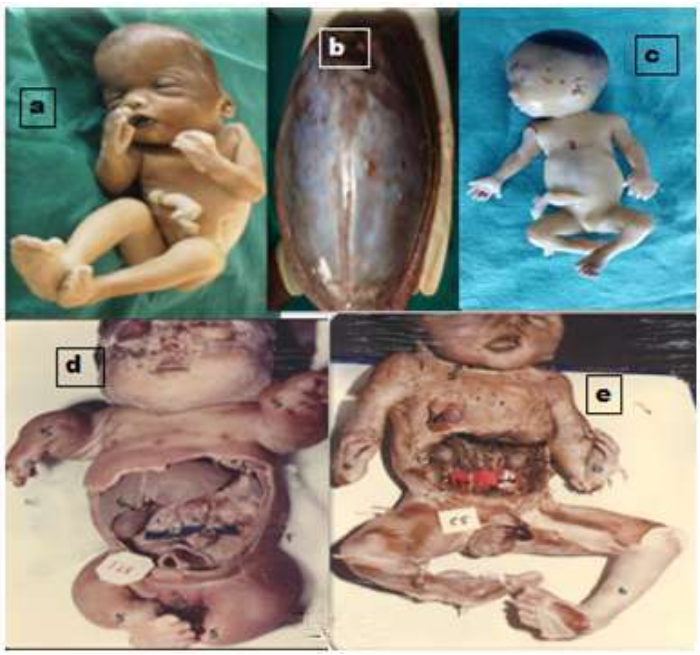

Fig. 7: (a)(b) Apert Syndrome.(c) Jeune syndrome (d) Phocomelia (e) Meromelia with talipesequinus

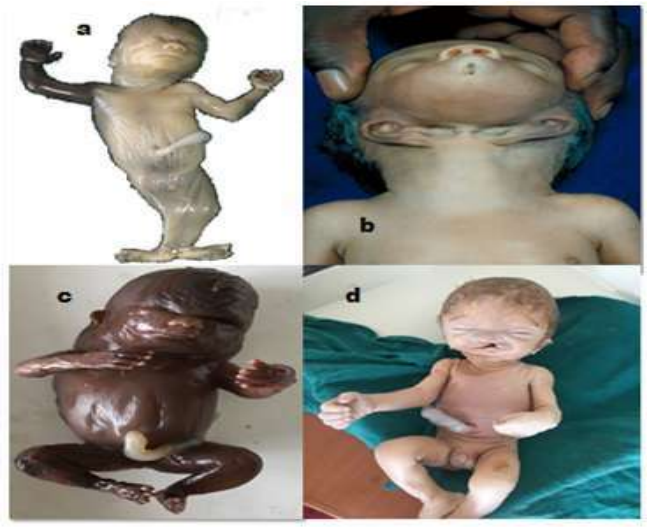

Fig. 8: (a) Sirenomelia (b) Otocephaly-Agnathia (c) Cystic hygroma (d) Cleft lip & palate

Fig. 9: (a) Gastroschisis. (b) Prune Belly Syndrome. (c) Omphalocele. (d) Ectopic Cardia

Fig. 10: Two very rare fetuses with multiple anomalies (a) (b) Case 1. (c) (d) Case 2

Discussion

Worldwide surveys have shown that birth prevalence of congenital anomalies varies greatly from country to country. It is reported to be 1.07% in Japan, 1.49% in South Africa, 2% in England, 2-3% in US and 4.3% in Taiwan [6]. This variation in prevalence could be due to social, racial and economic influences along with increase in the use of radiation, alkylating agents, smoking, alcohol and environmental pesticides [6]. According to study done in USA by Alexander Egbe *et al.* and Mohamed A. El Koumi *et al.* a prevalence of 28.9/1000 and 25/1000 live births respectively was reported [7,8]. Our study has shown the prevalence of CAs 2.3% (23/1000 live births) with 41 fetal deaths (87.23%) and 6 neonatal deaths (12.96%).

Studies have shown a significantly higher incidence of malformations in offspring of consanguineous parents [9]. In our study consanguinity was noted in 31 cases (65.95%). The incident of CAs was seen more in young primigravida between 19-29 years (70.21%) who belonged to low socioeconomic (74.46%) and had no prior regular antenatal check up (76.59%). This can be explained by the fact that these ill literate women were from rural background and had no knowledge of importance of antenatal visit, folic acid medication at the time of conception and ultrsonography. These findings correlated with the studies done by Basavanthappa SP *et al.* 13 and Swain S *et al.* [10, 11].

Occurrence of CAs is higher in abortus and preterm delievers [12]. The gestational age of low birth weight abortus fetus in our study were mostly between 16 to 28 weeks (52.17%) followed by 28 to 32 weeks (36.17%). All 6 neonates were preterm, low birth weight and survived for less than a week. This was consistent with studies done by Sangeetha et al. and Sachdeva *et al.* [13, 14]. The frequency of CAs was more in females (57.44%), 24 fetuses & 3 neonates than in males (40.42%), 16 fetuses & 3 neonates. This correlated with data of Sachdeva *et al.* and Parmer *et al.* [1] but was inconsistent with data of Sozan *et al.* [14,15,16]. One was ambiguous fetus of 32 weeks weighing 916 gms.

More than one CA was detected in each case with a total of 92 anomalies in 47 cases. Anomalies were commonly seen affecting the central nervous system (23.91%). Spina bifida (8.6%) was commonest followed by meningocele (6.5%) and anencephaly (3.2%). This coincided with studies of Sozan K *et al.* (37.7%) and Tomatir A.G. *at al.* (31.1%) [16,17]. Also rare anomalies like Craniorachischisis (2.17%), Arnold Chiari malformations type II (1%) and Dandy Walker malformations (1%) were detected. (Fig. 5).

Anomalies of genitourinary accounted for 13.04% was seen in 6 abortus foetuses with rare defects like Potter sequence (Redundent skin, low set ear, flat nasal bridge, Penial scrotal transposition, renal and bladder agenesis), bifid penis (Diphallus) and ambiguous external genitalia. (Fig. 6). Musculoskeletal defects (10.8%) included Apert's syndrome (craniosynostosis, syndactyly of fingers and toes) in 30 weeks female fetus, Jeune syndrome (thoracic dystrophy, rhizomelic dwarfism, bilateral polydactyly of hands) in 20 weeks female fetus, Phocomelia with rudimentary hands and feet attached to trunk in 28 weeks female fetus and Meromelia of upper limbs with talipes equinovarus in 32 weeks male fetus (Fig. 7). Face defects (10.8%) were cleft lip, cleft palate, Otocephaly –Agnathia (microstomia and synotia). Some of the other defects (10.8%) were 2 Sirenomelia and 2 bilateral cystic hygroma (Fig. 8). Prominent abdominal and gastrointestinal defects (7.6%) were gastroschisis, omphalocele & Prune belly syndrome having thin abdominal wall and severe facial dysmorphology. Cardiac anomalies (7.6%) were ectopic cardia, patent ductus arteriosus, atrial and ventricular septal defect seen in preterm neonates. (Fig. 9) Respiratory anomalies (2.17%) was bilateral lung hypoplasia.

Two very rare fetuses with multiple anomalies were seen. Case 1 was dead male fetus; 24 weeks, weighing 220 gms and showing Orofacial & chest deformity, meningocele, oligodactyly of hand, ill defined legs & foot with single toe on right lower limb (Figs. 10 A,B). Case 2 was macerated male fetus 30-32 weeks showing hydrocephalus, absence of neck, orofacial dysmorphology & vertebral and limb deformity (Figs.10 C,D).

Conclusions

Several studies in India and birth defect registry have addressed the prevalence of birth defects in the country. Their frequency varies from 1.94% to 2.03% of birth [18]. CAs in our study had a prevalence of 2.3% affecting most commonly the central nervous system. Young primigravida with a rural background, consanguineous marriage and no antenatal care were the risk factors. Strategies for the control of birth defects like double fortification of salt, flour fortification with multivitamins, counselling women about the benefits of folic acid supplementation especially preconceptional in

the high risk group, prenatal screening should be implemented especially in primary health centres in villages. Every district should have a birth defects register to record and develop future interventional trials to decrease the birth defects.

References

1. Tanteles GA, Suri M. Classification and etiology of birth defects. Pediatr Child Health. 2007;17:233–43.
2. Shatanik Sarkar Chaitali Patra, Malay Kumar Dasgupta. Prevalence of Congenital Anomalies in Neonates and Associated Risk Factors in a Tertiary Care Hospital in Eastern India. J Clin Neonatal. 2013 Jul-Sep;2(3):131–34.
3. Sharma R. Birth defects in India: Hidden truth, need for urgent attention Indian J Hum Genet. 2013 Apr-Jun;19(2):125–29.
4. Rajangam S *et al.* Consanguinity and chromosomal abnormality in mental retardation and or multiple congenital anomaly. Journal of the Anatomical Society of India. 2007;56:30–3.
5. Prathiba N. Doddabasappa, Adarsh E, Divya N. Prevalence of congenital anomalies: a hospital-based study. Int J Contemp Pediatr. 2018 Jan;5(1):119–23.
6. Rizk Francine, Salameh Pascale, Hamadé Aline. Congenital Anomalies: Prevalence and Risk Factors. Universal Journal of Public Health. 2014;2(2):58–63.
7. Alexander Egbe, Santosh Uppu, Simon Lee *et al.* Congenital Malformations in the Newborn Population: A Population Study and Analysis of the Effect of Sex and Prematurity. Pediatrics and Neonatology. 2015 Feb;56(1):25–30.
8. Mohamed A. El Koumi *et al.* Pattern of congenital anomalies in newborn: a hospital-based study. Pediatric Reports 2013;5:e5.
9. Tayebi N, Yazdani K, Naghshin N. Prevalence of congenital malformations and its correlation with consanguineous marriages. Oman Med J. 2010;25: 37–40.
10. Basavanthappa SP, Pejaver R, Srinivasa V, Raghavendra K, Suresh Babu MT. Spectrum of congenital malformations in newborns: in a medical college hospital in South India. Int J Adv Med. 2014;1:82–5.
11. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. India Pediatr. 1994;31:1187–91.
12. Congenital anomalies fact sheet N 370, World health organization. 2014:5.
13. Sangeeta Chippa et al. Study of congenital anomalies during pregnancy. International J Recent Trends Sci Tech. 2014;12(1):73.
14. Sachdeva S, Nanda S, Bhalla K, Sachdeva R. Gross congenital malformation at birth in a government hospital. Indian J Public Health. 2014;58:54–6.
15. Parmar A, Rathod SP, Patel SV, Patel SM. A Study of Congenital Anomalies in Newborn. NJIRM. 2010;1(1):13.
16. Sozan K. Ameen, Shahla Kareem Alalaf. Pattern of congenital anomalies at birth and their correlations with maternal characteristics in the maternity teaching hospital, Erbil city, Iraq. BMC Pregnancy and Childbirth. 2018;18:501.
17. Tomatir AG, Demirhan H, Sarkun HC, Kaksal A. Major congenital anomalies: a five years retrospective regional study in Turkey. Genet Mol Res. 2009 Jan;8(1):19–27.
18. Verma IC. Burden of genetic disorders in India. Indian journal pediatrics. 2000;67(12):893–8.