STUDY OF CONGENITAL MALFORMATIONS IN A TERTIARY CARE CENTRE, GOVERNMENT GENERAL HOSPITAL, KAKINADA

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ABSTRACT

BACKGROUND

Any deviation from the normal during morphogenesis constitutes an anomaly.[1] Congenital anomaly or malformation is an abnormality of structure, function or body metabolism which is present at birth and results in physical or mental disability. It is an important cause of perinatal mortality and morbidity.

The aim of the study is to determine the prevalence of congenital anomalies, types of anomalies and associated risk factors if any.

MATERIALS AND METHODS

This was a hospital-based cross-sectional study over a period of one year in the Department of Radiodiagnosis, Rangaraya Medical College, Kakinada, AP. All congenital anomaly cases detected antenatally were included in the study. Cases were analysed to find out the prevalence, types of anomalies and its relation with risk factors including maternal age.

RESULTS

Incidence of foetal congenital anomalies were 0.7%. Out of 96 cases of congenital anomaly cases detected Central Nervous System (CNS) deformity was the commonest defect observed in 41 cases (42.7%), out of which maximum cases (32) had neural tube defect. Anomalies were found more in younger age group, in primi gravidae and in women with anaemia and in low socioeconomic group. Various risk factors were associated in 16 number of cases out of 96.

CONCLUSION

Incidence of congenital anomalies were 0.7% and Neural Tube Defect (NTD) was the most common anomaly observed in our study. The risk factors are history of consanguinity (10.4%), previous history of abortions (3.13%) and family history of diabetes mellitus (2.08%). Routine anomaly scan is an important measure for early detection of malformations, primary prevention of disability and reducing perinatal mortality and morbidity.

KEYWORDS

Congenital Anomaly, Neural Tube Defect and its Risk Factors.

HOW TO CITE THIS ARTICLE: Kusumalatha P, Rani KR, Anuradha B, et al. Study of congenital malformations in a tertiary care centre, government general hospital, Kakinada. J. Evolution Med. Dent. Sci. 2017;6(81):5712-5716, DOI: 10.14260/jemds/2017/1239

BACKGROUND

Congenital anomaly or malformation is an abnormality of structure, function or body metabolism which is present at birth and results in physical or mental disability. Each year eight million children are born worldwide with congenital anomalies of which 3.3 million die before the age of five; 3.2 million of the survivors may be mentally or physically disabled.[2] It is supposed to have multi-factorial aetiology and approximately 40% to 60% are associated with unexplained morbidity.[3] The prevalence of congenital anomaly is comparable all over the world. In India, it is responsible for 8% - 15% of perinatal mortality.[4]

'Financial or Other Competing Interest': None.
Submission 28-08-2017, Peer Review 28-09-2017,
Acceptance 03-10-2017, Published 09-10-2017.
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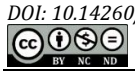
DOI: 10.14260/jemds/2017/1239



Knowing prevalence and type of anomaly in a particular population can be of help in primary prevention of disability and perinatal mortality and morbidity. Foetal anomaly scanning is the most powerful approach available for reducing the birth prevalence of infants with serious congenital abnormalities and increasing the chances of survival for those who are born. Finding of a correctable abnormality can be an indication for delivery to take place at a centre with facilities for paediatric surgery, the finding of a severe uncorrectable abnormality may lead to early termination of pregnancy. Hence, this study was carried out with the following aims and objectives-

1. To find out the incidence of congenital anomalies among antenatal women attending Department of Radiodiagnosis, Government General Hospital, Kakinada.
2. To know the type of anomaly.

MATERIALS AND METHODS

Present study is a hospital-based cross-sectional study carried out in the Department of Radiodiagnosis, Government General Hospital, Kakinada from 1st January 2016 to 31st December 2016. All congenital anomaly cases detected antenatally were

included in the study. Cases detected as congenital anomaly by antenatal ultrasound, but found to be normal at delivery were excluded from the study.

Ethical approval was taken from the Clinical Research Ethics Committee of Rangaraya Medical College and Hospital and written and informed consent was taken from each patient undergoing antenatal ultrasonography.

Patient data were obtained regarding age, area of residence, antenatal check-ups, gravida, parity, socioeconomic status, history of exposure to drug, radiation or history of fever in present pregnancy, previous history of foetal anomaly, history of diabetes, family history of diabetes, congenital anomaly, history of consanguineous marriage and personal history of alcohol consumption and smoking. Gestational age at the time of detection of anomaly was also noted. All Antenatally detected congenital anomaly cases were confirmed at delivery. Counselling and termination was done in cases of lethal anomaly. In some cases, ultrasonography (USG) or x-ray of baby/foetus was done for confirmation. Hb % and blood sugar was checked to find its relation with foetal malformation. Glucose tolerance test (GTT) was done in all the cases.

RESULTS

During the study period incidence of anomaly was found to be 0.7%, as 96 cases were detected out of 13,893 cases. Cases were divided into four age groups: below 20, 20 - 25, 25 - 30, more than 30 years with 5 (5.2%), 57 (59.4%), 24 (25%) and 10 (10.4%) cases respectively and having maximum number in 20 - 25 years age (Table 1).

Age in Years	Number of Cases	Percentage
<20	5	5.2%
20-25	57	59.4%
25-30	24	25%
>30	10	10.4%

Table 1. Showing Age Distribution

Regarding parity, anomalies were found to be more in primipara having 56 cases (58.30%) compared to higher parity as shown in Table 2.

Parity Distribution	Percentage
Primi	58.30%
2nd Gravida	21.87%
3rd Gravida	12.50%
4th Gravida and above	7.30%

Table 2

Only 15 cases (15.6%) were detected at 2nd trimester anomaly scan and majority either at 3rd trimester i.e. 42 cases (43.75%) or at delivery i.e. 39 cases (40.62%). In 24 unbooked cases, malformation was detected at delivery. Among 72 booked cases, only 15 picked up during 2nd trimester anomaly scan and 57 detected at delivery or at 3rd trimester as they failed to follow the advice regarding 2nd trimester USG. More cases were found in lower socioeconomic group with 83 (86.46%) cases as against 13 (13.54%) cases in middle and higher socioeconomic group. Amongst 96 cases of anomaly, 68 cases (71%) were anaemic with 65% in low socioeconomic group and 6% were in higher socioeconomic group.

Only 16 out of 96 (16.67%) cases had associated risk factors like consanguineous marriage, family history of diabetes mellitus, congenital anomaly, previous history of delivering anomalous baby and as shown in Table 4. GTT was normal in all cases.

Risk Factors	No. of Cases	Percentage
Antenatal exposure to radiation, drug intake	0	0%
History of febrile illness	0	0%
History of bleeding per vagina	0	0%
Family history of diabetes	2	2.08%
Family history of congenital anomaly	1	1.04%
Previous history of anomalous baby delivered	3	3.13%
History of consanguineous marriage	10	10.41%

Table 3. Showing the Number of Cases with Risk Factors

Regarding type of anomaly (Table 5) CNS defect was the commonest anomaly observed with 41 cases (42.7%), of which maximum number had neural tube defect i.e. 32 (33.32%) cases. This is followed by Genitourinary tract abnormality, ventral wall defect, limb deformity and cleft lip with or without cleft palate. Multiple congenital anomalies were seen only in 2 (2.08 %) cases. Anaemia was found to be significantly associated with neural tube defect, as out of 38 cases of NTD 36 cases were anaemic (Table 6).

Type of Anomaly	No. of Cases	Percentage
1. Central nervous system defects	41 cases	42.7%
i. Neural tube defects	32	33.32%
a. Anencephaly	18	
b. Hydrocephalus	10	
c. Meningocele	3	
d. Encephalocele	1	
ii. Arnold-Chiari malformation	1	1.04%
iii. Holoprosencephaly	3	3.12%
iv. Corpus callosal agenesis	2	2.08%
v. Schizencephaly	1	1.04%
vi. Dandy-Walker malformation	1	1.04%
vii. Microcephaly	1	1.04%
2. Genitourinary tract abnormality	11	11.6%
a. B/L PUJ obstruction	1	1.05%

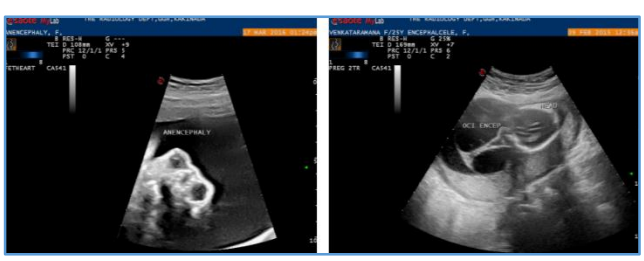
b. B/L Hydronephrosis	7	
c. B/L polycystic kidney	2	
d. Posterior urethral valves	1	
3. Ventral wall defect	8	8.33%
a. Omphalocele	5	
b. Gastroschisis	3	
4. Limb deformity	8	8.33%
a. Achondroplasia	2	
b. Polydactyly	1	
c. Talipes Equinovarus	4	
d. Thanatophoric dysplasia	1	
5. Cleft lip with/or palate	5	5.20%
a. Cleft palate	1	
b. Cleft lip	3	
c. Cleft lip with cleft palate	1	
6. Duodenal atresia	4	4.16%
7. TRAPS (Acardiac twin)	3	3.15%
8. Congenital heart defect	3	3.15%
9. Diaphragmatic hernia	2	2.08%
10. Amniotic band syndrome	2	2.08%
11. Multiple congenital anomalies	2	2.08%
12. Turner's syndrome	1	1.04%
13. Foetal ascites	1	1.04%
14. Situs inversus	1	1.04%
15. Mesenteric cyst	1	1.04%

Table 4. Showing Number of Cases according to the Types of Anomaly

	With NTD	**Without NTD**	**Total**	
Anaemia present	36 (38%)	30 (31%)	66 (69%)	P value < 0.0001
Anaemia absent	2 (2%)	28 (29%)	30 (31%)	
Total	**38 (40%)**	**58 (60%)**	**96 (100%)**	

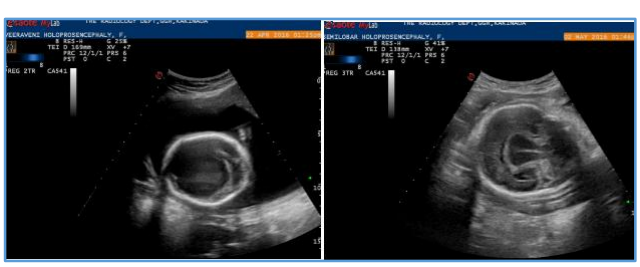
Table 5. Showing the Prevalence of NTD with Anaemia

Figure 1. Neural Tube Defects



Anencephaly with Frog Eye Appearance ***Occipital Encephalocele***

Figure 2. Holoprosencephaly



Monoventricle with Fusion of Midline Structures- Alobar Type ***Fusion of the Frontal Lobes with Incomplete Falx-Semilobar Type***

Figure 3. Alobar Holoprosencephaly with Proboscis

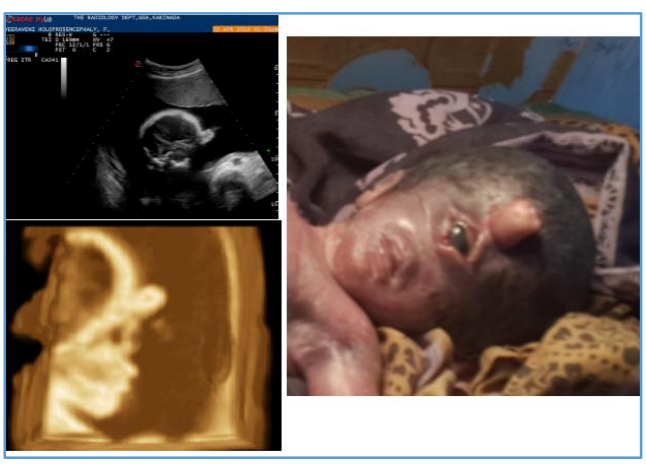
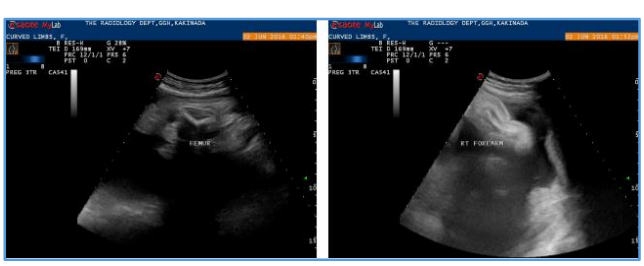
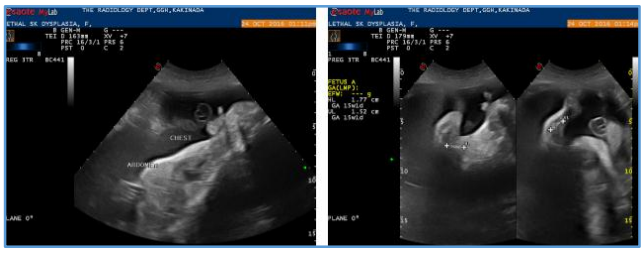


Figure 4. Non-Lethal Skeletal Dysplasia



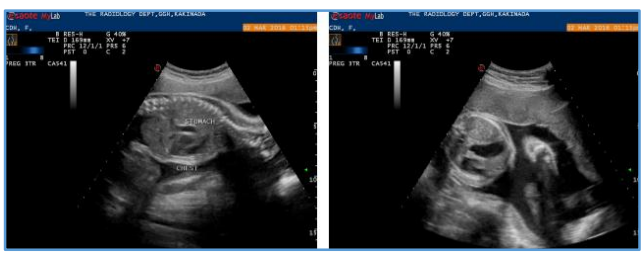
Curved Femur ***Curved Ulna and Radius***

Figure 5. Lethal Skeletal Dysplasia



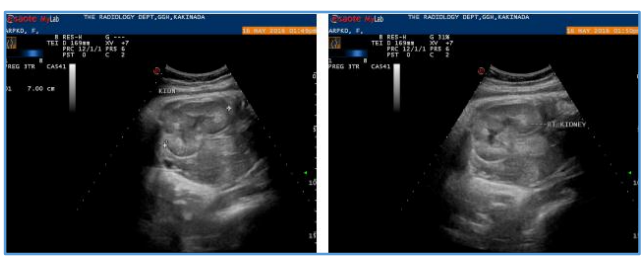
Narrow Thorax *Short Stubby Long Bones*

Figure 6. Congenital Diaphragmatic Hernia



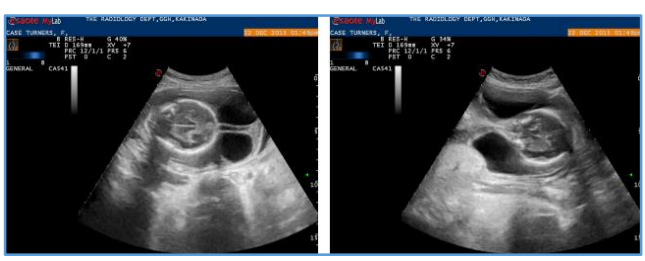
Stomach Bubble seen Herniating into the Thorax- Sagittal and Axial Views

Figure 7. Autosomal Recessive Polycystic Kidney Disease



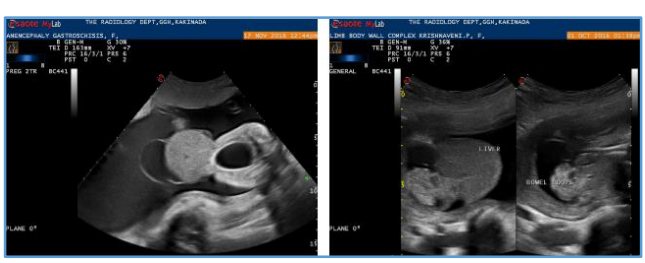
Bilateral Enlarged Echogenic Kidneys

Figure 8. Cystic Hygroma



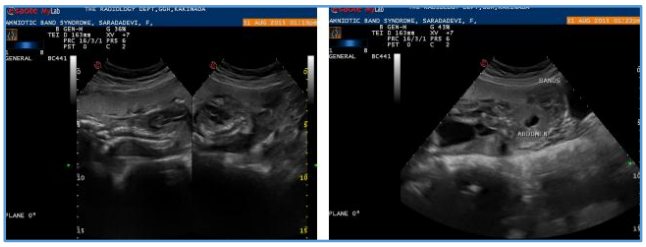
Axial and Coronal Views showing Cystic Lesions in the Region of the Neck

Figure 9. Anterior Abdominal Wall Defects



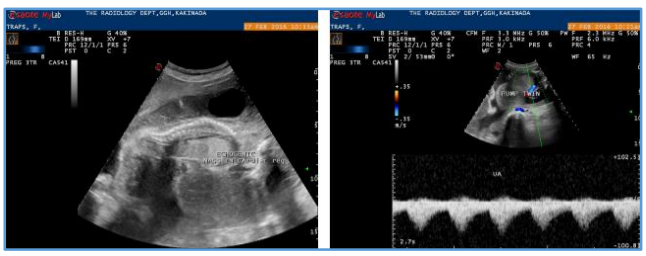
Omphalocele *Gastroschisis*

Figure 10. Limb Body Wall Complex



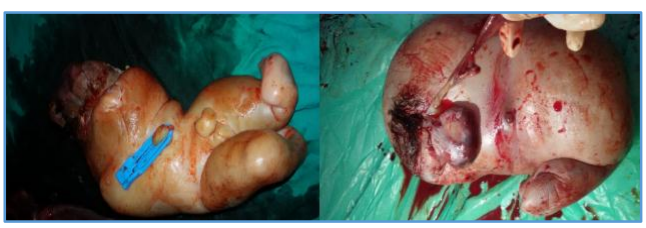
Deformed Spine *Amniotic Bands*

Figure 11. TRAPS



Acardiac Twin with Echogenic Mass in the Cardiac Region Flow Reversal in the Umbilical Artery of Pump Twin

Figure 12. TRAPS- Acardiac Acephalous Foetuses



Foetus with well-developed Lower Extremities **Amorphous Foetus**

DISCUSSION

In our study, incidence of congenital anomaly was 0.7% which is similar to the figure (0.69%) observed by Taboo ZAA,[5] but Chinara PK and Singh S,[6] Chaturvedi P and Banerjee KS[7] reported 2.08% and 2.27% respectively. This variation may be due to different geographical area, social factor and racial difference. Though elderly age group and higher parity are considered as risk factors for congenital anomaly, in our study higher incidence was observed in primipara and younger age group.[8] Similar findings were reported by Perveen F and Tyyab S in 2007.[9]

Out of 96, only 15 cases were detected in 2nd trimester and 81 cases were detected at 3rd trimester or at delivery either due to lack of antenatal check-up or not doing obstetric USG on time. Socioeconomically, highest number of pregnant women with anomaly belonged to lower class i.e. 86.5%. In a similar study conducted by Vrijheid M et al in 2001 reported that the risk of structural anomalies were more in population with increased socioeconomic deprivation; 65% cases (62) belonging to low socioeconomic status were anaemic. Out of 38 NTD cases 36 were anaemic ($P < 0.0001$) showing a significant correlation between lower socioeconomic statuses with anaemia and neural tube defect (NTD).

There were 3 cases of congenital anomaly having family history of diabetes, though GTT was normal which was similar to the observation reported by Sheffield SJ et al.[10]

Central nervous system defect was the commonest anomaly seen constituting 42.7% and maximum number had neural tube defect (39.58%). In a similar study conducted by Agarwal SS,[11] in 1999 neural tube defect was found to be the commonest malformation. Similarly, Perveen F and Tyyab S[9] in 2007 also found NTD as the commonest type of anomaly, i.e. 65.8%. Amongst NTDs, majority had hydrocephalus and anencephaly in our study.

However, Sigmund HE et al,[12] Krikunova NI et al[13] and Aziza et al[14] found cardiovascular, musculoskeletal and facial cleft respectively to be more common. This may be related to food habit and geographical variation.

CONCLUSION

In this study, incidence of foetal congenital anomaly was found to be 0.7%. Incidence was more in younger age group, primipara and lower socioeconomic status and among anaemic patients. In majority, there was no associated risk factor, which indicates all pregnancies are at potential risk of foetal malformation. Neural tube defect was found to be the commonest form of anomaly in our population and is significantly related to anaemia. Creating awareness regarding anaemia correction in preconception period, regular antenatal check-ups and importance of anomaly scan on time can help in primary prevention of disability and reducing perinatal mortality and morbidity.

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