Section A-Research paper

A SYSTEMATIC STUDY REGARDING ETIOLOGY OF FETAL ANOMALIES DURING PREGNANCY AND ITS NEONATAL OUTCOME IN TERTIARY CARE HOSPITALS

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ABSTRACT

Background: According to WHO Congenital anomalies are defined as structural or functional anomalies, including metabolic disorders which are present at the time of birth. Congenital anomalies are a major health problem accounting for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths in India. This study was done to know the frequency, pattern of congenital anomalies and various presentations, which may help to develop strategies for patient counseling and management. The objective of study was etiology of fetal anomalies during pregnancy in district headquarters hospital Khammam. **Methods:** A prospective observational study regarding etiology of fetal anomalies during pregnancy and its neonatal outcome, analytical hospital based study of 300 patients who delivered or aborted congenital anomalous baby from a period of 1st september 2021 to 31st March 2022.**Results:** In this study, we observed few multiple congenital anomalies are Acrania, atrioventricular septal defect and single umbilical artery, choroid plexus cyst and single umbilical artery, mega cisterna magna, small bowel obstruction and single umbilical artery, small bowel obstruction and hydronephrosis, vasa previa and Oligohydramnios. The neonatal outcome of all these anomalies

are found to be life threatening of fetus respectively. **Conclusions**: From the present study of various fetal anomalies involving multiple organ systems and we conclude that consanguinous marriages are associated with more significant development of fetal anomalies. Neural tube defects are the most common type of anomalies that are identified prenatally. These anomalies are associated with increased mortality and morbidity in the neonatal period. In view if increasing prevalence of fetal anomalies, there is a need to further identify other factors causing fetal anomalies such as pesticides, drugs and radiation causing mutations.

Keywords: Acrania, Fetal Anomalies, hydronephrosis, choroid plexus, hydronephrosis and Umbilical artery

INTRODUCTION

Congenital anomalies are a major health problem and are responsible for a remarkable proportion of mortality and morbidity in newborns. It affects 3-5% of live births in the United States, in India the reported incidence is 2.5%. Congenital anomalies account for 8% to 15% of perinatal deaths and 13% to 16% of neonatal deaths in India.1 According to WHO Congenital anomalies are defined as antenatal care prevents early diagnosis of the malformations[1,2].

Our hospital is a tertiary care medical college hospital receiving patients from nearby districts. It is important to know the frequency, pattern of congenital anomalies and various presentations. This in turn will help to develop strategies for patient counselling and management. Patient should be seen early in pregnancy and second trimester ultrasound scan should be performed. Neonatal management along with medical and surgical intervention counselling needs to a part of the strategy. Obstetric management planning will prevent complications as these patients have associated risk factors like anaemia, gestational diabetes, polyhydramnios, and malpresentations[3,4].

This study was aimed at presenting the spectrum of various congenital anomalies, epidemiological features of pregnant women with anomalous fetus. Fetal and neonatal details. Other associated antenatal complications and mode of delivery [5,6].

METHODOLOGY[7-15]

STUDY DESIGN: A prospective observational study regarding etiology of fetal anomalies during pregnancy and its neonatal outcome.

STUDY SITE: Govt. District headquarters hospital, Khammam.

STUDY PERIOD: 6 Months (Sept 2021-March 2022).

STUDY POPULATION: 300 subjects.

PATIENT ENROLLMENT: Patient who fulfill the criteria of inclusion and exclusion.

Inclusion criteria:-

1. Pregnant women
2. Age between >18 and <45.

Exclusion criteria:

1. Non pregnant women.

2. Patient's age <18.

3. Patients with other co-morbid conditions.

RESULTS AND DISCUSSION:

A prospective observational study was conducted in Khammam and Metpally locality for a period of 6 months. A total of 300 patients were enlisted under the inclusion and exclusion criteria.

DISTRIBUTION OF CASES BASED ON THE TYPE OF ANOMALY:

Table 01. Case distribution based on type of anomaly

Type of congenital anomalies	No. of cases collected	Percentage (%)
Isolated congenital anomalies	187	62.33
Multiple congenital anomalies	113	37.67
Total	300	100

In our study, we screened about 300 cases of fetal anomalies and its neonatal outcome. Out of 300 cases, 187 (62.33%) were isolated congenital anomalies and 113 (37.67%) were multiple congenital anomalies, and the results shown in the table no.01.

Fetal anomalies and its neonatal outcome in central nervous system and spine:

Table 02. Fetal anomalies and neonatal outcome in central nervous system and spine

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal Outcome	No. of Cases Noticed	Percentage (%)

1	Acrania	8	12.5	Life Threatening	6	75
2	Arachnoid Cyst	8	12.5	Seizures	6	75
3	Blake's Pouch Cyst	5	7.81	Hydrocephalus	3	60
4	Choroid Plexus Cyst	8	12.5			0
5	Agenesis Of Corpus Callosum	4	6.25	Andermann Syndrome	3	75
6	Dandy Walker Malformation	6	9.37	Abnormal Growth	5	83.33
7	Macrocephaly	3	4.68	Intractable Seizures	2	66.66
8	Megacisterna Magna	8	12.5			0
9	Microcephaly	3	4.68	Abnormal Growth	2	66.66
10	Ventriculomegaly	6	9.37	Mental Retardation	4	66.66
11	Open Spina Bifida	2	3.12	Paralysis	2	100
12	Hemivertebra	3	4.68	Scoliosis	3	100

In table no.02, represented the values of anomalies and its neonatal outcomes in central nervous system and spine reflects the anomalies Acrania cases 8(12.5%), its outcome life threatening cases 6 (75%), archnoid cyst cases 8 (12.5), its outcome seizures 6 (75%), blake's pouch cyst cases 5 (7.81%), its outcome hydrocephalus cases 3 (60%), choroid plexus cyst cases 8 (12.5%), its outcome is not applicable, agenesis of corpus callosum cases 4 (6.25%), its outcome andermann syndrome cases 3 (75%), dandy walker malformation cases 6 (9.37%), its outcome abnormal growth cases 5 (83.33%), Macrocephaly cases 3 (4.68%), its outcome intractable seizures cases 2 (66.66%), mega cisterna magna cases 8 (12.5%), and its outcome is not applicable, Microcephaly cases 3 (4.68%), its outcome abnormal growth cases 2 (66.66%), ventriculomegaly cases 6 (9.37%), its outcome mental retardation cases 4 (66.66%), open spina bifida cases 2 (3.12%), its outcome paralysis cases 2 (100%), Hemivertebra cases 3 (4.68%), its neonatal outcome scoliosis cases 3 (100%).

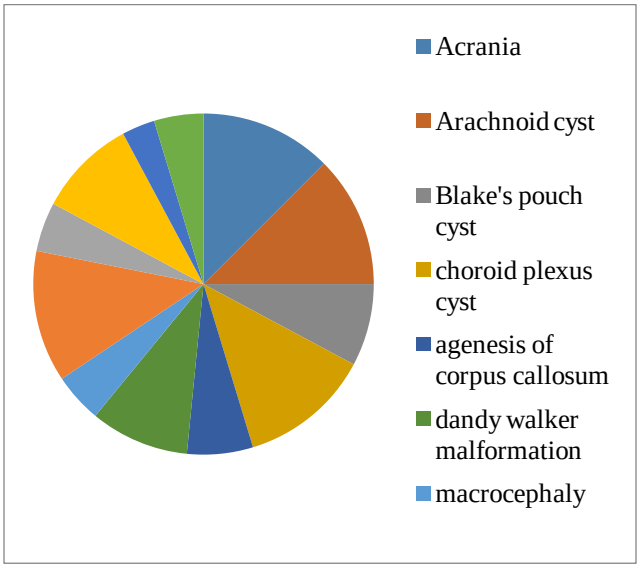

Figure 01. Fetal anomalies in central nervous system and spine

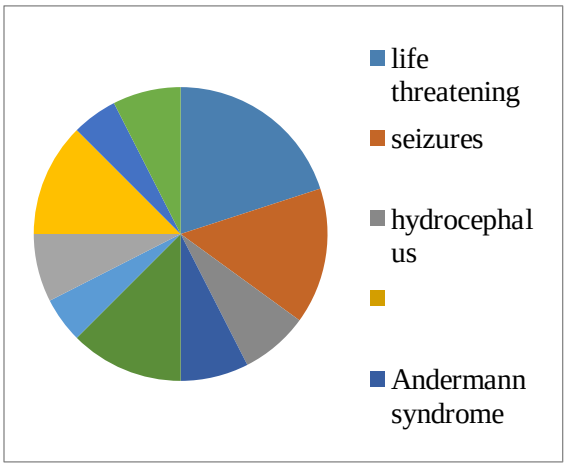

Figure 02. neonatal outcome in central nervous system

Fetal anomalies and its neonatal outcome in face and neck:

Table 03. fetal anomalies and its neonatal outcome in face and neck

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal Outcome	No. of Cases Noticed	Percentage (%)
1	Cataract	2	11.11	Abnormal Vision	1	50
2	Facial Cleft Binder	8	44.44	Dental Problems	8	100
3	Syndrome Cervical	6	33.33	Life Threatening	5	83.33
4	Teratoma	2	11.11	Life Threatening	2	100

In table no.03, represented the values of anomalies and its neonatal outcomes in face and neck reflects the anomalies cataract cases 2 (11.11%), its outcome abnormal vision cases 1 (50%), facial cleft cases 8 (44.44%), its outcome dental problems cases 8 (100%), binder syndrome cases 6 (33.33%), its outcome life threatening cases 5 (83.33%), cervical teratoma cases 2 (11.11%), its outcome life threatening cases 2 (100%).

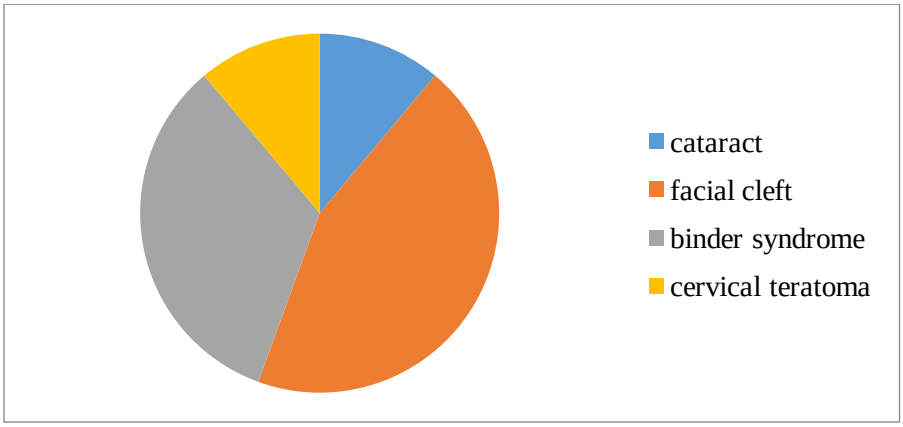

Figure 03. fetal anomalies in face and neck

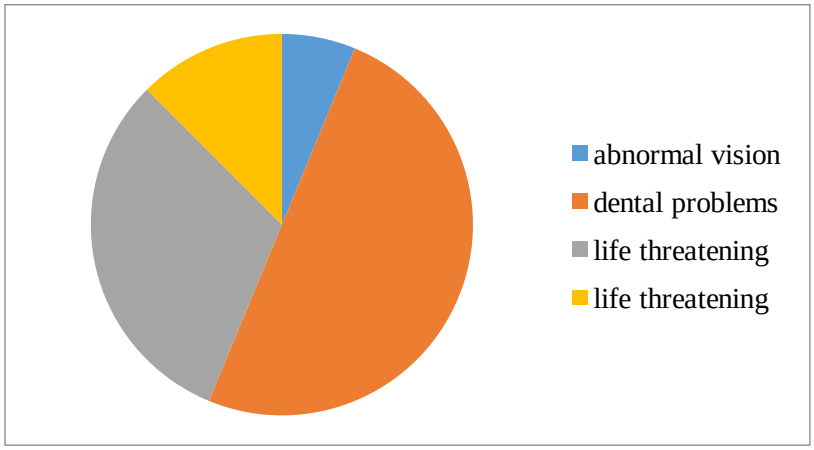

Figure 04. neonatal outcome in face and neck

Fetal anomalies and its neonatal outcome in thorax:

Table 04. fetal anomalies and its neonatal outcome of thorax

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal Outcome	No. of Cases Noticed	Percentage (%)

1	Pleural Effusion	5	50	Pulmonary Disorder	2	40
2	Diaphragmatic Hernia	2	20	GERD	2	100
3	CPAM	2	20	Life Threatening	2	100
4	CHAOS	1	10	Life Threatening	1	100

In table no.04, represented the values of anomalies and its neonatal outcomes in thorax reflects the anomalies pleural effusion cases 5 (50%), its outcome pulmonary disorder cases 2 (40%), diaphragmatic hernia cases 2 (20%), its outcome GERD cases 2 (100%), CPAM cases 2 (20%), its outcome life threatening cases 2 (100%), CHAOS cases 1 (10%), life threatening cases 1 (100%).

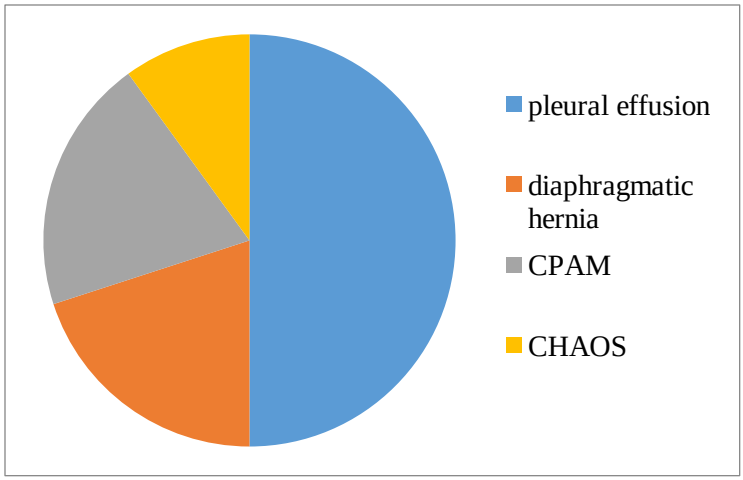

Figure 05. fetal anomalies in thorax

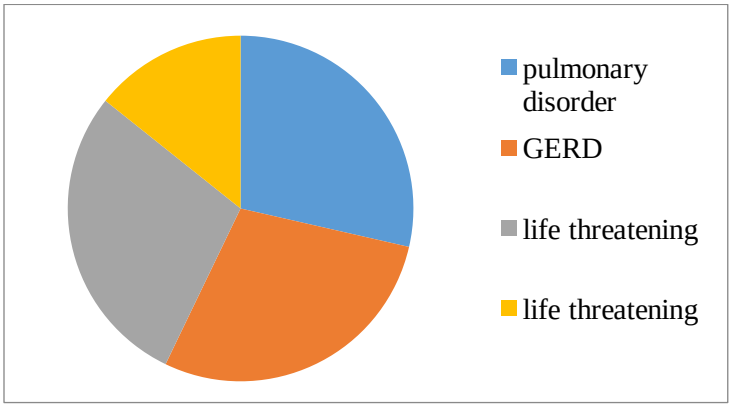

Figure 06. neonatal outcome in thorax

Fetal anomalies and its neonatal outcome in cardiovascular system:

Table 05. fetal anomalies and its neonatal outcome in cardiovascular system

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal outcome	No. of Cases Noticed	Percentage (%)
1	Atrioventricular Septal Defect	4	44.44	Heart Failure	2	50
2	Tetralogy of Fallot And RAA	2	22.22	Pulmonary Stenosis	1	50
3	Tricuspid Atresia With VSD	3	33.33	Patent Foramen Ovale	2	66.66

In table no.05, represented the values of anomalies and its neonatal outcomes in cardiovascular system reflects the anomalies atrioventricular septal defect cases 4 (44.44%), its outcome heart failure cases 2 (22.22%), tetralogy of fallot and RAA cases 2 (22.22%), its outcome pulmonary stenosis cases 1 (50%), tricuspid atresia with VSD cases 3 (33.33%), its outcome patent foramen ovale cases 2 (66.66%).

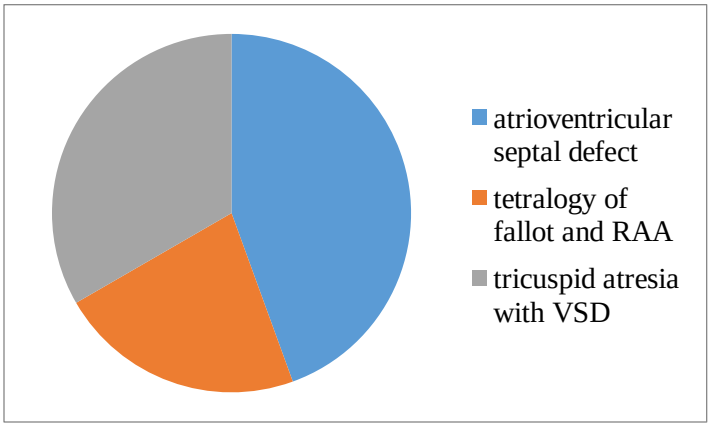

Figure 07. fetal anomalies in cardiovascular system

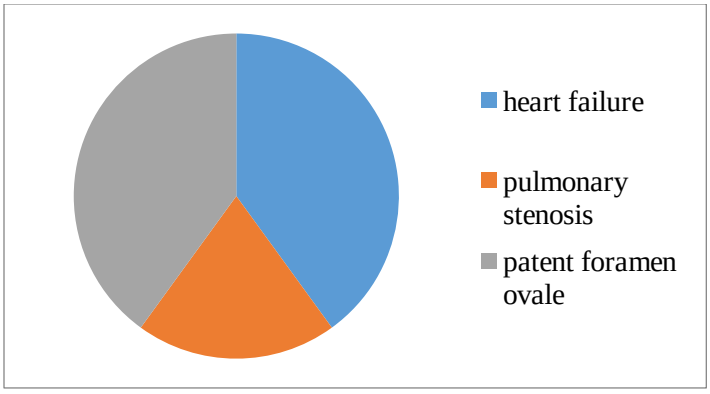

Figure 08. neonatal outcome in cardiovascular system

Fetal anomalies and its neonatal outcome in GIT & Abdomen:

Table 06. fetal anomalies in GIT & Abdomen

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal outcome	No. oxf Cases Noticed	Percentage (%)
1	Bladder Exstrophy	8	40	Urinary Incontinence	5	62.5
2	Small Bowel Obstruction	12	60	Peritonitis	10	83.33

In table no.06, represented the values of anomalies and its neonatal outcomes in GIT & abdomen reflects the anomalies bladder exstrophy cases 8 (40%), its outcome urinary incontinence cases 5 (62.5%), small bowel obstruction cases 12 (60%), its outcome peritonitis cases 10 (83.33%).

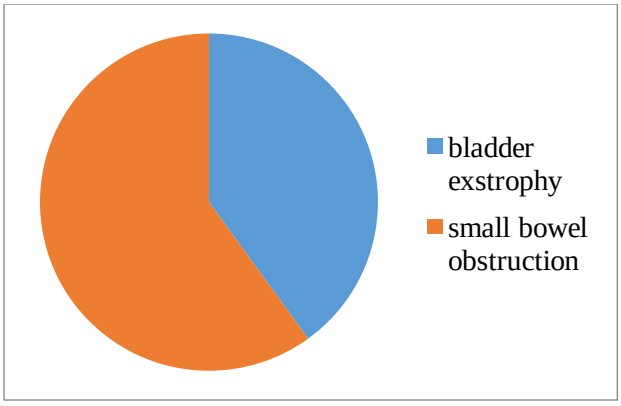

Figure 09. fetal anomalies in GIT & Abdomen

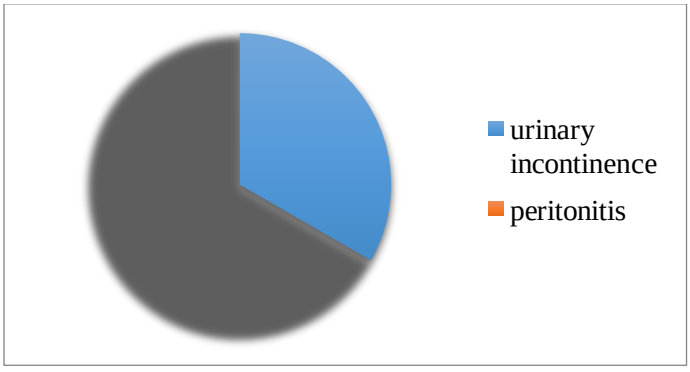

Figure 10. neonatal outcome in GIT & Abdomen

Fetal anomalies and its neonatal outcome in urinary tract:

Table 07. fetal anomalies and its neonatal outcome of urinary tract

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal Outcome	No. of Cases Noticed	Percentage (%)
1	Horseshoe Kidney	2	13.33	Hydronephrosis And Nephrolithiasis	1	50
2	Hydronephrosis	13	86.66	NA	0	0

In table no.07, represented the values of anomalies and its neonatal outcomes in urinary tract reflects the anomalies horse shoe kidney cases 2 (13.33%), its outcome hydronephrosis and nephrolithiasis cases 1 (50%), Hydronephrosis cases 13 (86.66%), and its outcome is not applicable.

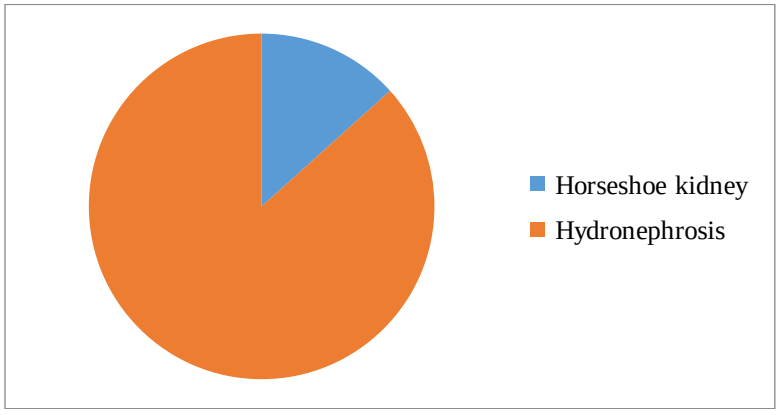

Figure 11. fetal anomalies in urinary tract

Fetal anomalies and its neonatal outcome of extremities:

Table 08. fetal anomalies and its neonatal outcome of extremities

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal Outcome	No. of Cases Noticed	Percentage (%)
1	Amniotic band syndrome	3	33.33	Life Threatening	3	100
2	Club foot	4	44.44	Neuromuscular Disorder	4	100
3	Club hands	2	22.22	Comorbity	1	50

In table no.08, represented the values of anomalies and its neonatal outcomes in extremities reflects the anomalies amniotic band syndrome cases 3 (33.33%), its outcome life threatening cases 3(100%), club foot cases 4 (44.44%), its outcome neuromuscular disorder cases 4 (100%), club hands cases 2 (22.22%), its outcome comorbity cases 1 (50%).

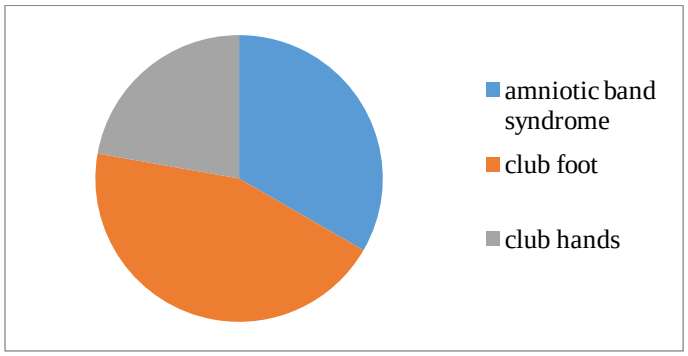

Figure 12. fetal anomalies of extremities

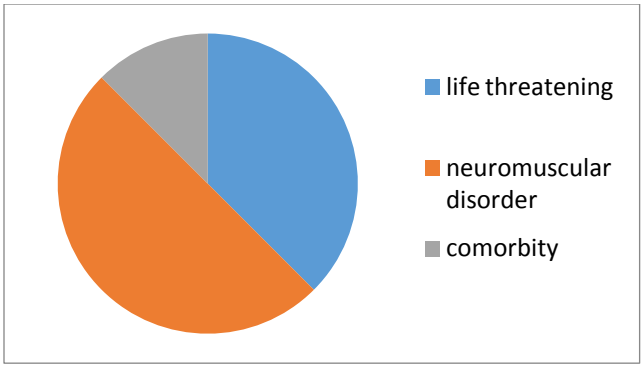

Figure 13. neonatal outcome of extremities

Fetal anomalies and its neonatal outcome of skeleton:

Table 09. fetal anomalies and its neonatal outcome of skeleton

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal outcome	No. of Cases Noticed	Percentage (%)
1	Achondrogenesis	4	33.33	Pulmonary Hypoplasia	3	75
2	Achondroplasia	3	25	Pulmonary Hypoplasia	2	66.66
3	Diastrophic dysplasia	5	41.66	Scoliosis	5	100

In table no.09, represented the values of anomalies and its neonatal outcomes in skeleton reflects the anomalies Achondrogenesis cases 4 (33.33%), its outcome pulmonary hypoplasia cases 3 (75%), Achondroplasia cases 3 (25%), its outcome pulmonary hypoplasia cases 2 (66.66%), diastrophic dysplasia cases 5 (41.66%), its outcome scoliosis cases 5 (100%).

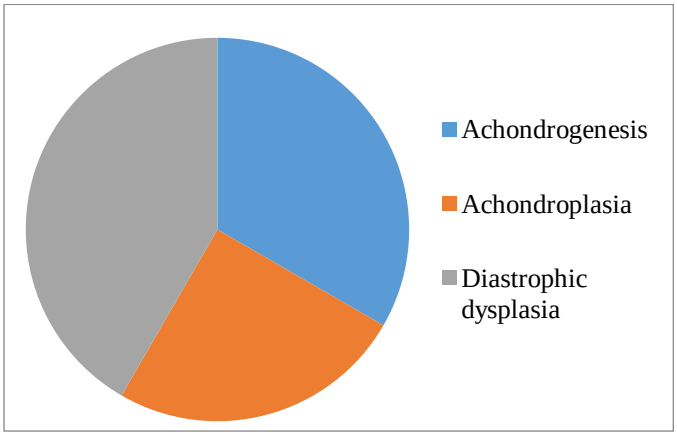

Figure 14. fetal anomalies of skeleton

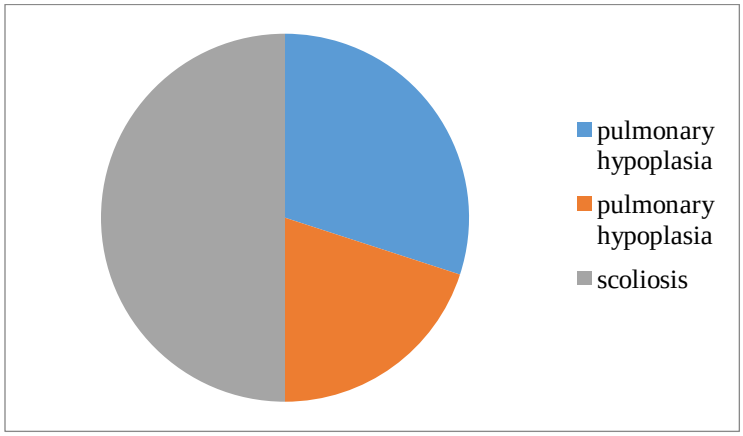

Figure 15. neonatal outcome of skeleton

Fetal anomalies and its neonatal outcome of umbilical cord and placenta:

Table 23. fetal anomalies and its neonatal outcome of umbilical cord and placenta

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal outcome	No. of Cases Noticed	Percentage (%)
1	Single Umbilical Artery	10	71.42	Growth Restriction	8	80
2	Vasa Previa	4	28.57	IUD	2	50

In table no.23, represented the values of anomalies and its neonatal outcomes in skeleton reflects the anomalies single umbilical artery cases 10 (71.42%), its outcome growth restriction cases 8 (80%), Vasa previa cases 4 (28.57%), its outcome IUD cases 2 (50%).

All this data was reflected in the below pie charts.

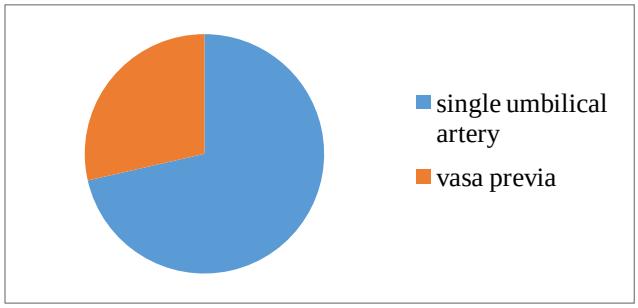

Figure 16. fetal anomalies of umbilical cord and placenta

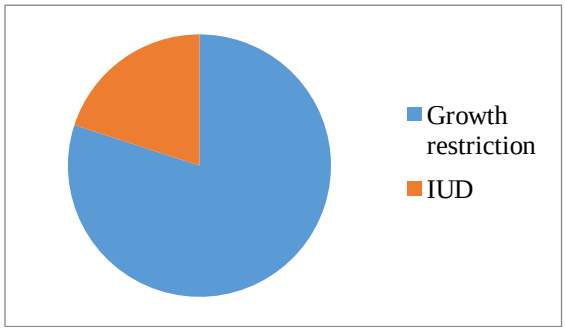

Figure 17. neonatal outcome of umbilical cord and placenta

Fetal anomalies and its neonatal outcome of amniotic fluid:

Table 10. fetal anomalies and its neonatal outcome of amniotic fluid

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal outcome	No. of Cases Noticed	Percentage (%)
1	Oligohydramnios	6	54.54	Immature Lungs	5	83.33
2	Polyhydramnios	5	45.45	SOB	3	60

In table no.10, represented the values of anomalies and its neonatal outcomes in amniotic fluid reflects the anomalies Oligohydramnios cases 6 (54.54%), its outcome immature lungs cases 5 (83.33%), Polyhydramnios cases 5 (45.45%), its outcome SOB cases 3 (60%).

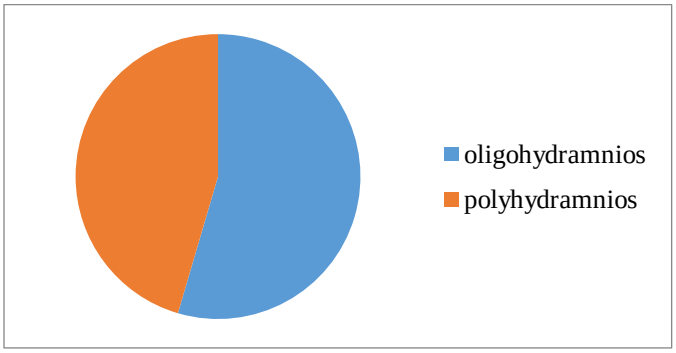

Figure 18. fetal anomalies of amniotic fluid

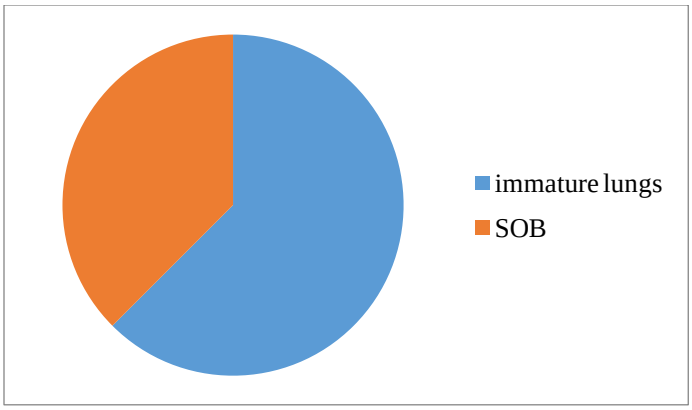

Figure 19. neonatal outcome of amniotic fluid

Fetal anomalies and its neonatal outcome of multiple pregnancies:

Table 11. fetal anomalies and its neonatal outcome of multiple pregnancies

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal outcome	No. of Cases Noticed	Percentage (%)
1	MC Twins: Conjoined Twins	2	40	Phyiscally Handicap	2	100
2	MC Twins: Death of One Fetus	3	60	Life Threatening	3	100

In table no.11, represented the values of anomalies and its neonatal outcomes in multiple pregnancies reflects the anomalies MC twins: conjoined twins cases 2 (40%), its outcome physically handicap cases 2 (100%), MC twins: death of one fetus cases 3 (60%), its outcome life threatening cases 3 (100%).

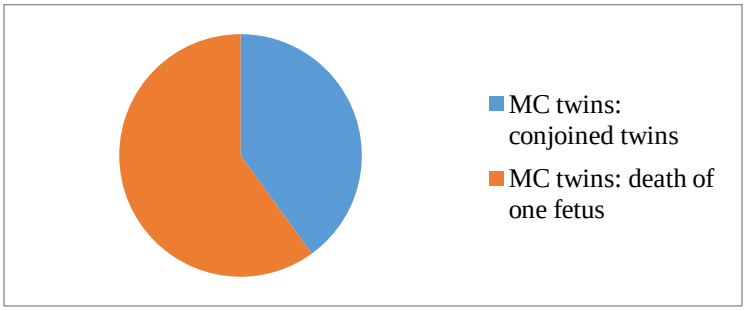

Figure 20. fetal anomalies in multiple pregnancies

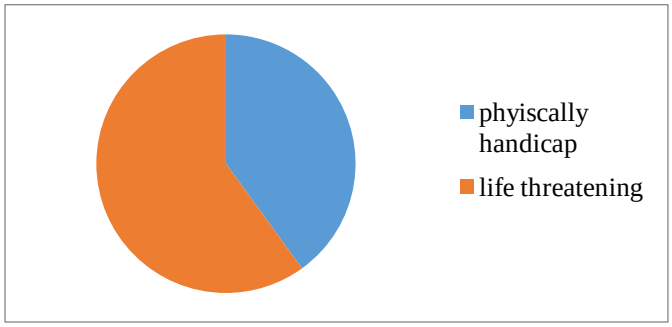

Figure 21. neonatal outcome in multiple pregnancies

Multiple congenital anomalies affecting the fetus:

Table 12. multiple congenital anomalies with its neonatal outcome

S.No	Anomalies	No. of Cases Noticed	Percentage (%)	Neonatal Outcome	No. of Cases Noticed	Percentage (%)
1	Acrania ,Atrioventricular Septal Defect& Single Umbilical Artery	15	13.27	Life Threatening	15	13.27
2	Choroid Plexus Cyst & Single Umbilical Artery	20	17.69	Life Threatening	20	17.69
3	Megacisterna Magna, Small Bowel Obstruction& Single Umbilical Artery	40	35.39	Life Threatening	40	35.39
4	Small Bowel Obstruction & Hydronephrosis	25	22.12	Life Threatening	25	22.12
5	Vasa Previa & Oligohydramnios	13	11.50	Life Threatening	13	11.50

In table no.12, represented the values of multiple congenital anomalies and its neonatal outcomes, the anomalies were acrania ,atrioventricular septal defect& single umbilical artery cases noticed 15 (13.27%), its outcome life threatening cases 15 (13.27%), choroid plexus cyst & single umbilical artery cases 20 (17.69%), its outcome life threatening cases 20 (17.69%), mega cisterna magna, small bowel obstruction& single umbilical artery cases 40 (35.39%), its outcome life threatening cases 40 (35.39%), small bowel obstruction & hydronephrosis cases 25 (22.12%), %), its outcome life threatening cases 25 (22.12%), Vasa Previa & Oligohydramnios cases 13 (11.50%), its outcome life threatening cases 13 (11.50%).

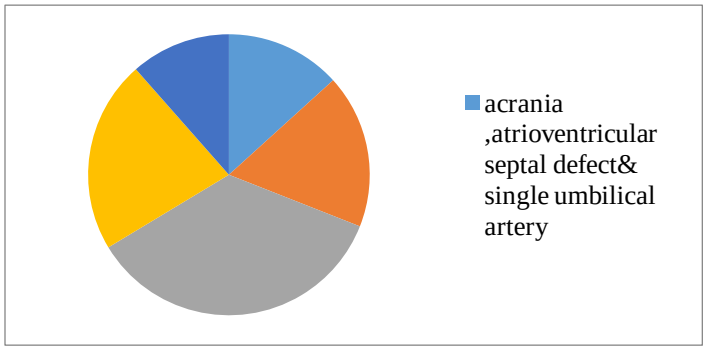

Figure 22. multiple congenital anomalies

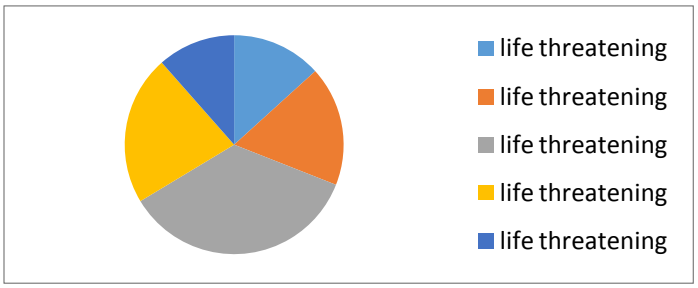

Figure 23. neonatal outcome in multiple congenital anomalies

Total isolated fetal anomalies and its percentage according to the system:

Table 13. Total isolated fetal anomalies and its percentage according to the system

S.No	Name of body organ(s)	No. of cases collected	Percentage (%)
1.	Central nervous system and spine	64	21.33
2.	face and neck	18	6
3.	Thorax	10	3.33
4.	Cardiovascular system	9	3
5.	Gastrointestinal tract & abdomen	20	6.66
6.	urinary tract	15	5
7.	Extremities	9	3
8.	Skeleton	12	4
9.	umbilical cord & placenta	14	4.66
10.	amniotic fluid	11	3.66
11.	multiple pregnancies	5	1.66
	TOTAL:	**187**	**62.33%**

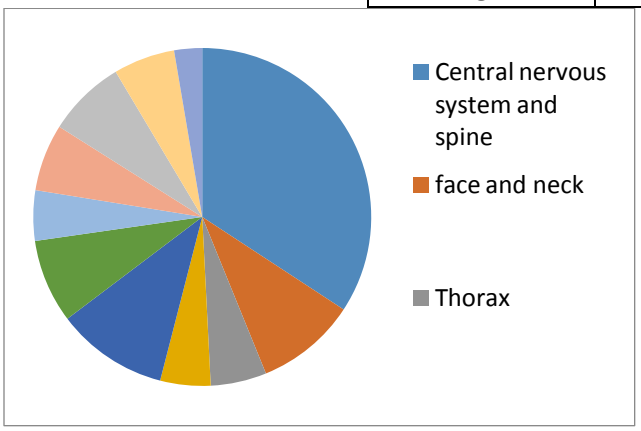

Figure 24. Total isolated fetal anomalies

Total no. of multiple congenital anomalies cases:

Table 14. Total no. of multiple congenital anomalies

S.No	Name of anomalies	Total no. of cases collected	Percentage (%)
1	Multiple congenital fetal anomalies	**113**	**37.67%**

In Table no.13, it represents the total no. of fetal anomalies according to the body organs, total no. of isolated cases are 187 and its percentage was found to be 62.33% and In Table no.14, it represents the total no. of multiple congenital fetal anomalies cases are 113 and its percentage was found to be 37.67 %.

CONCLUSION

From the present study of various fetal anomalies involving multiple organ systems and we conclude that consanguineous marriages are associated with more significant development of fetal anomalies. Neural tube defects are the most common type of anomalies that are identified prenatally. These anomalies are associated with increased mortality and morbidity in the neonatal period. In view if increasing prevalence of fetal anomalies, there is a need to further identify other factors causing fetal anomalies such as pesticides, drugs and radiation causing mutations.

CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

BIBILIOGRAPHY

1. Sadler TW. Birth Defects and Prenatal Diagnosis. In: Langman's medical embryology. 13th ed. Philadelphia: Wolters Kluwer; 2015. p. 126–40.

2.Congenital anomalies WHO2016. http://www.who.int/mediacentre/factsheets /fs370/en /. Accessed 7 May 2017.

3. Ndibazza J, Lule S, Nampijja M, Mpairwe H, Oduru G, Kiggundu M, et al. A description of congenital anomalies among infants in Entebbe, Uganda. Birt Defects Res A Clin Mol Teratol. 2011;91:857–61.

4. Oestergaard MZ, Inoue M, Yoshida S, Mahanani WR, Gore FM, Cousens S, et al. Neonatal mortality levels for 193 countries in 2009 with trends since 1990: a systematic analysis of Progress, projections, and priorities. PLoS Med. 2011;8:e1001080.

5. Liu L, Oza S, Hogan D, Perin J, Rudan I, Lawn JE, et al. Global, regional, and national causes of child mortality in 2000–13, with projections to inform post2015 priorities: an updated systematic analysis. Lancet. 2015;385: 430–40.

6. Sitkin NA, Ozgediz D, Donkor P, Farmer DL. Congenital anomalies in lowand middle-income countries: the unborn child of global surgery. World J Surg. 2015;39:36–40.

7. Kumar P. Dysmorphology. In: Kumar P, Burton B, editors. Congenital malformations. 1st edition. Blacklick. USA: McGraw-Hill Professional Publishing; 2008. p. 3–11.

8. Sarkar S, Patra C, Dasgupta MK, Nayek K, Karmakar PR. Prevalence of congenital anomalies in neonates and associated risk factors in a tertiary care hospital in eastern India. J Clin Neonatol. 2013;2:131.

9. Egbe A, Uppu S, Lee S, Stroustrup A, Ho D, Srivastava S. Congenital malformations in the newborn population: a population study and analysis of the effect of sex and prematurity. Pediatr Neonatol. 2015;56:25–30.

10. Ekanem TB, Okon DE, Akpantah AO, Mesembe OE, Eluwa MA, Ekong MB. Prevalence of congenital malformations in Cross River and Akwa Ibom states of Nigeria from 1980–2003. Congenit Anom. 2008;48:167–70.

11. Obu HA, Chinawa JM, Uleanya ND, Adimora GN, Obi IE. Congenital malformations among newborns admitted in the neonatal unit of a tertiary hospital in Enugu, south-East Nigeria - a retrospective study. BMC Res Notes. 2012;5:177.

12. Bakare TIB, Sowande OA, Adejuyigbe OO, Chinda JY, Usang UE. Epidemiology of external birth defects in neonates in south western Nigeria. Afr J Paediatr Surg. 2009;6:28.

13. Adeyemo AA, Gbadegesin RA, Omotade OO. Major congenital malformations among neonatal referrals to a Nigerian University teaching hospital. East Afr Med J. 1997;74:699–701.

14. Chen B-Y, Hwang B-F, Guo Y-L. Epidemiology of congenital anomalies in a population-based Birth registry in Taiwan, 2002. J Formos Med Assoc. 2009; 108:460–8.

15. Cosme HW, Lima LS, Barbosa LG. Prevalence of congenital anomalies and their associated factors in newborns in the city of Sao Paulo from 2010 to 2014. Rev Paul Pediatr Orgao 0Of Soc Pediatr Sao Paulo. 2017;35:33–8.