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A study to detect birth defects scenario in a tertiary care hospital New Delhi

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Abstract
Background: Birth defects are a group of structural or functional disorders which are present at birth and are of prenatal origin can be caused by genetic defects, environmental teratogens or multifactorial inheritance. A total of 290 newborns were diagnosed with birth defects in an observational cross-sectional study conducted in Department of Obstetrics and Gynaecology in collaboration with the Department of Pediatrics, at VMMC and Safdarjung Hospital, New Delhi. Before starting the study, permission was taken from the ethical committee of the institution. All women who delivered a live birth were tracked to study the spectrum of birth defect and its association with maternal risk factors. A total of 290 newborns were diagnosed with birth defects. Male female ratio was 1.30:1. Out of a total of 25,565 deliveries, 24,905 were live births and 660 were stillbirths. Prevalence of birth defects among stillbirths was higher, as compared to live births i.e. 4.54% and 1.04% respectively; the difference was statistically significant, p=<0.0001.The prevalence of birth defects among live births is 1.04% and among still births is 4.54%. Birth defects involving the central nervous system are the commonest i.e. 36.2%, followed by Oro-facial defects (cleft-palate and cleft-lip) i.e. 18.6% and GIT i.e. 14.13%.

Keywords: Birth defect, Teratogens, Live birth, Stillbirth, Oro-facial defect, Small Intestine atresia etc.

Introduction
Birth defects are a diverse group of structural or functional disorders that are present at birth and are of prenatal origin, which can be caused by genetic defects, environmental teratogens or multifactorial inheritance [1]. Every year 6% of children worldwide are born with serious birth defects due to genetic or environmental causes [3]; based on the latest annual birth data, the estimate would be nearly 9.78 million [5]. According to WHO estimates, in 2004, nearly 260,000 deaths worldwide (about 7% of all neonatal death) were caused by birth defects [6]. They are most prominent as a cause of death in settings where overall mortality rates are lower, for example in the European Region, where nearly 25% of neonatal deaths are due to birth defects [1]. It is estimated that, India has the largest number of affected children and that, every year, around 1.6 lack children are born with birth defects [7].

According to various studies, the reported prevalence of birth defects varies from 1.5 to 4.1% [8, 9, 10, 11]. They may affect various organ systems including central nervous system (CNS), musculoskeletal system (MSS), cardiovascular system (CVS) and genitourinary system (GUS). The most common serious birth defects are Congenital Heart Disease, Neural Tube Defects (NTD) and Down syndrome; Hemoglobinopathies (including thalassemia and sickle-cell disease) and glucose-6-phosphate dehydrogenase deficiency account for 6% of all congenital disorders [1]. A number of risk factors have been reported by various studies to be associated with birth defects [8, 9, 11]. Folic acid supplementation and maternal cigarette smoking have been implicated with increase risk of NTDs and CVS defects respectively [11]. Advanced maternal age, prematurity and increase birth order have also been reportedly associated with birth defects [9]. Consanguinity of marriage has also been observed to be an important risk factor in the occurrence of birth defect and inborn errors of metabolism [11, 12].

Various strategies have been suggested, to reduce the incidence of birth defects and to take care of affected children [1, 3]. These strategies require comprehensive maternal, newborn and child health programmes and provision of basic medical genetic services. Most birth defects of environmental origins can be prevented by public health approaches like vaccination against rubella and food fortification with iodine and folic acid [1]. Medical and surgical methods are required to treat and rehabilitate children with birth defects. All these measures would ensure

that people with birth defects or at reproductive risk of having children with birth defects, can live and reproduce as normally as possible [3].

A very few studies have shown the data regarding birth defects from developing countries. Besides, the studies lack a systematic approach and an adequate sample size. Keeping all this in view, the present study would be an effort to bridge the gaps in the existing literature. It aims to determine the prevalence and spectrum of birth defects at a tertiary care hospital.

Methodology

This was observational, cross sectional study. All women who delivered in labor room of Safdarjung Hospital New Delhi and who were willing to participate in the study protocol were enrolled from November 2013 to October 2014. These women were divided into two groups as follows.

Study group (n=290) comprising women who delivered a (live or stillborn) baby with birth defect, detected at birth or before the final outcome (discharge or death).

Control group (n=290) comprising women who delivered a live baby, without birth defect, consecutive to the study case.

All babies (live and still born) were examined clinically by a paediatrician for any major or minor birth defect. Mothers of all babies (study and control group) were interviewed through a predesigned questionnaire which include-

- Demographic profile of mother consisting of maternal age, religion, residence, education and socioeconomic status, Obstetric history including parity, previous pregnancy loses history of birth defects in previous babies, infection, ingestion of drugs including folic acid supplementation in its trimester, exposure to radiation, co-morbidities (Diabetes, thyroid disorder, hypertension disorder or any other medical illness), Consanguinity of marriage, Diabetes, hypertension, heart disease and any history of birth defects in any family member.

Statistical analysis – Simple descriptive tabulation were made from the data and it was analyzed by SPSS version 21.0. Qualitative analysis was done by using Chi-square test and p-value less than 0.05 was considered as significant.

Results and observations

Prevalence of birth defects

Out of a total of 25,565 deliveries, 24,905 were live births and 660 were stillbirths. A total of 290 newborns were diagnosed with birth defects, giving a prevalence of 1.13% or 113/10,000 births. Prevalence of birth defects among stillbirths was higher, as compared to live births i.e. 4.54% and 1.04% respectively; the difference was statistically significant, p=<0.0001. The sex wise distribution of birth defects was 56.6% males and 43.4% females ; Male female ratio was 1.30:1 (4 cases not included due to ambiguous genitalia) (Table-1).

Table 1: Birth Defects; Frequency and Sex Distribution.

	Total Cases (Number))	Birth Defects (Number)	Percent (%)	*p value*
Total Births	25,565	290	1.13	
Live births	24,905	260	1.04	
Still births	660	30	4.54	<0.0001
Male	13,783	162	1.17	
Female	11,782	124	1.05	
Ambiguous		4	1.37	

Spectrum of birth defects

Defects of CNS were the commonest, i.e. 36.2%, followed by Orofacial defects (cleft palate/cleft lip) i.e. 18.6% and GIT i.e.14.1 % respectively. Defects of CVS were observed in 10%, while musculoskeletal defects were observed in 7.6%. Chromosomal anomalies were seen in 2.1% cases. (Table-2)

Table 2: Spectrum of Birth Defects

System Involved	Number	Percent (%)
Central Nervous System (CNS)	105	36.2
Cardiovascular system(CVS)	29	10.0
Genitourinary(GUS)	21	7.2
Gastrointestinal(GIT)	41	14.1
Musculoskeletal(MSS)	22	7.6
Orofacial (Cleft lip/Cleft palate)	54	18.6
Chromosomal	6	2.1
Others	12	4.1

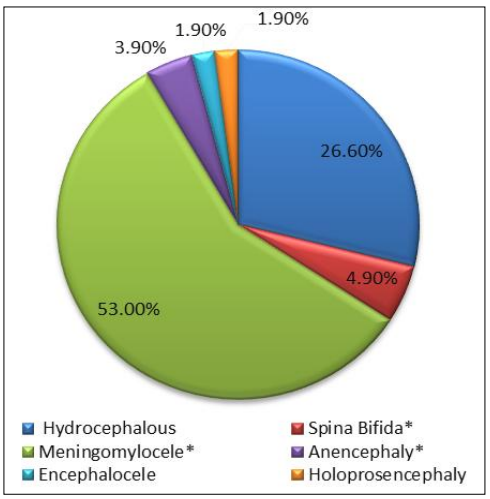

Fig 2: Central Nervous System (n=105)

Central Nervous System

Among CNS birth defects, neural tube defects (NTDs) were the commonest i.e. 65.6%, followed by hydrocephalous i.e. 26.6%. Holoprosencephaly, was present in 1.9% cases. (Figure-2)

Cardiovascular System

Amongst CVS birth defects, Cyanotic Congenital Heart disease (CCHD) was observed in 41.4%; of these, Tatrology of Fallot was the commonest. {Figure-2 (a)}

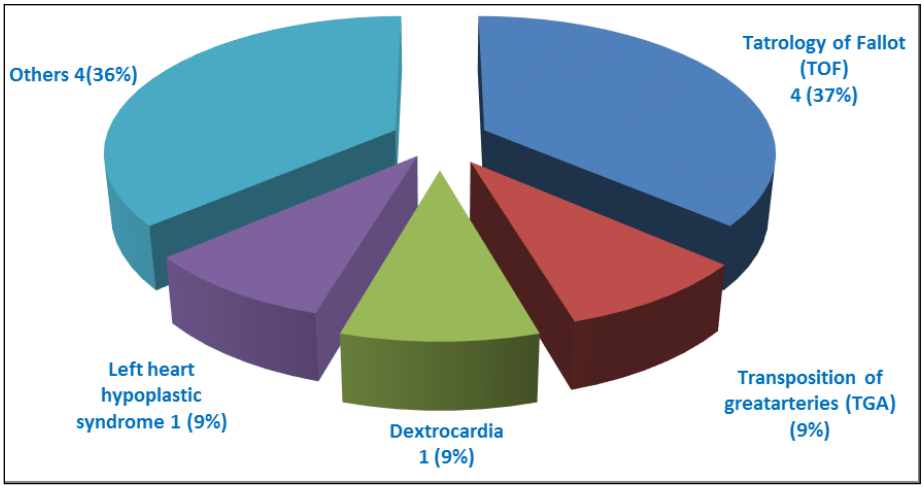

Fig 2 (a): Cyanotic Congenital Heart Disease (CCHD) (n=12)

Acyanotic congenital heart disease was observed in 58.6% cases; of these, Patent Ductus Arteriosus (PDA) was the commonest, followed by ventricular septal defect (VSD) and Atrial Septal Defect (ASD) respectively. {Figure 2(b)}

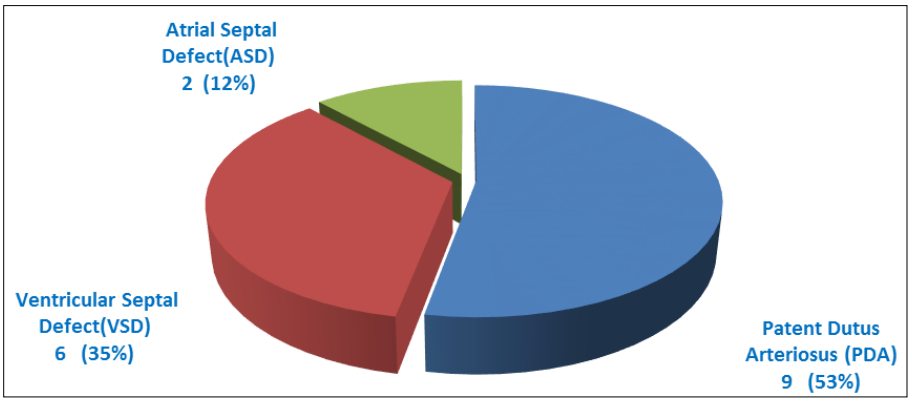

Fig 2 (b): Acyanotic Congenital Heart Disease (CCHD) (n=17)

Gastrointestinal Defects

Amongst GIT birth defects, small intestinal atresia was the commonest i.e. 29.2%, followed by Tracheo-esophageal Fistula i.e. 22.0 %. Anal Atresia was observed in 19.5% and Diaphragmatic Hernia was observed in 14.6%. {Figure-2(c)}.

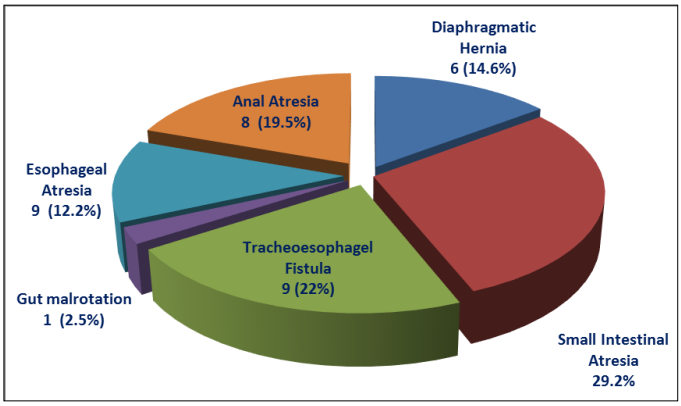

Fig 2 (C): Gastrointestinal Defects {GIT} (n=41)

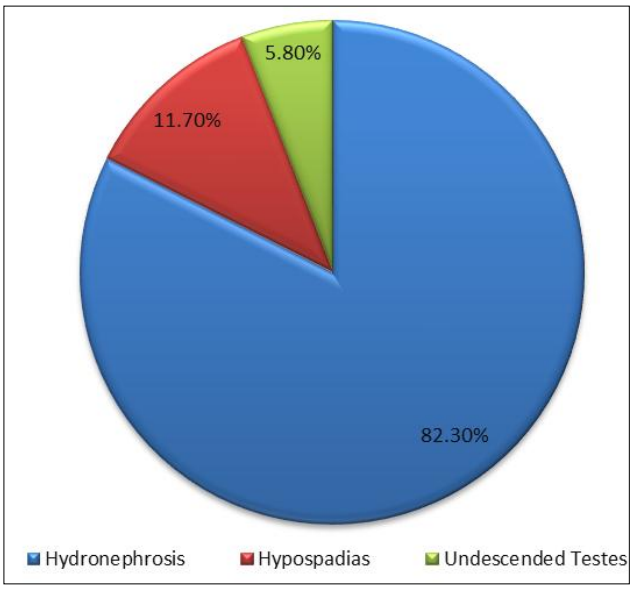

Fig 2 (D): Genitourinary Defects {GUS} (n=21)

Genito-urinary defect

The commonest genitourinary system (GUS) birth defect was hydronephrosis i.e. 66.7%, followed by ambiguous genitalia in 19.0%. Undescended testes (bilateral) was observed in 4.8%. {Figure 2(d)}

Musculo- Skeletal Defects

Amongst musculoskeletal defects, body wall defects and reduction deformity of upper/ lower limb were the commonest i.e. 31.8 % each, followed by CTEV i.e. 22.7%. {Figure 2(e)}

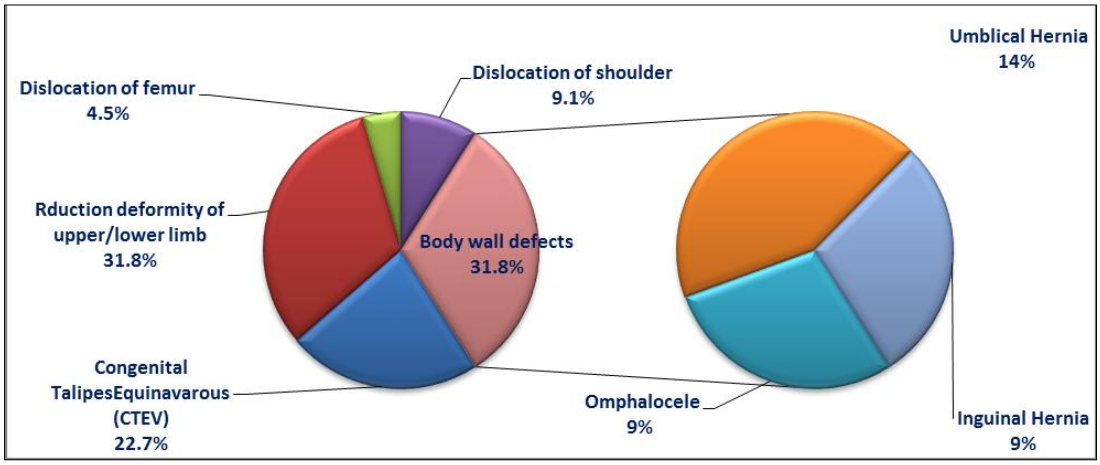

Fig 2(e): Musculo Skeletal Defects (MSS) (n=22)

Chromosomal, Minor and Other Birth Defects

Chromosomal birth defect (Down syndrome) was observed in 2.06 % cases. Other defects were observed in 4.1% cases (n=12). {Figure 2(f)} as follows:

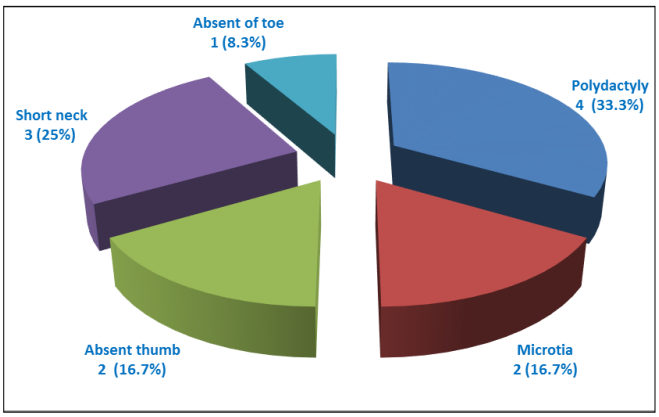

Fig 2(f): Other Defects

Discussion

The present study was designed to study the incidence and spectrum of birth defects in the Department of Obstetrics and Gynaecology, in collaboration with the Department of Pediatrics, VMMC and Safdarjung Hospital. It also aimed to study the associated maternal risk factors in these cases. The prevalence of 1.13% observed in the present study is comparable to that reported by Indian studies by Taksande *et al* and Gupta *et al* i.e. 1.91% and 1.53% respectively [1, 2]; only one study by Dutta *et al* has reported a relatively lower value i.e.0.08% [9]. Comparatively higher rates of birth defects have been reported by studies from Pakistan, Nigeria and Multan, i.e. 4.1%, 2.8% and 2.9% respectively [3, 4, 10]. The variation in observation is due to a number of factors i.e. the type of study. Some studies have been conducted in limited hospitals based settings, while others are community based. The presently observed rate of 1.13% does not reflect the picture of the general population, because this was a purely hospital based study, with no attempt whatsoever to obtain a sample that would be representative of the general population. Hospital based studies do not take into account, infants born at other centers, hence prevalence estimates based on hospital data are not a true estimate of the condition among a population.

Prevalence of birth defects among live birth and still births

The present study observed a significantly higher prevalence of birth defects among still births as compared to live births, i.e. 4.55% and 1.04% respectively, p <0.0001. Similar observations have been reported by Taksande *et al*, Taboo *et al* and Raza *et al* who reported higher rates of birth defects among still births as compared to live births [2, 4, 11] as per the observations of Taksande *et al*, the association was statistically significant, (p<0.01), similar to our observation [2]. This significant difference in fetal outcome, emphasizes the adverse impact of birth defects on perinatal mortality.

Prevalence of birth defects in male and female gender

The present study observed that a higher proportion of males were affected by birth defects as compared to females i.e. 56.6% and 43.4% respectively, (4 babies had ambiguous genitalia hence not included), giving a male: female ratio of 1.30:1. These observations are in concordance with those of Mohanti *et al* and Raza *et al*, who reported male preponderance amongst congenitally malformed babies [4, 6]. Naoom *et al* also reported that 9.2% of male babies had congenital anomalies, while only 2.4% females had birth defects, with a male: female ratio of 2:1 [13]. On the contrary, Jehangir *et al* and Taksande *et al* did not observe any significant role of gender in the occurrence of birth defects [10, 2] while Tabboo *et al* reported that fetal anomalies had a predilection for female gender with a male to female ratio 1:09 [12].

Spectrum of birth defects

The present study observed that birth defects involving the CNS were the commonest i.e. 36.2%, followed by Orofacial defects (cleft palate and/cleft lip) i.e. 18.6% and GIT 14.1%, in descending order. These observations are exactly similar to those of Obu *et al* and Jehangir *et al*, who reported CNS and Orofacial clefts to be the commonest birth defects [10, 3]. Studies by Gupta *et al*, Rad *et al* and Taboo *et al* also observed CNS anomalies to be the commonest of all birth defects [1, 7, 12] However, our observations differ from those of Taksande *et al*, Naoom *et al* and Ali *et al*, who reported CVS anomalies to be the most frequently observed, and from Raza *et al*, who found urogenital birth defects to be the commonest [2, 4, 5, 14].

The present study observed that cleft palate and cleft lip were the second most common birth defects. The high prevalence of these Orofacial clefts (OFCs), is comparable with the reports of Obu *et al* and Ali *et al* who observed them to be amongst the commonest congenital anomalies [3, 5] Dutta *et al* also reported a high incidence (17.4%) of facial defects [8].

The present study observed that the birth defects of the gastrointestinal tract were the third most common i.e. 14.13%.

Our observations are similar to those of Gupta *et al* and Jehangir *et al* who reported GIT birth defects to be third, in order of frequency [1, 10]. According to an Indian three-centre study also, GIT malformations were the third most common cause of congenital birth defects, and the incidence was 38.37 per 10,000 births [2] However, Raza *et al* and Taksande *et al* found GIT system to be less involved amongst birth defects [2, 4].

The relatively lower prevalence of birth defects of CVS observed in the present study, in probably because, antenatal fetal echo is not done regularly in our institution, even in high risk cases due to financial constraints. Taksande *et al* reported CVS anomalies to be the commonest, probably because regular fetal echo of high risk mothers is being done in their institution [2]. Another reason for the low prevalence estimate, is that most babies are discharged in <48 hours after birth. By this time, acyanotic congenital heart disease may not be detected because the neonate may remain asymptomatic. It is only after 4-6 weeks after birth, that the baby becomes symptomatic and develops signs and symptoms of heart failure, that the diagnosis is established.

Role of Obstetrician in Prevention of Birth Defects: For prevention of birth defects, the obstetrician has great responsibility than other medical discipline. As primary physician to women, obstetrician is the women's counselor during the preconceptional and interconceptional period. Many harmful intrauterine factors are risky in early pregnancy e.g. drugs, irradiation and viral infections. It is the obstetrician who advices and refers the women, as and when required, to higher specialized centre and for genetic counseling, if needed. He also advices the women regarding prevention of premature births, and the significance cause of better nutrition, improved healthcare and regular antenatal supervision. As such the treating obstetrician is the coordinator of various other relevant disciplines including pediatrician, physiotherapist, medicine and endocrinologist which are important for prevention of birth defects. In the vast relation of environmental factors and their relation to birth defects, even a greater opportunity how avails the obstetrician.

References

1. Gupta S, Gupta P, Soni JS. A study of incidence of various systemic congenital Malformations and their association with maternal factors. National Journal of Medical Research. 2012; 2(1). ISSN 2249-4995.
2. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in Central India: A rural medical college hospital based data. Indian J Hum Genet. 2010; 16(3):159-163.
3. Obu HA, Chinawa JM, Uleanya MD, Adimora GN, Obi IE. Congenital malformations among new borns admitted in a neonatal unit of a tertiary hospital in Enugu, South-East Nigeria-a retrospective study. BMC Research Notes. 2012; 10(5):177. doi: 10.1186/1756-0500-5-177.
4. Raza MZ, Sheikh A, Ahmed SS, Ali S, Naqvi Sma. Risk factors associated with birth defects at a tertiary care center in Pakistan. Italian Jourjnal Peadiatrics 2012, 38: 68 doi: 10.1186/1824-7288-38-68.
5. Ali WH AB, Balaha MH, Moghannum MSA, Hashim I. Risk factors and prevalence of birth defects and inborn error of metabolism in Al Ahsa, Saudi Arabia. Pan African Medical Journal. 2011; 8:14.
6. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G. Congnital malformation in newborn: A study of 10,874 consecutive births. J Anat Soc India. 1989; 38:101-111.
7. Abdi-Rad I, Khoshkalam M, Farrokh - Islamlou HR. The Prevalence at Birth of Overt congenital anomalies in Urmia, Northwestern Iran. Archives of Iranian Medicine. 2008; 11(2):148-151.
8. Dutta HK, Bhattacharya NC, Sarma JN, Giriraj K. Congenital malformations in Assam J. Indian Assoc Paedirt Surg. 2010; 15:(2):53-55.
9. Romitti PA. Utility of family history reports of major birth defects as a public health strategy. Pediatrics, 2007; 120(2):S71-7.
10. Jehangir W, Ali F, Jahangir T, Sajjad M. Prevalence of Gross Congenital Malformations at Birth in the Neonates in a Tertiary Care Hospital. A.P.M.C. 2009; 3(1):47-50.
11. Tayebi N *et al*. OMJ. 2010; 25:37-40. doi:10.5001/omj.2010.9
12. Taboo ZA-Alkader. Prevalence and Risk Factors for Congenital Anomalies in Mosul city. The Iraqi Postgraduate Medical Journal. 2012, 4.
13. Naoom MB, Alsaadi YI, Ghalib Yassin BA, Matloob HY. Congenital anomalies among new borns admitted in tertiary hospital; Iraqi experience. J Fac Med Baghdad. 2013; 55(2):106-110.
14. Mossey PA, Davies JA, Little J. Prevention of orofacial clefts: Does pregnancy planning have a role? Cleft Palate Craniofac J. 2007; 44:244-50.