

# Congenital Malformation Pattern in Duhok City

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## Abstract

This study was undertaken to estimate the prevalence of neonatal malformations in Duhok province by reviewing a database of all deliveries from January 1<sup>st</sup> 2006 to December 30<sup>th</sup> 2010 in Azadi hospital, The files of the delivery room and neonatal registry records were studied for all obvious congenital anomalies. During the five years study period a total of 79227 babies were born at Azadi University hospital of which 1760 were stillbirth and 369 were surviving babies who had one or more congenital anomaly giving an incidence of 4.65/1000 live birth. The most involved system was the Central Nervous System forming nearly half of the entire defects (44.98%) with an incidence of 2.09/1000 birth, Hydrocephalus, Myelomeningocele and microcephaly together were the most prevalent CNS anomalies among all reported congenital malformations, followed by congenital heart diseases (0.65 /1000) with ventricular septal defect (VSD) being the most common lesion. The proportion of live born infants with chromosomal anomalies was 32 cases, contributing 8.67% of all malformations. Down syndrome was the leading defect followed by Turner syndrome, while musculoskeletal, genitourinary and unclassified multiple system anomalies scored the incidence of (0.26/1000), (0.25/1000) and (0.35/1000) respectively. The less common defects were sensory (0.21/1000), gastrointestinal (0.17/1000), teratoma (0.15/1000) and respiratory (0.08/10000).

**Keywords:** Genetic disorders, Birth defects prevalence, Genetic counseling.

## 1.0 Introduction

Congenital malformations or birth defects are common among all races, cultures, It is an important cause of childhood death, chronic illness, and disability in many countries (Behrman *et al.*, 2004). Congenital malformations may be defined in terms of physical structure as an abnormality of physical structure or form usually found at birth or during the first few weeks of life (Hudgins and Cassidy, 2006). Congenital abnormalities are caused by problems during the fetus's development before birth, such as those of single-gene abnormalities, when one or more genes doesn't work properly or part of a gene is missing or problems with chromosomes, such as having an extra chromosome or missing part of a chromosome. Sometimes no simple mode of inheritance can be established and no teratogenic agents obvious. In such cases, abnormal development may be a result of failure of gene control, failure of cellular and tissue interactions or local environmental effects on gene expression during critical stages of the pregnancy (e.g. Spina bifida and cleft lip and palate. (Khoury and Gruss, 1983).

The treatment and rehabilitation of children with birth defects is usually costly and complete recovery is usually impossible (Petrini *et al.*, 1997) hence it is obligatory to find out causative and risk factors for birth defects and prevent them earlier. (Wald *et al.*, 1996).

However many researchers suggested that germ line mutations, paternal food deprivation, chemical mutagens, alcohol use, age, smoking habits and epigenetic alterations can affect birth outcomes (Abel, 2004; De Santis *et al.*, 2008; Sartorius and Nieschlag, 2009; Anderson *et al.*, 2014).

Although there have been different studies focused on birth anomalies problems in different parts of the world as well as in Iraq, To our knowledge, there was no previous studies regarding the prevalence of birth defects in Duhok, thus the aim of this study is a trial to identify the incidence, types of birth defects recorded over five years at birth among babies delivered at Azadi teaching hospital in Duhok.

## 2.0 Maternal and methods

This descriptive, hospital-based survey was undertaken in Azadi teaching hospital in Dohuk city. Azadi hospital contains the main maternity and obstetric unit in the region which receives most of the referral cases from rural areas. Approval by the hospital administration has obtained according to the document dated 21/09/2011 and numbered 327, under the supervision of a pediatrician in that hospital. The survey period was between January 1<sup>st</sup> 2006 to December 30<sup>th</sup> 2010. Data on 79227 births were collected during the study period. All neonates were included in the study (full term, preterm), dead fetuses were excluded. The cases were recorded from the files of the delivery room and neonatal registry records after they were approved by the pediatrician.

## 3.0 Result

During the five year study period, there was a total of 79227 deliveries, about 1760 were in a Stillbirth and about 369 were surviving babies who had one or more congenital anomaly giving an incidence of 4.65/1000 live

births, as can be seen in **Table 1**.

**Table1:** The annual number and percentage of birth, stillbirths and malformed live babies recorded in Azadi hospital over five years.

Year	Birth	Still birth / Percentage	Malformed /Percentage
2006	13518	246(1.81%)	78(0.57%)
2007	15326	300(1.92%)	67(0.43%)
2008	16667	405(2.42%)	71(0.42%)
2009	17839	402(2.25%)	80(0.44%)
2010	15877	407(2.56%)	73(0.45%)
<b>Total</b>	<b>79227</b>	<b>1760(2.22%)</b>	<b>369(0.46%)</b>

The number of live newborns with different birth defects in each year is shown in **Table 2**.

**Table2:** The number of live newborns with different types of malformations.

Types of malformations	2006	2007	2008	2009	2010	Total
<b>Central Nervous System</b> Hydrocephaly, Myelomeningocele , Microcephaly, Facial plusy, Arnold chiari syndrome.	39	35	30	33	29	166
<b>Congenital heart diseases</b> Ventricular septal defect (VSD) ,Cardiomegaly ,atrial septal defect (ASD),	9	10	10	11	12	52
<b>Chromosomal</b> Downs, Turner, Edward	6	6	6	8	6	32
<b>Musculoskeletal system</b> Limb deformities,Arythrogryposis, Osteogenesis imperfect,Chondrodysplasia.	4	3	5	5	4	21
<b>Genitourinary system</b> Posterior urethral valve,polycystic kidney,poter syndrome ,hydronephrosis,ambiguous genitalia	4	3	4	5	4	20
<b>Sensory</b> Icthosis,ectodermal dysplasia, Epidermolysis bullosa ,dysmorphic features	4	2	4	3	4	17
<b>Gastrointestinal system</b> Omphalocele,Jejunal atresia,Tracheaesophageal fistula, Cleft lip and/or palate,imperforated anus Diaphragmatic hernia	3	2	2	4	3	14
<b>Teratoma</b> Cystic hygroma Sacroccocygial teratoma	2	2	2	3	3	12
<b>Respiratory system</b> Congenital pneumonia,Choanal atresia	2	0	2	2	1	7
<b>Un classified multiple system anomalies</b>	5	4	6	6	7	28

The congenitally malformed neonates were diagnosed and classified according to the affected system using WHO classification (13). The most involved system was the Central nervous system with 166 cases

forming 44.98% of all cases giving an incidence of 2.09/1000 live births, followed by Congenital heart defects (n=52) 14.09% , Chromosome defects (n=32) 8.67% , Musculoskeletal (n=21) 5.6%, genitourinary system defects (n=20) 5.42% and multiple malformation syndrome (n=28) 7.6%. The less common defects included Sensory (skin, eye, ear, and face) (17) gastrointestinal system defects (n=14). Teratoma (n=12) Respiratory system (n=7). The annual incidences of malformations are shown in **Table 3**.

**Table 3-The annual rate of malformations per 1000 total birth estimated soon after birth.**

	2006	2007	2008	2009	2010	The average rate
<b>Birth number</b>	13518	15326	16667	17839	15877	
<b>Central nervous system</b>	2.88	2.28	1.79	1.84	1.82	2.09
<b>Congenital heart diseases</b>	0.66	0.65	0.59	0.61	0.75	0.65
<b>Chromosomal</b>	0.44	0.39	0.35	0.44	0.37	0.40
<b>Musculoskeletal system</b>	0.29	0.19	0.29	0.28	0.25	0.26
<b>Genitourinary system</b>	0.29	0.19	0.23	0.28	0.25	0.25
<b>Sensory</b>	0.29	0.13	0.23	0.16	0.25	0.21
<b>Gastrointestinal system</b>	0.22	0.13	0.11	0.22	0.18	0.17
<b>Teratoma</b>	0.14	0.13	0.11	0.16	0.18	0.15
<b>Respiratory system</b>	0.14	0	0.11	0.11	0.06	0.08
<b>Un classified multiple system anomalies</b>	0.36	0.26	0.35	0.33	0.44	0.35

#### 4.0 Discussion

The prevalence of congenital anomalies at birth in developed countries is reported to be between 3-5% (Park, 2005) ,the overall prevalence of congenital anomalies for Duhok city in this study was found to be 4.65 per 1000 births. ). if stillbirths were included, an autopsy back-up was available; our incidence would have been higher than that obtained in the series, because it is well known that higher rate of malformations present in stillbirths and neonatal deaths than live births (Halevi,1967).This prevalence was higher than that reported from Turkey for major congenital malformations 2.9/1000 births between 2000 to 2004 (Tomatir *et al.*,2009) and was lower than that reported in Basrah in 1998 (7.76 per 1000 births) (Al-Sadoon *et al.* , 1999 ), from Baghdad, Iraq in 2007 (12.36 per 1000 births ) (Hameed,2007) and also lower than that reported previously in Iran (16.55 per 1000 total births which increased from 10.46 in 2000 to 17.01 per 1000 births in 2004) (Dastgiri *et al.* ,2007) in United Arab Emirates (10.5 per 1000 live births during 1992–1994),( Al-Gazali *et al.*,1995), Bahrain (18.75 per 1000 live births in 1985) (Hammamy and Al-Awan,1994) .

The result of this survey shows that the most common types of malformations were central nervous system (CNS), congenital heart diseases (CHD), and chromosome abnormality syndromes respectively. Central nervous system anomalies ranked the first, forming nearly half of the entire defects (44.98%) with an incidence of 2.09/1000 birth. Hydrocephalus, Myelomeningocele and microcephaly together were the most prevalent CNS anomalies among all reported congenital CNS malformations. This rate is higher than incidence of CNS in other reports from Saudi Arabia (Fida *et al.* ,2007) and lower than that reported in Iran (Golalipour *et al.* ,2005).In many other international studies CNS defects were also the most common type of anomalies (De Galan-Roosen

*et al.*, 1998; Mir *et al.*, 1992; Tunçbilek *et al.*, 1999).

In Al-Anbar governorate it was found that 33 infants were delivered with (NTDs) giving an incidence of 3.3/1000 births in 2007 to 2008, most were of myelomeningocele and anencephaly types. Two-thirds of the cases found were from consanguineous marriages. (Al-Ani *et al.*, 2010). Similar rates were reported in Basrah for the period 1999–2000 (4.35 per 1000 births) (Al-Sadoon *et al.*, 1999), and Erbil (4.48 per 1000 live births) (Al-Rabbaty, 2001) also a similar rate of neural tube defects (NTDs) (4.7 per 1000 live births) was reported from Duhok, Iraq (Abdurrahman, 2007), higher rates of NTDs were reported in Baghdad (5.95 per 1000 births) (Hameed, 2007) and Diwaniyah (8.4 per 1000 total births in 2000) of Iraq (Al-Shammosy, 2002).

Many reports indicated that Maternal nutrition are very important factors which contribute to neural tube defects (Stein and Susser, 1997; Sheffer *et al.*, 1993) Much attention has been focused recently on the role of peri-conceptional folic acid supplementation during pregnancy for the primary prevention of birth defects particularly NTDs (Stevenson *et al.*, 2000; Willams *et al.*, 2005). Another possible explanation for the apparent higher percentage of these types of defects may be because they are obvious at birth and are recorded more carefully than other defects (Goynumer *et al.*, 2005).

In this study Congenital heart defects (CHD), were the second major malformation (14.09%). and VSD is found to be the most common Acyanotic CHD lesion, this is consistent with that reported by (Mustafa, 2012), While in Al-anbar city in 2012 it has been found that cardiovascular system was the most affected, followed by genito-urinary system. (Al-Ani *et al.*, 2012) This may be explained by the difference in genetic makeup and ethnicity, also Consanguinity proved to be a risk factor for CHD (Al-Ani, 2010)

According to the current report the proportion of live born infants with chromosomal anomalies was 8.67% of all cases (n=32). Down syndrome was the leading autosomal aneuploidy chromosome defect followed by Turner syndrome. this may accuse to Maternal age which is strongly associated with chromosomal anomalies and the rising proportion of older mothers is likely to contribute to increase in prevalence of anomalies (Kalter, 1983) due to the increase of aneuploidy with advancing maternal age.

In this study the incidence of Musculoskeletal malformations (0.26 per 1000 birth), genitourinary defects (0.25 per 1000 birth), and Gastrointestinal defects (0.17 per 1000 birth) were less than that reported in Baghdad, the incidence, for the Musculoskeletal malformations was 1.61 / 1000 live births in 2002 (Hameed, 2007), In Bahrain, the musculoskeletal system had the highest frequency at an average of 2.8 per 1000 births (Hammamy and Al-Awan, 1994), In Turkey for the anomalies of urogenital system was (2.1 per 1000 births) (Himmotoglu *et al.*, 1996), in Al-Anbar, the most common system involved was the cardiovascular system followed by genito-urinary system among total birth between 2010-2011 (Ali *et al.*, 2012), also in an Indian study, the Musculoskeletal malformations were the most common in live births followed by gastrointestinal and CNS defects (Datta and chatumedia, 2000).

In fact, The pattern and prevalence of congenital anomalies may vary over time or geographical location, thereby reflecting a complex interaction of known and unknown genetic and environmental factors, These variations may be explained by social, racial, ecologic, and economic influences (Temtamy *et al.*, 1998; Biri *et al.*, 2005). It seems that the Interaction of genetic and environmental mutagens, physical agents and infections in addition to the high rate of consanguineous marriages in our population might be an important cause of infant malformations. (Hanan *et al.*, 1989).

## 5.0 Conclusion

In Iraqi pediatric hospitals and clinics including Azadi university teaching hospital in duhok city, there is an obvious shortage in health care strategies, due to the absence of any nationwide birth defects monitoring, associated with having neither a genetic service network, genetic counseling for preventive health services, nor properly trained health care personnel to provide these services, which reflect a negative effect on the incidence of birth defects. To eliminate all of these deficiencies, various national and institutional steps need to be taken to consider possible preventive medical care.

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