第 56 卷 第 4 期 2021 年 8 月 JOURNAL OF SOUTHWEST JIAOTONG UNIVERSITY Vol. 56 No. 4 Aug. 2021

ISSN: 0258-2724

DOI : 10.35741/issn.0258-2724.56.4.49

Research article

Medicine

# IDENTIFICATION OF MOST COMMON CONGENITAL ABNORMALITY TYPES AMONG NEWBORN INFANTS: A HOSPITAL-BASED STUDY

识别新生儿中最常见的先天性异常类型:一项基于医院的研究

Manaf Authman Hreeth <sup>a</sup>, Omer Q. B. Allela <sup>b</sup>, <sup>\*</sup>, Imad Jebur Rashid <sup>c</sup> <sup>a</sup> Pediatrics Central Teaching Hospital Baghdad, Iraq <sup>b</sup> Alnoor University College Bartela, Iraq, <u>omerallela@alnoor.edu.iq</u> <sup>c</sup> Iraqi Centre for Cardiac Disease Hospital Baghdad, Iraq

Received: May 13, 2021 • Review: June 11, 2021 • Accepted: July 16, 2021 • Published: August 30, 2021

*This article is an open-access article distributed under the terms and conditions of the Creative Commons Attribution License* (<u>http://creativecommons.org/licenses/by/4.0</u>)

#### Abstract

The term "congenital abnormalities" signifies a disruption in the normal process of organogenesis occurring before birth: the earlier the insult, the grosser the abnormality. This research is the largest study aimed at identifying the most common congenital abnormality types among newborn infants in the neonatal care unit (NCU) of the Al-Kadhymia teaching hospital, Baghdad, Iraq. This prospective study was carried out during the period from February 1 to August 1, 2011. A total of 2700 neonates were admitted to the NCU, and 100 newborn infants in the nursery care unit were proven to have congenital abnormalities by physical examination alone. The questionnaire for neonatal evaluation included: gestational age, sex, body weight, and type of congenital anomaly. The results showed that of the total (100) affected neonates, 63 (63%) were full term, 55% had neurological abnormalities, followed by 12% with cleft lip and palate and then 11% with chromosomal abnormalities (most of them had Down syndrome, only 3 cases had Edward syndrome, and 1 case had Patau syndrome). It can be concluded that most of the affected newborns were full term, with a slight male predominance. The incidence of neurological abnormalities was higher than other types of birth defects.

Keywords: Congenital Abnormalities, Iraq, Newborn Infants

**摘要** 术语"先天性异常"表示出生前发生的正常器官发生过程的中断:损伤越早,异常越严重。 这项研究是规模最大的研究,旨在确定伊拉克巴格达 Al-Kadhymia 教学医院新生儿护理室(中央 控制单元)新生儿中最常见的先天性异常类型。这项前瞻性研究于 2011 年 2 月 1 日至 8 月 1 日期 间进行。中央控制单元共收治 2700 名新生儿,仅通过体检就证实 100 名托儿所新生儿患有先天性 畸形。新生儿评估问卷包括:胎龄、性别、体重和先天异常类型。结果显示,在受累新生儿总数 (100 名)中,足月 63 名 (63%),神经系统异常 55%,唇裂和腭裂 12%,染色体异常 11% (多 数为唐氏)综合征,仅 3 例为 Edward 综合征,1 例为帕陶综合征)。可以得出结论,大多数受 影响的新生儿都是足月,男性略占优势。神经系统异常的发生率高于其他类型的出生缺陷。

关键词: 先天性异常, 伊拉克, 新生儿

### I. INTRODUCTION

There are two types of congenital abnormalities, the major congenital anomaly, which is a structural abnormality present at birth that has a significant effect on function or social acceptability, e.g., cleft lip, and the minor congenital anomaly, which is a structural abnormality present at birth that has minimal effect on clinical functions, but may have a cosmetic impact, e.g., preauricular pit [1].

Congenital malformations or birth defects are common among all races, cultures, and socioeconomic strata. Congenital disabilities can be isolated abnormalities or part of a syndrome and continue to be an important cause of neonatal and infant morbidity. Based on a World Health Organization (WHO) report, about 3 million infants are born each year with major congenital malformations; congenital malformations accounted for an estimated 495,000 deaths worldwide in 2005 [2].

Regarding etiology, congenital abnormalities can result from monogenic, chromosomal, maternal infections, maternal illness, twinning, environmental agents, medication, nutritional and unknown etiologies [3].

Congenital anomalies can be classified either based on the timing of the insult, underlying histological changes, or their medical and social consequences.

Congenital anomalies based on insult can be placed into the following three categories: malformation, disruption, and deformation. Classification based on underlying histological changes includes aplasia, hypoplasia, hyperplasia, and dysplasia [4].

Regarding managing congenital abnormalities, newborns with one or more malformations should receive ongoing care and may require multidisciplinary care and case management. Some clinical problems or physical findings may evolve over time and become more apparent with age [5, 6].

### **II. PATIENTS AND METHODS**

#### **A. Sample Population**

This prospective study was carried out in the Al-Kadhymia teaching hospital (Neonatal care unit (NCU)) from 1<sup>st</sup> February to 1<sup>st</sup> August 2011.

A total of 2700 neonates were admitted to the NCU. The inclusion criteria include all children with a congenital abnormality. One hundred newborn infants in the nursery care unit were proven to have congenital abnormalities by physical examination alone. The questionnaire for neonatal evaluation included: gestational age, sex, body weight, and type of congenital anomaly.

#### **B. Study Hypotheses**

1. Child demographic variables are associated with a congenital abnormality.

2. More than half of babies have Full-term babies.

3. Neurological disorder has a high percentage among the type of congenital anomalies.

#### C. Statistical Analysis

The Statistics Package for Social Science (SPSS) version 17 was used for data analysis. The results are expressed in numbers, percentages, and Chi-square Pearson correlation, which was statistically significant at a P value of less than 0.05 and statistically not significant at a P-value of more than 0.05.

#### **III. RESULTS**

Of the total (100) affected neonates, 63 (63%) were full-term compared with 37 (37%) preterm babies (<37 weeks gestation), as shown in Table 1, with a significant difference (P <0.05).

Table 1. Gestational age of newborn babies

Gestational age	Number	(%)
Preterm babies	37	37%

Full-term babies	63	63%
Total	100	100%

Table 2 shows that, out of the 100 neonates who were proven to have congenital anomalies, 55 (55%) were male, and 45 (45%) were female, with no significant difference (P > 0.05). The male to female ratio was 1.3:1.

Table 2.

Gender distribution of neonates with congenital abnormalities

Gender	Number	(%)
Male	55	55%
Female	45	45%
Total	100	100%

Newborns with congenital abnormalities had an average weight of 3 kg, ranging from <1 kg to 3.5 kg. Of the total (100) cases, only 2 (2%) cases had body weight <1 kg, with the highest occurrence of congenital abnormalities in newborns with bodyweight >2.5 kg (56 cases, 56%), with significant difference (P <0.05), as shown in Table 3.

Table 3.

Distribution of body weight in newborns with congenital anomalies

Bodyweight	Number	(%)
<1 kg	2	2%
1-2.5 kg	42	42%
>2.5 kg	56	56%
Total	100	100%

Results showed that 55% were diagnosed with neurological abnormalities, followed by 12% with cleft lip and palate and then 11% with chromosomal abnormalities (most of which were Down syndrome, with only 3 cases of Edward syndrome and 1 case of Patau syndrome), with significant difference (P <0.05), as shown in Table 4.

Table 4.

Distribution of newborns according to the type of congenital anomalies

Types of anomalies	(%)
Neurological (mainly neural tube defects)	55
Cleft lip & palate	12
Chromosomal	11
Cardiovascular system (C.V.S)	6
Musculoskeletal system	5
Alimentary system	5
Genitourinary system (GUS)	5
Respiratory system	1

## **IV. DISCUSSION**

Our results indicate that the prevalence of congenital malformation disorders in the NCU of

the Al-Kadhymia teaching hospital was 3.7% (100/2700), where the number of males was higher than the number of females, and the male to female ratio was 1.3:1.

This finding agrees with a recent study performed in Iran in June 2008, where 2.9% of live births had major congenital abnormalities [7], while another study in Kuwait documented 1.25% of children with congenital abnormalities [8]. Considerable variation in frequency has been reported in different populations, from as low as 1.07% in Japan [9] to as high as 4.3% in Taiwan [10]. This wide variability could be due to genetic differences in these populations.

In addition, in this study, out of 100 newborn infants with congenital abnormalities, 63% were full-term, with bodyweight ranging between 2.5-3.5 kg, and only 37% were preterm babies, with bodyweight ranging between 1-2.5 kg, whereas a slight increment in incidence was found in preterm and small for gestational age infants in studies carried out in the U.K. [11] and India [12]. Such results could be attributed to the absence of facilities for antenatal diagnosis of such anomalies in our country, leading to the delivery of full-term babies with such anomalies.

Moreover, this study showed that the most common anomalies were neurological (55%), followed by cleft lip and/or palate (12%), and chromosomal abnormalities (11%); among the neurological anomalies, neural tube defects were the most common.

Different observations were recorded in other studies. A study in India [13,14] revealed an increase in the frequency of musculoskeletal anomalies (30%), neurological anomalies (20.5%), and cleft lip and palate (18.5%), while other studies in Iran and Tunis [15,16] showed a higher incidence of cleft lip and palate. A study in Saudi Arabia [17] reported the major anomalies as genitourinary (25%), cardiovascular (15%), and neurological (10%).

This data partly implies the poor compliance of pregnant women regarding the intake of folic acid and poor antenatal care in terms of screening for such anomalies.

The neurological anomalies in this study accounted for 55% of cases, while the number of such cases was 1% in Wales [18] and 2% in Germany [19]. This difference could be related to the inadequate education of our people concerning supplementation of folic acid during pregnancy and poor antenatal care; meanwhile, in Wales and Germany, there are facilities for prenatal diagnosis and interruption of an affected pregnancy. The chromosomal abnormalities in this study accounted for 11% of cases, which is similar to the study in the United Kingdom but higher than a study in Norway, which had 0.1% of such cases [20].

## REFERENCES

[1] MYRIANTHOPOULOS, N.C. & CHUNG, C.S. (1974) Congenital malformations in singletons: epidemiologic survey. Report from the Collaborative Perinatal Project. *Birth Defects Original Article Series*, 10(11), pp. 1–58.

[2] LEPPIG, K.A., WERLER, M.M., CANN, C.I., COOK, C.A., & HOLMES, L.B. (1987) Predictive value of minor anomalies. I. Association with major malformations. *The Journal of Pediatrics*, 110(4), pp. 531–537.

[3] WYNSHAW-BORIS, A. & BIESECKER, L.G. (2008) Dysmorphology. In KLIEGMAN, R.M., BEHRMAN, R.E., JENSON, H.D., STANTON, B.F. (Eds.) Nelson textbook of Pediatrics. (18th ed) Philadelphia: Elsevier Saunders Ch. 108: pp. 788.

[4] KUMAR, P. & BURTON, B.K. Congenital malformation evidence-based evaluation and management, 15<sup>th</sup> ed., McGraw-Hill Companies; Ch. 1:7.

[5] AMERICAN COLLEGE OF MEDICAL GENETICS FOUNDATION. (2003) Definition and evaluation of the newborn with single or multiple congenital anomalies.

http://www.health.state.ny.us/nysdoh/dpprd/e xec.htm

[6] PORT, K.E., MOUNTAIN, H., NELSON, J. & BITTLES, A.H. (2005) Changing profile of couples seeking genetic counseling for consanguinity in Australia. *American Journal of Medical Genetics. Part A*, 132 A, pp. 159-163.

[7] MADI, S.A., AL-NAGGAR, R.L., AL-AWADI, S.A. &, BASTAKI, L.A. (2005) Profile of major congenital malformations in neonates in Al-Jahra Region of Kuwait. *Eastern Mediterranean Health Journal*, 11, pp.700-706.

[8] IMAIZUMI, Y., YAMAMURA, H., NISHIKAWA, M., MATSUOKA, M. & MORIYAMA, I. (1991) The prevalence at birth of congenital malformations at a maternity hospital in Osaka city, 1948 – 2003. *Jinrui Idengaku Zasshi (The Japanese Journal of Human Genetics)*, 36, pp. 275-287. [9] CHEN, C.J., WANG, C.J., YU, M.W., & LEE, T.K. (2004) Perinatal mortality and prevalence of major congenital malformations of twins in Taipei city, Taiwan. *Acta Geneticae Medicae et Gemellologiae*, 41, pp. 197-203.

[10] ROIZEN, N.J. & PATTERSON, D. (2003) Congenital abnormalities: occurrence and outcome in the U.K. *Lancet*, 361, pp. 1281-1289.

[11] SINGH, A. & GUPTA, R.K. (2009) Pattern of congenital anomalies in newborn: a hospital based prospective study. *JK Science: Journal of Medical Education & Research*, 11(1), pp. 34-36.

[12] GROVER, N. (2000) Congenital malformations in Shimla. *Indian Journal of Pediatrics*, 67(4), pp. 249-51.

[13] DATTA, V. & CHATURVEDI, P. (2000) Congenital malformations in rural Maharastra. *Indian Journal of Pediatrics*, 37, pp. 988-1001.

[14] KHATEMI, F. & MAMORRI, G.A. (2005) Survey of congenital major malformations in 10,000 newborns. *Iranian Journal of Pediatrics*, 15, pp. 315-20.

[15] KHROUF, N. (1996) Malformation in 10,000 consecutive births in Tunis. *Acta Pediatrica Scandinavia*, 75, pp. 534-39.

[16] HASSIB, N. & NAJI, K. (2005) Congenital malformations: Are they more prevalent in populations with a high incidence of consanguineous marriages? *Annals of Saudi Medicine*, 17(2), pp. 254– 256.

[17] MORRIS, J.K., & WALD, N.J. (1999) Quantifying the decline in the birth prevalence of neural tube defects in England and Wales. *Journal of Medical Screening*, 6, pp. 182.

[18] KLUSMANN, A., HEINRICH, B., STOPLER, H. et al. (2005) A decreasing rate of neural tube defects following the recommendations for periconceptional folic acid supplementation. *Acta Paediatrica*, 94, article ID 1538.

[19] KAZAURA, M.R., & LIE, R.T. (2002) Down's syndrome and paternal age in Norway. Paediatric and Perinatal Epidemiology, 16(4), pp. 314–319.
[20] CHATURVEDI, P. & BANERJEE, K.S. (1993) An epidemiological study of congenital malformations in newborn. Indian Journal of Pediatrics, 60 (5), pp. 645-655.

## 参考文:

[1] MYRIANTHOPOULOS, N.C. 和 CHUNG, C.S. (1974) 单胎先天性畸形:流 行病学调查。来自协作围产期项目的报告 。出生缺陷原始文章系列,10(11),第 1-58页。

[2] LEPPIG, K.A., WERLER, M.M., CANN, C.I., COOK, C.A., 和 HOLMES, L.B. (1987) 轻微异常的预测值。一世. 与重大畸形的 关联。儿科杂志, 110(4), 第 531-537 页 。

[3] WYNSHAW-BORIS, A. 和
BIESECKER, L.G. (2008) 形态学。在
KLIEGMAN, R.M., BEHRMAN, R.E.,
JENSON, H.D., SANTON, B.F. (编辑) 纳尔
逊儿科教科书中。(第 18 版)费城:爱
思唯尔桑德斯(第 108:第 788 页。

[4] KUMAR, P. 和 BURTON, B.K. 先天性 畸形循证评估和管理, 第 15 版,麦格劳-希尔公司;通道 1:7。

[5] 美国医学遗传学基金会。(2003) 新生 儿单发或多发先天性异常的定义和评估。

http://www.health.state.ny.us/nysdoh/dpprd/e xec.htm

[6] PORT, K.E., MOUNTAIN, H., NELSON, J. 和 BITTLES, A.H. (2005) 在澳 大利亚寻求血缘遗传咨询的夫妇的变化情 况。美国医学遗传学杂志。一种部分.

132一种,第159-163页。

[7] MADI, S.A., AL-NAGGAR, R.L., AL-AWADI, S.A. 和 BASTAKI, L.A. (2005) 科

威特贾赫拉地区新生儿主要先天畸形概况 。东地中海健康杂志,11,第 700-706 页 。

[8] IMAIZUMI, Y., YAMAMURA, H., NISHIKAWA, M., MATSUOKA, M. 和 MORIYAMA, I. (1991) 大阪市一家妇产医 院出生先天性畸形的患病率, 1948 – 2003.
日本人类遗传学杂志, 36,第 275-287页
[9] CHEN, C.J., WANG, C.J., YU, M.W., 和 LEE, T.K. (2004) 台湾台北市双胞胎主要 先天畸形的围产期死亡率和患病率。医学 与遗传学学报, 41,第 197-203页。

[10] ROIZEN, N.J. 和 PATTERSON, D. (2003) 先天性异常:英国柳叶刀的发生和 结果, 361, 第 1281-1289 页。

[11] SINGH, A. 和 GUPTA, R.K. (2009) 新 生儿先天性异常的模式:基于医院的前瞻 性研究。JK 科学:医学教育与研究杂志 ,11(1),第 34-36页。

[12] GROVER, N. (2000) 西姆拉的先天性 畸形。印度儿科杂志, 67(4), 第 249-51 页。

[13] DATTA, V. 和 CHATURVEDI, P. (2000) 马哈拉施特拉邦农村的先天性畸形。印度儿科杂志, 37, 第 988-1001 页。

[14] KHATEMI, F. 和 MAMORRI, G.A.(2005) 10,000 名新生儿先天性重大畸形调查。伊朗儿科杂志, 15, 第 315-20 页。

[15] KHROUF, N. (1996) 突尼斯连续10,000 例新生儿畸形。斯堪的纳维亚儿科学报,75,第 534-39页。

[16] HASSIB, N. 和 NAJI, K. (2005) 先天性 畸形:它们在近亲结婚率高的人群中是否 更为普遍?沙特医学年鉴,17(2),第 254-256页。 [17] MORRIS, J.K.,和 WALD, N.J. (1999)
量化英格兰和威尔士神经管缺陷出生率的下降。医学筛查杂志,6,第182页。
[18] KLUSMANN, A., HEINRICH, B., STOPLER, H. 等。(2005)根据围孕期补充叶酸的建议,神经管缺陷率下降。儿科学报,94,文章 ID 1538。
[19] KAZAURA, M.R.,和 LIE, R.T. (2002)

唐氏综合症和挪威的父亲年龄。儿科和围 产期流行病学,16(4),第 314-319 页。

[20] CHATURVEDI, P. 和 BANERJEE, K.S. (1993) 新生儿先天畸形的流行病学研 究。印度儿科杂志, 60 (5), 第 645-655 页

o