

## CONGENITAL MALFORMATIONS AMONG LIVE BIRTHS AT ARVAND HOSPITAL, AHWAZ, IRAN - A PROSPECTIVE STUDY

Ahmadzadeh Ali<sup>1</sup>, Safikhani Zahad<sup>2</sup>, Abdulahi Masoumeh<sup>3</sup>, Ahmadzadeh Azar<sup>4</sup>

### ABSTRACT

**Objective:** The aim of the study was to determine the profile of congenital malformations (CM) among live births at Arvand hospital, in Ahwaz city.

**Methodology:** In this prospective study all of the neonates born at Arvand hospital in Ahwaz from 2004 to 2006 were registered. Stillbirths and those who died in a few hours after birth were excluded and finally 4660 newborns were enrolled.

**Results:** Of the 4660 live births 94 (20.2/1000) had at least a CM. The predominant systems involved were musculoskeletal (7.9/1000), followed by genitourinary (7.1/1000), central nervous (2.4 /1000), digestive (1.1/1000) and chromosomal anomalies (0.9/1000).

**Conclusions:** Although the frequency of malformations in the study was approximately similar to other investigations, if we include abortions, stillbirths and if we used screening tests and genetic studies, this rate was more than 20.1/1000.

**KEY WORDS:** Congenital malformation, Anomaly, Ahwaz, Iran.

Pak J Med Sci January - March 2008 Vol. 24 No. 1 33-37

### INTRODUCTION

Congenital malformations (CM) are structural defects of prenatal origin that result from defective embryogenesis or an intrinsic abnormality

in the development process. They are found in approximately 3% of newborns.<sup>1</sup> Congenital anomalies are a common cause of medical intervention, long-term illness and death.<sup>2</sup> Despite the advances in the etiology and pathogenesis of CM, even in the USA, the infant mortality from major congenital malformations is about 22%.<sup>3</sup> The pattern and prevalence of congenital anomalies may vary over-time or with geographical location, thereby reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables.<sup>4</sup> The causes of congenital malformations are varied and few studies have evaluated the etiology of malformation in newborns.<sup>5</sup> The largest number (86%) of congenital malformations are isolated and most isolated malformations are believed to be the consequence of multi-factorial inheritance.<sup>2</sup>

Surveys on congenital malformations in Iran have been carried out in Tehran,<sup>6</sup> Mashhad<sup>7</sup> and Gorgan,<sup>8</sup> cities in the Central, Eastern and Northern areas. But, a similar study has not

1. Ahmadzadeh Ali, Department of Pediatrics, Abuzar Children's Hospital,
2. Safikhani Zahad,
3. Abdulahi Masoumeh, Department of Embryology and Anatomy, School of Medicine,
4. Ahmadzadeh Azar, Department of Obstetric and Gynecology, Emam Khominee Hospital,
- 1-4: Jondishapor University of Medical Sciences, Ahwaz - Iran.

#### Correspondence

Ahmadzadeh Ali,  
Department of Pediatrics,  
Abuzar Children's Hospital,  
Jondishapor University of Medical Sciences,  
Ahwaz - Iran.  
E-mail: dr\_ahmadzadeh\_ali@ajums.ac.ir

- \* Received for Publication: May 4, 2007
- \* Revision Received: November 20, 2007
- \* Revision Accepted: November 30, 2007

been conducted among newborns in Ahwaz, a city with different ethnic groups located in the Southwestern of Iran. The purpose of the present study was to provide the profile of congenital malformation in this area of Iran.

### METHODOLOGY

This was a prospective study which involved all babies born at Arvand hospital in Ahwaz city during three years (from April 2003 to December 2006). Arvand hospital is one of the hospitals in this city with about 1/300/000 population. All the population was Muslims and the majority of them were Arabs. The consanguinity rate is high because marriage between first cousins was common. All of live newborns delivered in this hospital during the study period were examined and screened for congenital malformations only by the author. The charts of newborns with congenital malformations were extracted. Variables recorded included demographic data: the date of birth, sex, ethnicity and maternal age as well as weight, length, head circumference and Agar scores. Ethnicity was classified in 2 groups: Arab and Fars.

The diagnosis of a congenital abnormality was based on clinical examinations or prenatal ultrasonographic data confirmed over a few weeks after birth. Further appropriate investigations such as radiography, ultrasonography, echocardiography, chromosomal analysis helped in making an accurate diagnosis. All congenital malformations suspected during the first week of life, but whose evaluations were complete subsequently were included. The abnormalities were divided into Genitourinary, Musculoskeletal, Central nervous system, Gastrointestinal, Cardiovascular, Pulmonary systems and miscellaneous disorders. Newborns with transient abnormalities such as minimal foot deformity and hydrocele were excluded. For each case, a detailed antenatal history including history of exposure to teratogens and family history, including the level of consanguinity, were obtained by reviewing the maternal and labor ward records and by interviewing the parents.

### RESULTS

Over the three years period, 4660 babies were born at Arvand hospital: 2427 males and 2233 females. Forty six mothers gave birth to twins, two triplets and one quadruple. There were 2415 (51.8%) Arabs and 2245(48.2%) Farses. The number of babies with congenital malformations diagnosed at birth or within the first week of life was 94(2.02%), 58 males and 36 females (Table-I).

The male to female ratio in malformed newborns was 1.6. The rate of congenital abnormality was different between males and females (2.38% and 1.61 %, respectively). One hundred and forty (6%) boys had mild to severe hydrocele, 84 (3.46%) boys had undescended testis and 92(2%) babies from both sexes who had minimal foot deformity were excluded. Eighty-three of the 94 newborns (88.3%) were full-term, whereas 11(11.7%) were pre-terms. The maternal age of malformed newborns were younger than 20 years in 5(5.3%) cases, 21-25 years 28(29.8%), 26-30 years 54(57.4%) and older than 30 years 7 (7.5%).

Of the 94 patients 56 were Arabs, 38 were Farses. Consanguinity was considered in 85% and 50% of the parents of newborns in Arab and Fars, respectively. The frequency of CM in Arab and Fars was 2.3% and 1.7%, respectively.

Table-II shows systemic distribution and the frequency of congenital malformations. Musculoskeletal system was the most affected, involving 37(39.3%) of 94 patients. Among this group, the most frequent anomalies were club foot, congenital dislocation of hip (CDH), polydactyly, syndactyly and oligodactyly. The genitourinary system was second in frequency, involving 33 (35.1%) out of 94. Among this group

Table-I: Congenital malformations: Frequency and sex distribution

	<i>Total live births</i>	<i>Malformed newborns</i>	<i>Percentage</i>
Males	2427	58	2.38
Females	2233	36	1.61
Total births	4660	94	2.02

Table-II: Congenital malformations (CM) among 4660 live birth newborns by system involved

Malformation/system	No. of CM (%)	Rate/1000 births
Musculoskeletal system	37(39.3)	7.9
Clubfoot	15	3.2
Congenital dislocation of hip	15	3.2
Polydactyly	4	0.9
Syndactyly	2	0.4
Oligodactyly	1	0.2
Genitourinary system	33(35.1)	7.1
Hypospadias	23	4.9
Hyronephrosis	5	1.1
Epispadias	3	0.7
Polycystic kidney disease	1	0.2
Microphallus	1	0.2
Central nervous system	11 (11.7)	2.4
Meningocele/ meningocele	5	1.1
Microcephaly	5	1.1
Anencephaly	1	0.2
Digestive system	5(5.3)	1.1
Imperforated anus	3	0.7
Cleft lip/ cleft palate	2	0.4
Chromosomal anomalies	4(4.3)	0.9
Down's syndrome	4	0.9
Cardiovascular system	3(3.2)	0.6
Acyanotic heart disease	3	0.6
Respiratory tract	1(1.1)	0.2
Pulmonary hypoplasia	1	0.2
Total	94 (100%)	20.2 /1000

the most frequent lesions were hypospadias, hyronephrosis and epispadias.

The central nervous system (CNS) came third in frequency, involving 11 (11.7%) of the patients. Meningomyelocele or meningocele, microcephaly followed by anencephaly were the most prominent CNS lesions. Anomalies of the gastrointestinal tract were the next highest group, involving 5 (5.3%) out of 94 patients. In this group imperforated anus, cleft lip and cleft palate were the most prominent lesions. Chromosomal anomalies were found in 4 (4.3%) patients, all of them had Down's syndrome. Congenital heart defect was diagnosed

Table-III: Frequency of congenital (CA) in Ahwaz, other regions of Iran and selected countries

Location / reference	Rate of CA per 1000 live births
Libya, Benghazi (14)	70
Iran, Tehran (9)	35
The USA, Atlanta (15)	31
Bahrain (12)	27
Oman (17)	24.6
Afghanistan, Kabul(21)	24
Saudi Arabia (22)	22.7
Spain (11)	20.3
UAE, Abu Dhabi (23)	16.6
Singapore(20)	15.1
Malaysa,Perak (24)	14.3
India, Maharashtra (10)	10.8
Iran, Arak (8)	10.4
UK, in five British regions (25)	8.2
Ahwaz, Iran (present study)	20.2

in 3 (3.2%) patients. Unilateral pulmonary hypoplasia was considered in a male newborn (1.1%).

## DISCUSSION

In the present study, the overall frequency of congenital malformations in the newborns was 20.2/1000. This study was similar to Mashhad, located in the North-East of Iran.<sup>7</sup> Their frequency of major congenital malformations in the study was 18/1000 which is close to our findings. But, a lower frequency was reported from Arak,<sup>8</sup> located in the center of our country. There are other reports from Iran,<sup>6,9</sup> other Islamic countries in the Middle East and other parts of the world representing different frequency of congenital malformations.<sup>10</sup> Although we got nearly the same result as reported in other studies but, if we include abortions, stillbirths and if we used screening tests and genetic studies, the frequency of CM would be more than this rate.

Table-III shows the frequency of congenital malformations in live newborns from different parts of Iran and other countries. The commonest system involved in the present study was the musculoskeletal which is in conformity with the study reported from Tehran.<sup>9</sup> But, it is much higher than the other previous reports

Table-IV: Comparison of different type of congenital abnormalities (CA) in Ahwaz with other studies.

Location	Rate of CA per 1000 birth						
	<i>clubfoot</i>	<i>cystic spina bifida</i>	<i>anencephaly</i>	<i>cleft palate± cleft lip</i>	<i>imperforated anus</i>	<i>hypospadias</i>	<i>Down syndrome</i>
Britain <sup>1</sup>	1.2	3.4	3.9	1.3	0.4	1.9	1.3
The USA <sup>26</sup>	-	0.8	0.5	1.6	0.43	3.9	0.7
India, Moharashtra <sup>10</sup>	1.04	0.34	0.69	1.04	-	0.34	0.69
Iran, Tehran <sup>27</sup>	2.9	0.92	0.08	1.61	0.38	4.45	1.23
Egypt, Giza <sup>13</sup>	-	0.66	-	1.66	0.33	2.33	1.33
Iran, Gorgan <sup>16</sup>	1.5	1.8	0.8	1.4	1.3	1.8	0.6
Oman, Nizwa <sup>17</sup>	0.68	0.59	1.27	2.41	-	-	2.00
Iran, Ahvaz (present study)	3.21	1.07	0.21	0.42	0.64	4.93	0.85

from other parts of the world<sup>10-12</sup> (Table-IV). However, some workers have reported CNS defects as highest<sup>13,14</sup> while one study has reported highest frequency of gastrointestinal malformations.<sup>15</sup> In the present study the frequency of neural tube defects was 1.1 /1000. This compares favorably with the studies from other part of Iran,<sup>8,9</sup> but lower than Britain.<sup>12</sup> The frequency of genitourinary system anomalies, including hypospadias (4.9/1000) was much more than the studies reported from India,<sup>10</sup> but in conformity with the study conducted in Tehran.<sup>9</sup> In our series, the frequency of chromosomal anomalies was 0.9/1000 which was comparable to the reports from Iran<sup>9</sup> and Egypt,<sup>13</sup> but it was higher than India.<sup>10</sup> The frequency of Down's syndrome ranging from 0.6 to 2/1000,<sup>11,16,17</sup> the highest frequency being reported from Oman. In contrast to the Oman study, the maternal age of most newborns (92.6%) in our series was younger than 30 years. In the present series, like some of the earlier studies we could observe sex predilection among babies with club foot and CDH (M:F was 4:1 and 7:8, respectively). Heridofamilial and consanguineous marriages are reported to play a major role in the occurrence of congenital malformations.<sup>2</sup> In the present study like Kuwait and the United Arab Emirates most of the malformed babies had been born out of consanguineous marriages.<sup>18,19</sup> Most of the major malformations in our study

could be prevented by using methods of pregnancy screening for neural tube defects, screening of older mother for Down's syndrome, assessing family history and prenatal diagnosis. In our region there are no well-accepted preventive measures despite the high risk of recurrence of CM, which indicates the strong need for comprehensive preventive measures for congenital anomalies in this region. There have been some efforts to address this issue, such as maternal care during pregnancy and educational programs on congenital malformations and the consequences of consanguineous marriages.

## CONCLUSION

Although the frequency of malformations in the study was approximately similar to other investigations, if we include abortions, stillbirths and if we use screening tests and genetic studies, this rate was more than 20.1/1000.

## ACKNOWLEDGMENT

The authors wish to express their thanks to Dr. Afzali for his help during this study. The authors also express their gratitude to the Director of Arvand hospital.

## REFERENCES

1. Kalter H, Warkang J. Congenital malformations: Etiology factors and their role in prevention (first of two parts). *N Eng J Med* 1983;308(8):424-31.



2. Hudgins L, Cassidy SB. Congenital anomalies. In: Martin RJ, Fanaroff AA, Walsh MC. Neonatal- Perinatal Medicine, 8<sup>th</sup> ed Philadelphia, Mosby-Elsevier 2006;561-81.
3. Lee K, Khoshoond B, Chen L. Infant mortality from congenital malformations in the USA, 1970-1990. *Obstet Gynecol* 2001;98:620-70.
4. AIHW NPSU: Birch MR, Grayson N, Sullivan EA. Recommendations for development of a new Australian birth anomalies system: A review of the congenital malformations and birth defects data collection. AIHW cat. No. PER 23. Sydney: AIH National Perinatal Statistics Unit (Birth Anomalies series no.1), 2004.
5. Evaluation of the newborn with single or multiple congenital anomalies: A clinical guide line. American College of Medical Genetics Foundation. WWW. Health.state.ny/nysdoh/dpprd/main.htm#fulldoc. Accessed on 7 August 2004.
6. Shajari H, Mohammadi N, Karbalai AM. Prevalence of congenital malformations observed in neonates in Shariati hospital (2002- 4). *Iranian J Pediatrics* 2006;16 (3):308-11.
7. Khatemi F, Mamoori GA. Survey of congenital major malformations in 10/000 newborns. *Iranian J of Pediatrics* 2005;15(4):315-20.
8. Shamohammadi F, Ahmadi MA. The survey of congenital malformations in live births in Taleghani hospital, Arak, Iran. *J Arak Uni Med Sci* 1997;1(4):23-9.
9. Farhud DD, Walizideh GhR, Kamali MS. Congenital malformations and genetic diseases in Iranian infants. *Human genetics*.1986;74:382-5.
10. Datta V, Chatuvedi P. Congenital malformations in rural Maharashtra. *Indian Pediatrics*, 2000;37:988-1001.
11. Martinez-Frias ML. Epidemiological aspects of Mendelian syndromes in a Spanish population sample: II. Autosomal recessive malformation syndromes. *Amer J Med Genetics* 1991;38:626-9.
12. Al Arrayed SS. Epidemiology of congenital abnormalities in Bahrain. *Eastern Mediterranean Health J* 1995;1(2):248-52.
13. Temtamy SA. A genetic epidemiological study of malformations at birth in Egypt. *Eastern Mediterranean Health J* 1998;4(2):252-9.
14. Singh R, Al Sudani O. Major congenital anomalies in Benghazi, Libyan Arab Jamahirya 1995. *Eastern Mediterranean Health J* 2000;6(1):65-75.
15. Rasmussen SA. Evaluation of birth defects histories obtained through maternal interviews. *Amer J Human Genetics* 1990;41(3):299-305.
16. Golallipour MJ, Ahmadvour-Kacho M, Vakili MA. Congenital malformations at a referral hospital in Gorgan, Islamic Republic of Iran. *Eastern Mediterranean Health J* 11(4)2005;707-13.
17. Sawardekar K. Profile of major congenital malformations at Nizwa hospital, Oman: 10- year review. *J Paediatr Child Health* 2005;41:323-30.
18. Madi SA, Al-Naggar RL, Al-Awadi SA, Bastak LA. Profile of major congenital malformations in neonates in Al-Jahra Region of Kuwait. *Eastern Mediterranean Health J* 2005;11(4):700-6.
19. Al-Ghazali LI. The profile of major congenital abnormalities in the United Arab Emirates (UAE) population. *J Med Genetics* 1995;32:7-13.
20. Thein MM, Koh D, Tan KL, Lee H-P, Yip YY, Tye CY, et al. Descriptive profile of birth defects among livebirths in Singapore. *Teratology* 1992;46(3):277-84.
21. Singh M. Congenital malformations at birth among live-born infants in Afghanistan, a prospective study *Indian Paediatrics* 1982;49:331-5.
22. Refat MYM. Major birth defects at King Fahd Hofuf Hospital: prevalence, risk factors and outcome. *Annals of Saudi Medicine* 1995;15(4):339-43.
23. Al Talabani J, Shubar AI, Mustafa KE. *Annals of Human Genetics* 1998;62:411-18.
24. Thong MK, HO JJ, Khatijah NN. A population-based study of birth defects in Malaysia. *Annals of Biology*. 2005;32(2):180-7.
25. Rankin J, Pattenden S, Abramsky L, Boyd P, Jordan H, Stone D. Prevalence of congenital anomalies in five British regions, 1991-99. *Archives of Disease in Childhood* 2005;90:374-9.
26. Forfar JO. Demography, vital statistics and the pattern of disease in childhood .In: Campbell AGM, McIntosh N eds. *Forfar And Arneil, s textbook of paediatrics*, 4<sup>th</sup> ed. London, Churchill Livingstone,1992:1-17.
27. Arbabi AH, Babak D. The study of major anomalies in newborns. *Proceedings of the 10<sup>th</sup> Pediatric congress*, Tehran, Islamic Republic of Iran. 1991;583-90.
28. Porter RW. Clubfoot: congenital talipes equinovarus. *J Royal College Surgeons Edinburgh*. 1995;40(1):66-71.