

Prevalence and Associated Risk Factors of Congenital Anomalies at a tertiary care hospital

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ABSTRACT

Aim: To determine the prevalence of various congenital anomalies in the neonates born at Mardan medical complex, Mardan and to determine their associated risk factors.

Methodology: This descriptive cross sectional study was done on all the congenitally anomalous babies who were born between 1st May 2016 and 30th April 2017 at Mardan Medical Complex. Meticulous history about the anomaly and possible risk factors was taken from the mother and recorded in predesigned proforma.

Results: Out of 9453 deliveries, 117 (1.23%) neonates had congenital anomalies. Hydrocephalus (27.3%), anencephaly (18%), meningocele (11%) and encephalocele (9.4%) were the most common anomalies. Mean maternal age was 25.59±7.6 years. 66 (56.4%) mothers were in age range of 21-30 years and 66 (56.4%) were multigravidas. Consanguinity was positive in 72 (61.5%) and there was lack of folic acid intake in 83 (71%) cases. 33 (28.2%) received antenatal care whereas 84 (71.8%) were deprived of it. Weight of babies in 68 (58%) cases was 1-2.5kg.

Conclusion: Consanguineous marriages and lack of folic acid are the topmost risk factors. Women and families should be awarded regarding the intake of folic acid and avoidance of consanguineous marriages.

Keywords: Congenital anomalies, Consanguineous, Antenatal Care, Hydrocephalus

INTRODUCTION

The World Health Organization has defined the term congenital anomaly as structural, biochemical, physiological or molecular defects that may develop in the fetus from conception till delivery and may present at birth, or may be detected later¹. Congenital anomalies are disorders of antenatal origin which may be caused by single gene mutation, multifactorial, chromosomal abnormalities, environmental teratogenic substances and deficiency of micronutrients. Maternal rubella, diabetes mellitus, iodine and folic acid deficiency, certain medicines, substance abuse like alcohol and tobacco, chemicals, and irradiation are other factors leading to congenital anomalies². Congenital anomalies, birth defects and congenital abnormalities are terms used for the defects present at birth³.

The frequency and specification of congenital anomalies differ from one country to another and from one area to another. This depends on the their definition, how were they detected, for how long the population was observed, ethnic and socio-economic circumstances of the population studied⁴. Congenital anomalies are classified as major and minor, major require surgical intervention or even can lead to

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death of the neonate. Minor congenital anomalies are detrimental to quality of life and effects health of neonate⁵.

In our country, 6% to 9% of perinatal mortality is due to congenital anomalies. Within this figure, in 40% to 60% cases cause is unknown, 20% are due to multiple factors, 7.5% are single gene defects, 6% are due to chromosomal abnormalities, and 5% are due to maternal ailments⁶. Congenital anomalies are responsible for 495,000 deaths worldwide⁷. They are considered as the major cause of perinatal mortality, and morbidity and disability in children worldwide⁸. These are preventable in 60% cases⁹ but needs epidemiological information. Congenital anomalies have association with poor pregnancy outcomes like perinatal mortality, neonatal mortality and morbidity, intrauterine growth restriction, preterm birth, breech presentation, preeclampsia, placental abruption and obviously if discovered antenatally is a source of stress for the pregnant mother¹⁰.

Congenital anomalies is a problem prevailing since centuries and is very well recognised. It is an issue for research because of the high prevalence and the devastating consequences they can have on the neonate and whole family. We have carried out this study to determine the prevalence of different congenital anomalies and their associated risk factors

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in our local set up. This can help us to modify these risk factors and in the long run can help to decrease the incidence of these anomalies in our own society.

MATERIALS AND METHODS

This descriptive cross sectional study was conducted at Gynaecology and Obstetrics Department of Mardan Medical Complex, Mardan from 1st May 2016 to 30th April 2017 for the duration of one year. Mothers delivering congenitally anomalous babies during this one year were included in the study. They were queried and a predesigned proforma used to collect all the information gathered. The congenitally anomalous babies were sent to Paediatrician for detailed examination and abnormality was recorded.

Approval was taken from the Hospital Ethical Committee. Verbal consent was taken from all the mothers recruited into the study. Detailed history regarding the risk factors was taken from all the subjects regarding maternal and paternal age, maternal medical disorders e.g., diabetes, epilepsy etc., active or passive smoking, periconceptional use of folic acid, history of congenital anomalies in family, consanguineous marriage, drug history, antenatal TORCH infections, antenatal radiation exposure, antenatal checkups and past history of abortions or congenital anomalies. Karyotyping being important part of this type of study was unable to be performed due to affordability problems and also it was not available. Ages were determined from identity cards. Three categories of marriages were included i.e., first cousin, second cousin and non consanguineous.

All the data was analyzed using SPSS version 20.0. Mean and standard deviation were calculated for numerical data whereas frequency and standard deviation was calculated for the categorical data. All the data was presented in the form of tables.

RESULTS

Out of the 9,453 deliveries, 117 (1.23%) babies had various congenital anomalies. Anomalies of central nervous system were in majority comprising 92 (78.6%) of subjects with hydrocephalus on the top effecting 32 (27.3%) babies, followed by anencephaly in 21 (18%), meningomyelocele in 13 (11.1%), encephalocele in 11 (9.4%), hydrocephalus+meningocoele in 8 (6.8%) and microcephaly in 7 (6%) babies. If systems are considered, CNS was followed by musculoskeletal, gastrointestinal, renal, facial deformity and miscellaneous disorders in descending order (Table I)

Maternal age was divided into four groups, 10 (8%) cases fell into age group of <20 years, 66 (56.4%) fell into 21-30 years age range, 29 (24.7%) were of 31-40 years age group and 12 (10%) were greater than 40 years age. Mean age was 25.59±7.6 years. Primigravidas were 19 (16.2%), multigravidas were 66 (56.4%) and grand multigravidas were 52 (27.3%). 33 (28.2%) patients received antenatal care whereas 84 (71.8%) were deprived of it. Regarding weights of newborns, 68 (58%) had 1-2.5kg weight, 46 (39%) had 2.6-4kg and 3 (2.56%) had more than 4kg weight.

Among the different risk factors which were evaluated, consanguinity and lack of folic acid intake was the most common, with positive consanguinity in 72 (61.5%) cases and lack of folic acid intake in 83 (71%) subjects. Other risk factors were responsible for comparatively lesser number of cases (Table II). Regarding the consanguineous relationships, 50 (42%) were first cousins and 22 (18%) were second cousins.

Table I: risk factors of congenital anomalies (n=117)

Risk factor	Yes	No
Consanguinity	72 (61.5%)	45 (38.4%)
Folic acid intake	34 (29%)	83 (71%)
Past h/o miscarriages	15 (12.8%)	102 (87%)
Maternal medical illness	14 (12%)	103 (88%)
Maternal age >40 years	12 (10%)	105 (90%)
h/o drug intake	11 (9.4%)	106 (90.5%)
Past h/o congenital anomalies	10 (8.5%)	107 (91.5%)
Past h/o stillbirth/neonatal death	7 (6%)	110 (94%)
h/o Radiation exposure	1 (0.85%)	116 (99.1%)
Smoking	-	7 (100%)

Table II: Types of anomalies

Anomaly	Frequency	%age
CNS Anomalies	92	78.6
Hydrocephalus	32	27.3
Anencephalus	21	18
Meningomyelocele	13	11
Encephalocele	11	9.4
Hydroceph+ Meningomyelocele	8	6.8
Microcephaly	7	6
Musculoskeletal Anomalies	7	6
Talipes equinovarus	4	3.4
Polydactyly	2	1.7
Achondroplasia	1	0.85
Gastrointestinal Anomalies	5	4.27
Omphalocele	2	1.7
Gastroschisis	2	1.7
Fetal ascites	1	0.85
Renal Anomalies	4	3.4
Polycystic kidneys	2	1.7
Multicystic/Dysplastic kidneys	2	1.7

Facial Deformities	2	1.7
Miscellaneous	7	6

DISCUSSION

Major congenital anomalies occur in 2-3% of births. Depending on the nature of anomaly, it may be detected earlier after birth, or later. Keeping in view the importance of detection of these congenital anomalies in time, developed countries have designed special programmes for estimating prevalence and prevention of congenital anomalies, and also timely diagnosis in case they occur¹¹.

The frequency of congenital anomalies in our hospital was 1.23% or 12.3/1000 total births. This corresponds to a study by Parveen F et al at Liyari General hospital where this rate was found to be 11.4/1000 total births¹². Madi et al showed an incidence of 1.25% in their study at Kuwait¹³. Likewise, 1.9% was rate reported by an author in Iran¹⁴ and 1.4% was reported in a study at Lady Reading Hospital, Peshawar¹⁶.

In our study, anomalies of central nervous system were in majority, succeeded by musculoskeletal anomalies. Same pattern was reported by Gul F et al in their study at Kohat, Khyber Pakhtoonkhwa¹¹. Nasreen A et al and Karim R et al also observed same order of affected systems in their studies^{15,16}. This finding is further supported by two other international studies^{17,18}.

Looking at the results of our study, consanguinity was the topmost risk factor in the causation of congenital anomalies. Our country is having highest rates of 61% for consanguineous marriages¹⁹. Nasreen A et al reported 46% consanguinity rate in their study¹⁵ whereas Sheridan E et al in their study at Bradford reported doubling of risk of congenital anomalies in consanguineous marriages²⁰, whereas a study from Iran shows 3.5 times more frequency of congenital anomalies in consanguineous marriages²¹. Lack of folic acid intake, which is an easily preventable factor was admitted by 71% women in our study. Shawky RM et al reported only 72.5%¹ whereas according to Gul F et al, 66.7% subjects denied intake of folic acid¹¹. Blencowe et al in their meta-analysis has shown a 70% reduction in the reduction of neural tube defects with the intake of folic acid²².

Advanced maternal age is also a contributory factor in aetiology of congenital anomalies. Increased chromosomal meiotic errors occur with increasing maternal age, resulting in congenital anomalies. Mean maternal age was 25.59±7.6 years whereas only 10% mothers were more than 40 years of age in our study. Our results are very close to those observed by

Gul F et al where the mean age was recorded as 26.10±7.4 years and 6.5% were greater than 40 years age¹¹. Tootoonchi et al reported 25.69 + 5.54 years in their study²³ and Tomatir AG et al had 8.7% subjects in their study who were more than 35 years age²⁴. 56.4% cases in our study were multigravidas, supported by an Egyptian study where this figure was 54%¹ and a local study by Gul F et al where 85% women were multigravidas¹¹.

In our study, 58% babies had birthweight of less than 2.5kg. Two local studies from Abbotabad and Kohat also reported the same findings^{11,25} and it is a known fact that there is association of congenital anomalies with intrauterine growth restriction and low birth weight, IUGR may be cause or effect of the congenital anomaly or both may co-exist²⁵.

Focussing on our study, the rate is much lower compared to the international figures. The reason may be missing the inborn errors of metabolism which are diagnosed later on and the patients with cardiac lesions who were diagnosed after the patients are discharged from maternity wards. Secondly, no paediatric cardiology unit is available in Mardan Medical Complex so the cases were referred to Peshawar. CNS lesions may be more prevalent because of the fact that they are easily diagnosed at birth.

There are several limitations in our study. As this is a hospital based study, so its results cannot be generalizable. Folate levels estimation could not be done due to high cost despite that it was an important risk factor. Chromosomal abnormalities could not be diagnosed because of lack of availability of karyotyping. Lastly, only Mardan Medical Complex was the study place. Inclusion of hospitals from the same locality could have given better idea about prevalence and risk factors in that area.

CONCLUSION

Consanguineous marriages and lack of intake of folic acid are the two most common risk factors as determined from our study. It is recommended to create general awareness among masses to emphasize the intake of folic acid around and in early conception time and to avoid inter-cousin marriages in families. It is recommended that antenatal care should be made universal and encouraged as it has important role in prevention as well as timely diagnosis and further management in case the congenital anomalies occur.

REFERENCES

1. Shawky RM, Sadik DI. Congenital malformations prevalent among Egyptian children and associated risk factors. *Egyptian J Med Hum Gen* 2011; 12: 69- 78.

2. El Koumi MA, Banna EA, Lebda I. Pattern of congenital anomalies in newborn: a hospital based study. *Paed Reports* 2013; 5: 5.
3. Tanteles GA, Suri M. Classification and etiology of birth defects. *Pediatr Child Health* 2007; 17: 233- 43.
4. Aihw NP, 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: AIHW National Perinatal Statistics Unit (Birth Anomalies Series no.1): 2004.
5. Sadler TW: *Langman Medical Embryology*. 10th Edition. Lippincott Williams and Wilkins 2011; 1: 11.
6. Korejo R, Bhutta S, Noorani KJ. An audit and trends of perinatal mortality at Jinnah Postgraduate Medical Centre, Karachi. *J Pak Med Ass* 2007; 57(4): 168- 72.
7. Kalter H. Five decades international trends in the relation of perinatal mortality and congenital malformations: stillbirth and neonatal death compared. *Int j epidemiol* 1991; 20: 173.
8. Dastgiri S, Sheikhzadeh Y, Dastgiri A. Monitoring of congenital anomalies in developing countries: a pilot model in Iran. *Studies in Health Technology and Informatics* 2011; 164: 157- 61.
9. Czeizel AE, Intody Z, Modell B. What proportion of congenital anomalies can be prevented?. *BMJ* 1993; 306(6876): 499-503.
10. Ali WHA, Balaha MH, Moghannum MSA, Hashim I. Risk factors and birth prevalence of birth defects and inborn errors of metabolism in Al Ahsa Saudi Arabia. *Pan African Med J* 2011; 8(14): 1- 9.
11. Gul F, Jabeen M, Khan AS. Frequency of congenital malformations and associated risk factors at Liaqat Memorial Hospital, Kohat. *Khyber Med Univ J* 2012; 4(3): 119- 24.
12. Perveen F, Tyyab S. Frequency and pattern of distribution of congenital anomalies in the newborn and associated maternal risk factors. *J Coll Physicians Surg Pak* 2007; 17(6): 340- 3.
13. Madi SA, Al-Naggar RL, Al-Awadi SA, Bastaki LA. *East Mediterr Health J* 2005; 11: 700- 6.
14. Movafagh A, Zadeh ZP, Javadi MH, Mohammad FM. Occurrence of congenital anomalies and genetic diseases in a population of Ghazvin province, Iran. *Pak J Med Sci* 2008; 24(1): 80- 5.
15. Nasreen A, Naib JM, Ibrar M. Frequency of birth defects and associated risk factors. *PJMHS* 2016; 10(2): 541- 3.
16. Karim R, Wahab S, Akhtar R, Jamala F, Jabeen S. Frequency and pattern of antenatally diagnosed congenital anomalies and the associated risk factors. *J Postgrad Med Inst* 2014; 28(2): 184- 8.
17. Verity C, Firth H, French C. Congenital abnormalities of the central nervous system. *J Neurol Neurosurg Psychiatry* 2003; 74: 13- 8.
18. Fatemaq K, Begum F, Akter N, Zaman SMM. Major congenital malformations the newborns in BSMUU hospital. *Bangladesh Med J* 2011; 40(1): 7- 12.
19. Rukannudin AR, Hasan HK. *Maternal and child health. Chap 10. Pakistan demographic and Household Survey 1990- 91. Islamabad: Government of Pakistan.*
20. Sheridan E, Wright J, Small N, Corry CP. Risk factors for congenital anomalies in a multi ethnic birth cohort: an analysis of the born in Bradford study. *Lancet* 2013; 382: 1350- 9.
21. Mosayebi Z, Movahedian AH. Pattern of congenital malformations in a consanguineous marriage versus non consanguineous marriages in Kashan, Islamic republic of Iran. *East Mediterr Health J* 2007; 13(4): 868- 75.
22. Blencowe H, Cousens S, Modell B, Lawn J. Folic acid to reduce neonatal mortality from neural tube disorder. *Int J Epidemiol* 2010; 39: 110- 21.
23. Tootoonchi P. Easily identifiable congenital anomalies: prevalence and risk factors. *Acta Medical Irania* 2003; 41(1): 15- 9.
24. Tomatyr AG, Demirhan H, Sorkun HC. Major congenital anomalies: a five year retrospective regional study in Turkey. *Genet Molecul Res* 2009; 8(1): 19- 27.
25. Khoury MJ, Erickson JD, Cordero JF. Congenital malformations and intrauterine growth restriction: a population study. *Paediatrics* 1988; 82: 83- 90.