ORIGINAL ARTICLE

Prevalence and pattern of congenital malformations among neonates in the neonatal unit of a teaching hospital

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Abstract

Objective: To determine the prevalence and pattern of congenital malformations among neonates in a teaching hospital.

Methods: The prospective hospital-based study was conducted over a period of 18 months in the neonatal unit of Combined Military Hospital, Kharian, from September 2011 to February 2013. All neonates from newborn to 28 days of age admitted to the unit irrespective of their condition comprised the study population. Neonatal examination was done by the Registrar at the time of admission followed by neonatologist/paediatrician. Information regarding gender, weight, gestational age, mode of delivery, consanguinity, maternal age, antenatal visit record and family history were recorded on a predesigned proforma. After clinical examination, if required, relevant investigations like ultrasonography, radiology, echocardiography, laboratory and genetic studies were done to confirm diagnosis. Data was statistically analysed by using SPSS 20.

Results: Out of 3,210 total admissions, 226 (7%) neonates were congenitally malformed. Of them, 130 (57.52 %) were male and 96 (42.47 %) females. Among different body systems affected, anomalies related to the central nervous system were 46(20.35%) musculoskeletal 42(18.58%), genitourinary 34 (15.04%), cardiovascular system 30 (13.27%), ear, eye, face, neck 27(11.94%), digestive system 19 (8.40%), syndromes and skin 14 (6.19%) each.

Conclusion: Congenital Malformations are not rare in our community and central nervous system is the most commonly affected system. Healthcare managers must stress upon primary prevention in the form of vaccination, nutrition and drugs to decrease preventable share of congenital malformations.

Keywords: Congenital malformations, Neonates, Pakistan, Prevalence, Pattern. (JPMA 64: 629; 2014)

Introduction

Congenital malformation (CM) means any abnormality, genetically or otherwise, that is present at birth. To be more precise, it is an abnormality of physical structure that is seen at birth or within few weeks after birth.[1] According to World Health Organisation (WHO) document of 1972, the term congenital malformation should be confined to structural defects present at birth. CM may be minor or major. Minor malformation is defined as structural abnormality present at birth which has minimal effect on clinical function, but may have a cosmetic effect e.g. preauricular tag. Major malformation has a significant effect on function or on social acceptability e.g. ventricular septal defect (VSD) and cleft lip.[2] Initially, malformations can be categorised into three groups:[2] Single malformation; Multiple malformations, recognisable pattern (syndrome); and Multiple malformations, pattern not recognisable.

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A syndrome is defined as a pattern of multiple abnormalities that are related by pathophysiology and result from common, defined aetiology. Dysmorphology is study of abnormalities of human form and mechanism that cause these abnormalities. About 20-30% of infant deaths and 30-50% post-neonatal deaths are due to CM. First trimester, especially between the 3rd and 8th weeks of gestation, is the crucial period for morphogenesis of organs. Any insult in any form during this period can cause congenital abnormality. This is the period where preventive intervention strategy will reduce the incidence of developing CMs.[3]

The aetiology of CMs may be genetic, environmental or unknown. Among genetic causes, 6% are due to chromosomal abnormality, 25% single-gene disorders, and 20-30% multifactorial. In 50% cases, the cause is unknown.[4] Among preventable causes Folate supplementation during peri-conception period and first trimester definitely helps in the prevention of neural tube defects.[5] In some developed countries food fortification with folic acid significantly reduced incidence of neural tube defects.[5] Incidence of CM is much higher in low birth weight (LBW) than normal birth weight children[3] and in consanguineous than non-consanguineous marriages.[4]

Consanguinity is very common in Pakistan. Among other risk factors for CM are maternal age, drug intake, teratogens, radiation exposure, maternal illnesses, smoking and alcohol consumption.[6] Different antenatal screening methods like maternal serum markers, chorionic villus sampling, amniocentesis, cordocentesis and ultrasonography can be used to detect anomalies. On the basis of findings of these modalities, termination of pregnancy or therapeutic manipulation can be done. In utero intervention for some CMs like hydrocephalus, Posterior Urethral Valves (PUVs), cleft lip and hydronephrosis is gaining popularity.[7]

Prevalence of CM ranges between 3-7% and varies in different geographical, racial and ethnic parts of world.[8,9] As far as involvement of different systems of the body is concerned, brain has the highest incidence of CM i.e. 10/1000 followed by heart 8/1000, kidney 4/1000, limb 1/1000 and miscellaneous 6/1000 live births.[10] Incidence of CM is higher in black children than white. In Pakistan, 6-9% perinatal deaths are due to CM.[11] Morbidity and mortality of children with CM is much higher than normal children.[12]

Preventive measures are primary and secondary. Primary prevention involves folic acid supplementation, maternal disease prevention by vaccination, especially against rubella and chickenpox. Secondary prevention is targeted at early antenatal detection followed by termination of pregnancy, but it involves social, legal and religious issues.[13]

We conducted this study to find out the prevalence and pattern of congenital malformations in different categories of neonates admitted to our neonatal unit. It is of paramount importance from preventive strategic point of view to know about the characteristics of neonates and malformations. Hopefully information available from this study will help the health managers to evolve preventive strategy to decrease the prevalence of CM in the study area.

Subjects and Methods

The prospective hospital-based study was conducted in the neonatal unit of Combined Military Hospital, Kharian, from September 2011 to February 2013. All neonates from newborn to 28 days of age admitted to the neonatal unit were included. The study was regardless of gender, gestational age, weight, race, ethnicity, geographical distribution and socioeconomic status. Meticulous neonatal examination for neonatal care and detection of any kind of CM was done by the registrar at the time of admission which was followed by neonatologist/paediatrician. Necessary radiological, haematological and genetic investigations were done where required. Ultrasonography and radiological investigations were performed by the hospital radiologist to detect and rule out multiple internal anomalies, where it was considered necessary. Echocardiography and genetic studies were requested from our sister tertiary care medical facilities. Variables like gender, weight, gestational age, mode of delivery, consanguinity, maternal age, antenatal visit record and family history were recorded on a predesigned proforma. Autopsy on neonatal death was not done due to social and religious factors. Written consent of parents for participation in the study was taken. Data was analysed using SPSS version 20.

Results

A total of 3,210 neonates were admitted during the study period and of them 226 (7%) were found to have different types of CMs; 130(57.52%) were males and

Table-1: Study parameters.

Parameter		Frequency	Percentage
Gender		**226**	
	Male	130	57.52%
	Female	96	42.47%
Gestational Age		**226**	
	Pre-Term	116	51.32%
	Term	84	37.16%
	Post-Term	26	11.50%
Weight		**226**	
	<2.5KG	126	55.75%
	2.5-4KG	86	38.05%
	>4KG	14	6.19%
Mode of Delivery		**226**	
	LSCS	126	55.75%
	Others	100	42.24%
Consanguinity			
	Yes	156	69.02%
	No	70	30.97%
Gravada			
	One	90	39.82%
	Two or More	136	60.17%
Antenatal Visit			
	1st Trimester	40	17.69%
	2nd Trimester	120	53.09%
	3rd Trimester	66	29.20%
Family History			
	Yes	50	22.12%
	No	176	77.87%
Maternal Age		**226**	
	>30 Years	100	44.24%
	25-30 Years	30	13.27%
	20-25 Years	26	11.50%
	<20 Years	70	30.97%

LSCS: Lower Section Caesarean Section.

Table-2: Distribution of Congenital Malformations (n=226).

System	Malformation Type	Frequency	Percentage of Total Malformed Cases
(1) CNS		**46**	**20.35%**
	Meningomyelocoele+/-hydrocephalus	35	
	Microcephaly	5	
	Encephalocoele	3	
	Sacrococcygeal teratoma	2	
	Anencephaly	1	
(2) Musculoskelatal System		**42**	**18.58%**
	Talipes	20	
	DDH	8	
	Polydactyly	5	
	Craniosynostosis	4	
	Syndactyly	2	
	Spinabifida	2	
	Rhizomelia	1	
(3) Genitourinary System		**34**	**15.04%**
	Polycystic Kidney	3	
	Hydronephrosis	4	
	Duplex system	2	
	PUV	2	
	Hypospadias	8	
	Epispadias	4	
	Micropenis	1	
	Hydrocoele	5	
	Undescended testes	5	
(4) Cardiovascular System		**30**	**13.27%**
	Acyanotic	20	
	Cyanotic	8	
	Complex	2	
(5) Digestive System		**19**	**8.40%**
	Oesophageal atresia	3	
	Diaphragmatic hernia	1	
	Duodenal atresia	1	
	Jejunal atresia	2	
	Bands	2	
	Anorectal malformations	6	
	Gastroschiasis	1	
	Exomphalocoele	1	
	Malrotation	2	
(6) Ear, Eye, Face, Neck		**27**	**11.94%**
	Abnormal pinna	4	
	Atresia, external canal	1	
	Buphthalmous	1	
	Cleft lip and palate	16	
	Micrognathia	4	
	Choanal atresia, bil	1	
(7) Syndromes		**14**	**6.19%**
	Downs syndrome	10	
	AMC	2	
	Pier Robin syndrome	1	
	Prune belly syndrome	1	
(8) SKIN		**14**	**6.19%**
	Preauricular tags	10	
	Haemangioma	2	
	Giant hairy nevus	2	
Total		**226**	**100%**

CNS: Central Nervous System. DDH: Developmental Dysplasia of the Hip. PUV: Posterior Urethral Valve. AMC: Arthrogryposis multiplex congenita.

96(42.47%) females. LBW neonates were 126(55.75%) and further distribution in weight category revealed that 86(38.05%) were between 2.5kg to 4kg and 14(6.19%) >4kg. Regarding gestational age, 116(51.32%) were pre-term, 84(37.16%) full-term and 26(11.50%) post-term. Besides, 126(55.75%) neonates were born by lower-section Caesarean Section (LSCS) and 100(44.24%) by simple vaginal delivery (SVD). Maternal age parameter revealed that 100(44.24%) mothers were above 30 years, 30(13.27%) 25-30 years, 26(11.50%) 20-25 years, and 70(30.97%) below 20 years. History of consanguinity was positive in 156 (69.02%) couples. Family history of CMc was positive in 50(22.12%) cases. A total of 120(53.09%) mothers visited first time for antenatal check-up during the 2nd trimester; 90(39.82%) mothers were primi-gravida (Table-1). Most commonly involved body systems were the central nervous system (CNS) 46 (20.35%), followed by Musculoskeletal 42 (18.58%), Genitourinary 34(15.04%), cardiovascular system (CVS) 30 (13.27%), Ear, eye, face, neck 27(11.94%), Gastrointestinal 19 (8.40%), Skin 14 (6.19%) and Syndromes 14 (6.19%) (Table-2).

Discussion

Prevalence rate of CMs in our study was 7%. True prevalence of CMs depends upon many factors like place of study, nature of sample, ethnicity, geographical distribution and socioeconomic status. That is why, any two studies are never comparable in the strict sense of the term. Worldwide it is 3-7% but varies from country to country.[9] Our prevalence rate was low compared to 13% reported earlier.[7,14] It was comparable with a hospital neonatal unit based study[15] (4.23%), and high compared with international data. Prevalence from Nigeria[12] has been reported as 2.7%, Taiwan[16] 4.3%, Oman[16] 2.46%, Bahrain[16] 2.7% and India[17] 1.5%.

Most commonly affected system in our study was the CNS followed by Musculoskeletal, Genitourinary, CVS, Ear eye face neck, Gastrointestinal systems and Syndromes in descending order of frequency. It is in comparison with a study from Saudi Arabia[18] that also reported CNS as the most commonly affected system followed by Musculoskeletal and then Renal. Similarly, a study from Iran[19] reported CNS, Musculoskeletal, Gastrointestinal, Urogenital and Chromosomal disorders in descending order of frequency. An Indian study[20] revealed first ranking for CNS followed by Musculoskeletal and then CVS. Similarly, another study[21] also reported CNS anomalies as the commonest. But in contrast an Iranian study[22] reported Genitourinary system as the one most often affected. Some studies from Iran[23-25] and India[26] showed Musculoskeletal anomalies as the commonest.

Studies from Pakistan[7,14] reported Gastrointestinal defects as the commonest but others[13,15] supported CNS findings. A plausible explanation for these minor differences is that in our set-up sub-specialties like Plastic Surgery, Paediatric Surgery, Neurosurgery, Urology and Neonatology exist and neonates after birth or in utero are referred here for tertiary care.

In our study, the rate of CMs outnumbered in males compared to females and was consistent with a study from Brazil[27] (55.5% male and 43.6% in females). Another study[22] also reported that CMs were more common in males than females (2.1:1 ratio). It was also consistent with results of other studies,[13,25,28] but in contrast with results of one study[29] (female 59.1%).

The incidence of CMs in our study was significantly higher in pre-term babies compared to the full-term ones. It represented the Phenomenon of Nature's Selection and was consistent with results of a study from Brazil[27] (67% pre-term and 33% term), and others,[21,28] but in contrast with another study[13] which reported tendency of anomalies more common in Pakistan in term neonates.

Association of LBW with increased incidence of anomalies was found in our study and was in accordance with results others.[13,27-29]

We also had LSCS preponderance as the mode of delivery which was consistent with earlier results[27] but in contrast with another.[16] Most probable explanation for it is foetal indication due to prenatal diagnosis of CM.

Maternal age's association with congenital anomalies is considered an important factor. Our study revealed that mothers above 30 years of age had high incidence of producing malformed babies. It was in accordance with earlier studies.[21,28] One Pakistani sudy[29] has reported the highest (80.6%) incidence between 20-40 years age group.

High incidence of CM among gravida 2 or more than primi-gravida was reported by our study and was similar to earlier reports.[28,29] It indicates that the incidence of CM increases as the birth order increases.

Consanguinity is considered a controversial association with CMs. Increased incidence of CM in consanguineous couples is due to homozygous expression of recessive genes inherited from common ancestors.[16] The current study found CMs to be more common in consanguineous

parents than non-consanguineous which was in accordance with an earlier study (61.3%).[29] Another study[22] also reported that CMs were 3.5 times more common in consanguineous than non-consanguineous marriages. This association is also supported by another study.[16]

Antenatal care helps in early detection of CMs. In Pakistan it is reported that 95% of pregnant women receive antenatal care from lady health visitors (LHVs) or nurse and only 5% from doctors. In our study, the minority of mothers received antenatal care during the 1st and majority during the 2nd trimester. It is partially in accordance with a study from Nigeria[3] where all mothers whose babies had CMs received antenatal care in the 2nd trimester. But a Pakistani study[29] reported that only 32.3% mothers with CM babies received antenatal care irrespective of the stage of pregnancy. Apparent cause for this difference is socioeconomic and education status of the studied population. Availability of health facilities definitely has impact on this count.

Family history of CMs was present in nearly quarter of our cases and was comparable to 8.6%, 9% and 17% reported earlier.[29-31] We could not find any definite explanation for these differences except that cousin marriages were very common in the study area.

In terms of limitations, the current study was based on a hospital neonatal unit and, as such, is not representative of the situation in the community at large. Besides, the hospital did not have paediatric cardiology facilities and many cases of coronary heart diseases are likely to have been missed.

Conclusion

CMs are not rare in our set-up and CNS was the most commonly affected system in our study. Prematurity, LBW, male gender, consanguinity, advanced maternal age and family history of CMs were associated risk factors for CMs in neonates. Knowledge of incidence and pattern of CMs are important to plan preventive strategies at different levels by healthcare providers.

References

1. Hudgins I, Cassidy SB: Congenital malformations among live births at Arvand Hospital Ahwaz, Iran. Pak J Med Sci 2008; 24: 33-7.
2. Evaluation of infant with single or multiple congenital anomalies. Guide lines American College of Medical Genetics. (Online) (Cited 2013 January 26). Available from: URL: http/www.health.ny.gov/nysdoh/dpprd/exec.htm.
3. Obu H A, Chinawa JM, Uleanya ND, Adimora GN, Obi IE. Congenital malformations among newborns admitted in the neonatal unit of a tertiary hospital in Enugu, South East Nigeria: a retrospective study. BMC Res Notes 2012; 5: 177.
4. Rajangam S, Devi R. Consanguinity and chromosomal abnormality in mental retardation and or multiple congenital anomalies. J Anat Soc India 2007; 56: 30-3.
5. Castilla EE, Orioli IM, Lopez-camelo JS, Dutra MG. Preliminary data on changes in neural tube defect prevalence rates after folic acid fortification in South America. Am J Med Genet 2003; 123: 123-8.
6. Parveen F, Tayyub S. Frequency and pattern of congenital anomalies in newborn and associated maternal risk factors. J Coll Physicians Surg Pak 2007; 17: 340-3.
7. Rafi M, Iqbal Z, Saleem M, Waseem M, Anwar J, Saleem M. Pattern of congenital malformations and their outcome at Sheikh Zayed Medical College/Hospital Rahim Yar Khan. Pak J Med Health Sci 2011; 5: 94-6.
8. Kucik JE, Alverson CJ, Gilboa SM, Correa A .Racial/ethnic variations in the prevalence of selected major birth defects, 1994-2005. Public Health Rep 2012; 17: 52-61.
9. Park K. Congenital malformations.In: K Park(Ed). Park's Text book of Preventive and Social Medicine.15th edition. BANARSIDAS BHANOT PUBLISHERS, 2005; pp 379-80.
10. Kumar V. Congenital abnormality of brain. In:Kumar V, Abbas A, Fausto N (eds). Robbins and Cotrans pathological basis of disease. Seattle Washington: W B Sanders, 2010; pp 470.
11. Korejo R, Bhutta S, Noorani KH, Bhutta ZA. An audit and trends of perinatal mortality at the Jinnah postgraduate Medical Centre Karachi. J Pak Med Assoc2007; 57: 168-71.
12. Ambe JP, Madzica AG, Akpede GO, Mava Y. Pattern and outcome of congenital malformations in newborn babies in Nigerian teaching hospital. West Afr J Med 2010; 29: 24-9.
13. Khan AA, Khattak TA, Shah SHA, Roshan E, Haq AU. Pattern of congenital anomalies in the newborn. JRMC; 2012; 16: 171-3.
14. ShamimS,Chohan N, Qmar S. Pattern of congenital malformations and their neonatal outcome. J Surgery 2010; 15: 34-7.
15. Gillani S, Kazmi NHS, Najeeb S, Hussain S, Raza A. Frequency of congenital anomalies among newborns admitted in nursery of Ayub teaching hospital Abbottabad, Pakistan. J Ayub Med Coll Abbottabad 2011; 23: 117-21.
16. Tayebi N, Yazdani K, Naghshin N. Prevalence of congenital malformations and its correlation with consanguineous marriages. Oman Med J 2010; 25: 37-40.
17. Singh A, Gupta R K. Pattern of congenital anomalies in newborn: A hospital based prospective study. J K Science 2009; 11: 34-6.
18. Asindi AA, Al Hifzi I, Bassuni WA. Major congenital malformations among Saudi infants admitted to Asir central hospital. Ann Saudi Med 1997; 17: 250-3.
19. Abdi-Rad I, Khoshkalam M, Farrokh-IslamlouHR.The prevalence at birth of overt congenital anomalies in Urmia Northwest Iran. Arch Iran Med 2008; 11: 148-5.
20. Gupta S, Gupta P, Soni J S. A study on incidence of various systemic congenital malformations and their association with maternal factors.Natl J Med Research 2012; 2: 19-21.
21. Parmar DA, Rathod D S P, Patel DSV, Patel DSM. A study of congenital anomalies in newborn . NJIRM 2010; 1: 13-7.
22. Mosayebi Z, Movahedian AH. Pattern of congenital malformations in consanguineous versus non consanguineous marriages in Kashan, Islamic Republic of Iran. East Mediterr Health J 2007; 13: 868-75.
23. Ali A, Shafikhani Z, Abdulahi M. Congenital malformations among live births at Arvand Hospital Ahwaz, Iran. A prospective study. Pak J Med Sci 2008; 24: 33-7.
24. Khatemi F, Mamorri GA. Survey of congenital major malformations in 10,000 newborns. Iranian J Pediatr 2005; 5: 315-20.

25. Golalipour MJ, Ahmadpour-Kacho M, Vakili MA. Congenital malformations at a referral hospital in Gorgan,Islamic Republic of Iran. East Mediterr Health J 2005; 11: 707-15.
26. Gupta RK, Gupta CR, Singh D. Incidence of congenital malformations of the musculoskeletal system in live newborns in Jammu. JK Science 2003; 5: 157-60.
27. Oliveira CI, Costa AR, Ferrarese VCC, Mozvaz DC, Fett-conte AC. Birth defects in newborns and stillborns: An example of the Brazilian reality. BMC Notes 2011; 4: 343.
28. 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: 159-63.
298. 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: 119-24.
30. Ahmed AM, Elkader SA, Hamid AA, Gaafar HM. Assessment of risk factors for fetal congenital anomalies among pregnant women at Cairo university hospitals. J Am Sci 2011; 7: 899-908.
31. Sridhar K. A community based survey of visible congenital anomalies in rural Tamil Nadu. Indian J Plast Surg 2009; 42 (suppl); S184-S91.