A Study of the Spectrum of Congenital Malformations in Neonates Admitted in the Neonatal Unit of the Pediatric Department of Government Medical College Srinagar

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ABSTRACT

Introduction: Congenital abnormalities play a major role in mortality and morbidity of children. The treatment and rehabilitation of these children can be very costly, hence the need to identify the causative and risk factors and prevent them early wherever possible. Objectives of the study were to study the spectrum of congenital abnormalities in neonates admitted in the special newborn care unit and to identify any risk factors associated with the occurrence of these abnormalities

Material and method: This was a prospective observational study conducted in the Pediatric Department of Govt Medical College Srinagar. The diagnosis of congenital abnormality was based on a detailed physical examination and relevant investigations.A detailed history including the antenatal history and possible risk factors, was sought on a pre designed proforma.

Results: Out of 4987 admissions during the study period 145 neonates had one or more congenital abnormalities. Musculoskeletal system defects accounted for majority of the cases 20.8% (n=41), followed by GIT 17.7% (n=35), CVS 16.75% (n=33), genitourinary 10.65% (n=21), CNS 8.1% (n=16) and others 16.24% (n=32). Congenital anomalies were more common in males, premature and low birth weight babies and babies born to mothers >30 years of age.

Conclusion: Musculoskeletal system was most commonly affected in our study. Prematurity, LBW, male gender, advanced maternal age were associated risk factors for congenital malformation in neonates.

Keywords: Congenital anomaly, risk factors, newborns, genetic, environmental, multi-factorial

INTRODUCTION

Birth defects are abnormalities of body structure or function that occur during intrauterine life, can be identified prenatally, at birth or later Sometimes also called as Congenital anomalies, Congenital abnormalities or Congenital malformations.[1] The etiology of congenital abnormality may be genetic (30-40%) or environmental (5-10%). Among the genetic causes chromosomal abnormality makes upto about 6%, single gene disorders about 7.5% and multifactorial factors 20-30%. In about 50% of cases, the cause is not known.[2,3]

Early intrauterine period (between the 3rd and 8th week of gestation) is the vital period of life for the normal development of organs. Any insult within that period may result in congenital abnormalities. It can further be argued that interventions within this period targeted at preventing insults (or removing the effect of insults) to the developing foetus will reduce the likelihood of an abnormality developing. For instance, it is known that folic acid supplementation helps in the prevention of neural tube defects especially in the first trimester. It is however observed that better maternal care and improved standards of living have little effect on the overall frequency of congenital malformations[4,5]

Congenital abnormalities can be classified into four descriptive categories on the basis of, location into external or internal birth defects, health impact into major or minor birth defects, clinical presentation into isolated or multiple birth defects and on the basis of pathogenesis into malformations, deformations, disruptions and dysplasias.

Congenital abnormalities play a major role in mortality and morbidity of children. However the treatment and rehabilitation of these children can be very costly, hence the need to identify the causative and risk factors and prevent them early wherever possible. The birth of an infant with major malformations whether diagnosed antenatally or not evokes an emotional parental response. Early recognition of anomalies is important for planning and care. Parents are likely to feel anxiety and guilt on learning of the existence of a congenital anomaly in the child and require sensitive counselling.

Prevalent studies of congenital anomalies are useful to establish baseline rates, to document changes over time and to identify clues to etiology. They are also important for health services planning and evaluating antenatal screening in populations with high risk. The study is also important as it may help to raise the awareness of surgical pediatric intervention and to emphasize the loss of babies with congenital abnor-

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malities.

We are not aware of any study of this nature from Kashmir province in general. In addition since no work has been done on the pattern of presentation of congenital abnormalities in newborns in the area, this study [A hospital based prospective study of the spectrum of congenital abnormalities among neonates of Kashmir province admitted in neonatal unit] was designed to bridge this gap. It was hoped that this would add to the body of knowledge available on these disorders and may stimulate further research in the area on this subject.

Aims and objectives of the study were to study the spectrum of congenital abnormalities seen among the neonates admitted in the special newborn care unit and to determine the various birth and maternal characteristics and to identify any risk factors which may be associated with the occurrence of these abnormalities.

MATERIALS AND METHODS

This study [A hospital based prospective observational study of spectrum of congenital abnormalities among neonates of Kashmir province admitted in neonatal unit] was conducted over a period of one year from 1st April 2013 to 31 March 2014 in the special newborn care unit of the Pediatric Department of Government Medical College Srinagar. All the neonates admitted were included in the study. The diagnosis of congenital abnormality was based on a detailed physical examination of the neurological, cardiovascular, respiratory, abdominal and musculoskeletal systems along with relevant investigations e.g X-Ray chest/abdomen for diaphragmatic hernia, ultrasound examination for renal abnormalities, echocardiography for congenital heart disease and karyotyping for chromosomal disorders (wherever indicated).

A detailed history including the antenatal history, history of exposure to teratogens/drugs, history of consanguinity, maternal age, parity, type of delivery, gestational age, maternal illness and congenital abnormality was sought on a pre designed proforma. The congenital anomalies were classified according to the International Statistical Classification of Diseases and Related Health Problems 10th Revision[6]

Multiple congenital anomalies were grouped depending upon whether those anomalies qualified as a specific syndrome or not. If they qualified as a specific syndrome they were categorized into that syndrome. If two systems were involved, both systems were recorded. When multiple anomalies of a system were present, they were counted as separate.

RESULTS

Out of 4987 admissions during the study period 145 neonates had one or more congenital abnormalities. Out of these 109 had a single anomaly and 36 had multiple anomalies.

Musculoskeletal system defects accounted for majority of the cases 20.8% (n=41), followed by gastrointestinal 17.7% (n=35), CVS 16.75% (n=33), genitourinary 10.65% (n=21), CNS 8.1% (n=16) and others 16.24% (n=32).

Among the musculoskeletal abnormalities CTEV was the most common malformation (n=21). In the GIT cleft palate was most common (n=15). VSD was the most common cardiovascular anomaly (n=13). In the genitourinary system ambiguous genitalia were most frequent (n=10) and in the nervous system congenital hydrocephalus was most common (n=5).

Among the syndromes Down Syndrome was predominant (n=15), followed by Pierrie Robin Sequence (n=7).

MUSCULOSKELETAL/Q65-Q79/41		
Congenital Malformation	**ICD Code**	**Number**
CTEV	Q66	17
POLYDACTYLY	Q69	8
SYNDACTYLY	Q7O.9	4
AMC	Q74.3	1
LIMB REDUCTION	Q71	2
DDAO	Q79.9	2
FACIAL ASYMMETRY	Q67.0	1
CDH	Q79	2
GASTROINTESTINAL/Q35-Q45/35		
CLEFT PALATE	Q35	15
CLEFT LIP	Q36	12
TEF	Q39.1	2
CHPS	Q40	2
Imperforate anus	Q42.3	1
INGUINAL HERNIA	Q45.9	2
MACROGLOSSIA	Q38.2	1
CIRCULATORY/Q20-Q28/33		
VSD	Q21	13
PDA	Q25	4
DTGA	Q20.3	4
TOF	Q21.3	3
ASD	Q21.1	3
AV canal defect	Q21.2	2
TAPVC	Q26.2	2
HLHS	Q23.4	1
TRICUSPID ATRESIA	Q22.4	1
NERVOUS SYSTEM/Q00-Q07/16		
CONG.HYDROCEPHALUS	Q03	5
MENINGOMYELOCELE	Q 05	4
MICROCEPHALY	Q02	4
ANENCEPHALY	Q00	1
CHOROID PLEXUS CYST	Q04.6	1
ENCEPHALOCELE	Q01	1
GENITOURINARY/Q50-Q64/21		
AMBIGUOUS GENITALIA	Q52/55	11
UNDESENDED TESTIS	Q53	6
HYPOSPADIAS	Q54	3
RENAL AGENESIS (u/l)	Q60	1

EYE EAR FACE NECK/Q10-Q18/19		
MICROOPHTHALMIA	Q11.2	2
CONGENITAL ECTROPION	Q10.1	2
PRE AURICULAR TAG	Q17	8
MICROTIA	Q17.2	2
LOW SET EARS	Q17.4	5

OTHERS/SYNDROMES/Q80-99/32		
DOWNS SYNDROME	Q90	15
EDWARDS SYNDROME	Q91	1
THORACOPHAGUS	Q99	1
APERT SYNDROME	Q87	1
LAMELLAR ICTHYOSIS	Q80.2	5
HEMANGIOMA	Q82.5	2
PIERREROBIN Sequence	Q87	7

Among 145 neonates 89/61.37% were males and 56/38.62% were females. LBW neonates were 85 (58.62%) and further distribution in weight category revealed that 51 (35.17%) were between 2.5kg to 4kg and 9 (6.2%)>4kg. Regarding gestational age, 76 (52.41%) were preterm, 55 (37.93%) full-term and 14 (9.65) post-term. Besides, 84 (57.93%) neonates were born by lowersection Caesarean Section (LSCS) and 61 (42.06%) by simple vaginal delivery (SVD). Maternal age parameters revealed that 64 (44.13%) mothers were above 30 years, 24 (16.55%) 25-30 years, 20 (13.79%) 20-25 years, and 37 (25.51%) below 20 years.

DISCUSSION

Prevalence rate of Congenital Malformations in our study was 2.9%. True prevalence of Congenital Malformations 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, prevalence from Nigeria[7] has been reported as 2.7%, Oman[8] 2.46%, Bahrain[9] 2.7% and India[10] 1.5%.

In this study, the most common system involved was the musculoskeletal system (20.8%), GIT (17.7%), CVS (16.7%). These findings were comparable to the studies conducted by other investigators in India, Kuwait[11], Saudi Arabia[12] and Iran.[13,14,15] Some studies, however, recorded a higher incidence of CNS and CVS malformations followed by GIT and musculoskeletal system.[16]

In our study, the rate of CMs outnumbered in males compared to females and was consistent with a study from Brazil (59 % male and 41% in females).[17,18] Another study In India also reported that CMs were more common in males than females (2.1:1 ratio). It was also consistent with results of other studies.

The incidence of CMs in our study was 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 fromBrazil (67% pre-term and 33% term)[17], and others but in contrast with another study which reported tendency of anomalies more common in Pakistan in term neonates.[19]

Association of LBW with increased incidence of anomalies was found in our study and was in accordance with result of other studies in Saudi Arabia, India[20, 21]. We also had LSCS preponderance as the mode of delivery which was consistent with earlier results.

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. One Pakistani study has reported the highest (80.6%) incidence in 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. It indicates that the incidence of CM increases as the birth order increases. Likewise H/O maternal illness was associated with congenital anomalies in 54.4% of neonates and this association is well supported by studies done earlier. Folic acid supplementation was absent in 13 cases, among them one was a case of meningocoele. None of the mothers was a smoker nor was their h/o alcohol intake during pregnancy

LIMITATIONS

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 neurosurgical facilities and many cases of CNS/surgical are likely to have been missed.

CONCLUSION

CMs are not rare in our set-up and MS was the most commonly affected system in our study. Prematurity, LBW, male gender, , advanced maternal age 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 for better outcome of these neonates

ABBREVIATIONS

ASD	Atrial septal defect
AMC	Arthrogryposis multiplex congenita
CDH	Congenital diaphragmatic hernia
CHPS	Congenital hypertrophic pyloric stenosis
CM	Congenital Malformation
CNS	Central nervous system
CTEV	Congenital talipesequinovarus
CVS	Cardiovascular system
DDAO	Deficiency of Depressor AnguliOris
DTGA	Dextro transposition of great arteries

GIT	Gastrointestinal tract
HLHS	Hypoplastic left heart syndrome
ICD	International classification of diseases
LBW	Low birth weight
LSCS	Lower segment caesarian section
PDA	Patent ductus arteriosus
SGA	Small for gestational age
TAPVC	Total anomalous pulmonary venous connection
TEF	Tracheoesophageal fistula
TOF	Tetrology of Fallot
VSD	Ventricular septal defect

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