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Prevalence of Congenital Malformations in Newborns Delivered in a Rural Medical College Hospital, West Bengal

Dr. Abhay Charan pal[1], Dr. Dipto Kanti Mukhopadhyay[2], Dr. Debasis Deoghuria[3], Dr. Sumanta Kumar Mandol[4], Dr. Aparesh Chandra Patra[5], Dr.Shibsankar Murmu[6]

1Associate Professor, Department of Pediatric Medicine; BS Medical College, Bankura, West Bengal
2Associate Professor, Dept. of Community Medicine; College of Medicine and Sagar Dutta Hospital, Kamarhati: Kolkata -58.
3Associate Professor, Dept. of Radiodiagnosis, BS Medical College, Bankura, West Bengal, India.
4Associate Professor , Dept. of Radiodiagnosis, BS Medical College, Bankura, West Bengal, India.
5Associate Professor , Dept. of Dermatology, BS Medical College, Bankura, West Bengal, India.
6Post-Graduate Trainee, Dept of Gyne and Obst; BS Medical College, Bankura, West Bengal .

Abstract: *Congenital malformations are among leading cause of mortality and morbidity among neonates and also beyond neonatal age group. An institution - based cross –sectional, observational study was conducted in B.S Medical college, Bankura, west Bengal, India. Among total 14079 neonates included in the study, prevalence of congenital malformations was 2.3%. In the present study cardiovascular, musculoskeletal, and genitourinary system were found to be most commonly involved. Different maternal risk factors were also studied as well as the role of preventive interventional strategies. This study helps us to know the pattern of congenital malformation in this part of country.*

Key Words*: Congenital malformations; cross-sectional study; observational study; prevalence and pattern of malformations; India*

I. Introduction

Congenital abnormality refers to any abnormality, whether genetic or not, which is present at birth(1). It can also be defined as abnormality of physical structure or form seen at birth or few weeks after birth (2).

Structural defects of prenatal origin are classified into the following three groups, according to the cause, timing and extent of the developmental disturbance:

- Malformations (defective organogenesis)
- Dysplasia (abnormal cell or tissue structure)
- Deformation (mechanically induced changes of normal structure) (3).

Primary malformations are caused by endogenous disturbances of primordial tissues. Secondary malformations (disruption) arise when organs develop abnormally from a normal primordium.

Major congenital abnormalities are structural defects of the body and organs that impair viability and require intervention. Minor congenital abnormalities are small structural developmental disturbances that do not impair viability and do not need to be treated.

A distinction is drawn between singular (isolated) malformation, combined malformations (more than one malformation in a single organ category in one individual), multiple malformations (more than one malformation in different organ system in one individual) and syndromes (combination of developmental defects showing a common etiology). Sequences are developmental defects due to cascade of events originating from a primary morphological defect. Associations are non-random combination of defects of unclear etiology and without any apparent heritability (1, 3)

About 20% of all major congenital malformation are genetically transmitted by a monogenetic abnormality ,5-10%b are due to chromosomal anomalies ,2-10% are due to viral infection . In about 60% the cause is unknown and appears to multifactorial (4).

Exogenous etiological factors include teratogenic medicines like vit-A derivatives and maternal metabolic disease such as diabetes mellitus. Toxic effects on the human embryo has been demonstrated for the following substances. ; alcohol, androgen, carbamazepine , coumarin derivatives,iodine(overdose),cocaine,polychlorinated,biphenylsphenobarbitol/primidone,phenytoin,retinoids,thalidomide,valproic acid and cytostatic agents.Ionising raditions is also embryotoxic(5).

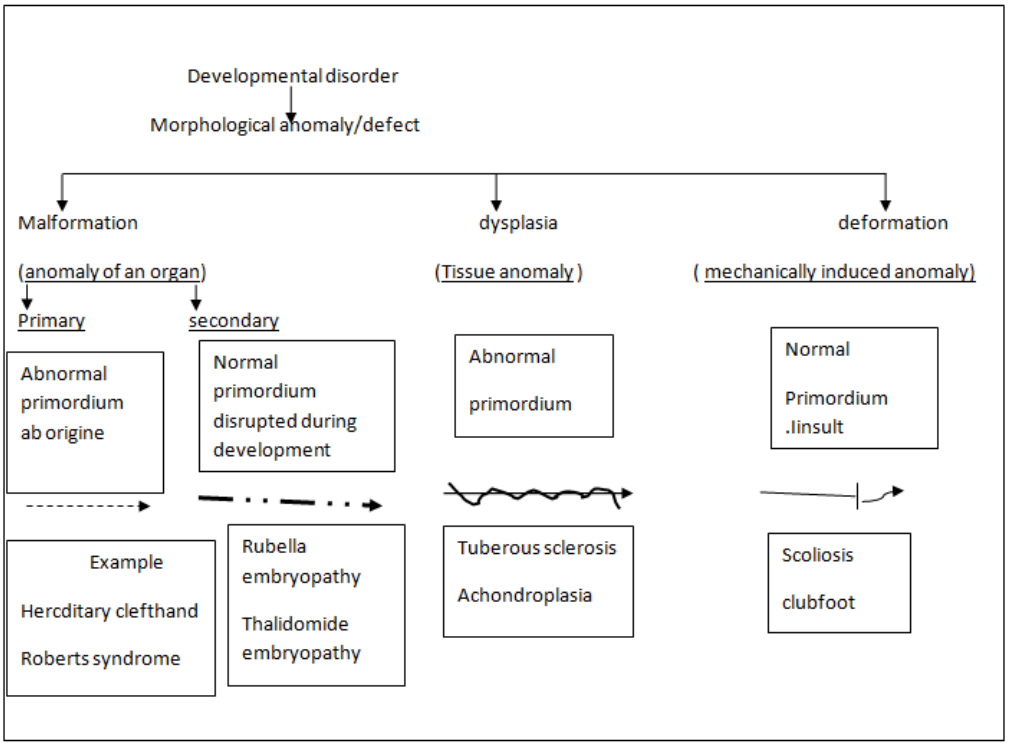

Diagram -1 The pathological mechanism and nomenclature of developmental disorders:

A malformation is the result of abnormal organogenesis. A primary malformation is due to an abnormality of the organ primordium ab origine e.g. because of a genetic mutation. Secondary malformations are caused by an external event (teratogen) interfering with the previously normal course of development. A dysplasia is a disorder of a particular type of tissue that can be observed in all organs where this tissue is present. Dysplasia may be evolutive (primary) or degenerative (secondary). A deformation is a mechanically induced change of an organ. If the mechanical impediment can be removed the organ continues to grow normally

By International convention frequency of congenital malformation is reported as prevalence rather than incidence, as congenital malformations are not newly arising disease in the usual sense ; but rather disorders affecting a given population at a given moment of time (the time of birth). Prevalence of major malformation has been variously reported as 3-4% to 6-8%(6,7). Optimally designed active surveillance systems demonstrate higher prevalence. About one fifth of all such malformations are severe and life threatening (8, 9, 10)

Congenital abnormalities plays a major role in a morbidity and mortality of neonates and children (11). Due to high cost of treatment and rehabilitation of these anomalies, early identification of causative and risk factors and early prevention is necessary where possible (11). In the tropical countries malnutrition and infection are main causes of infant morbidity and mortality while in temperate zones cancer, accidents and congenital abnormalities are the key causes of infant morbidity and mortality (11).

Prevalence studies of congenital malformation are useful to establish baseline rates, to document changes over time and to identify clues to etiology. They are also important for health service planning and evaluating antenatal screening in population with high risk. Such studies are important as those help to raise the awareness of surgical pediatric intervention and to emphasize the loss of babies with congenital malformation (12).

The present study was conducted with an intention to determine the prevalence of congenital malformation among the newborns delivered in the department of Gynecology and Obstetrics B.S Medical College, Bankura, West Bengal, India. It is hoped that this study will add to the knowledge available on the subject.

II. Objective of the study

In the developed countries congenital malformations are a leading cause of death . Though in developing countries like India the leading cause of neonatal mortality is infection and low birth weight; in the coming decades due to improved perinatal and neonatal care, mortality due to sepsis and low birth weight will be reduced remarkably and congenital malformation will become a leading cause of neonatal mortality .

The present study was conducted with the objective to determine the overall prevalence of congenital malformation, prevalence in live birth and still birth as well as involvement of various organ systems at a Rural Medical College Hospital in West Bengal and to compare them to previous studies in this field.

III. Materials and methods

This prospective study was conducted in the department of Pediatrics Medicine, B S Medical College, Bankura; a rural Medical College Hospital in West Bengal . The study was conducted in collaboration with the department of Gynecology and Obstetrics. All the intramural deliveries between May 2014 to June 2015 comprised the study material. There were total 13791 live births and 288 still births during this period. The study material comprised 14079 births and there were 13986 mothers (93 mothers give birth twin babies). All the newborn (liveborn and stillborn) were looked for congenital abnormalities soon after birth (after stabilization and initial resuscitation as required) and everyday during routine ward round . Relevant information regarding maternal age , gestational age , sex, community , birth weight , birth order and consanguinity were documented . Antenatal history like maternal illness , ingestion of drugs , exposure to radiation and complication of labor was recorded. Antenatal ultrasonographic (USG) findings were noted . Relevant radiological, histohematological and genetic tests were carried out. Autopsy on stillborn and neonates who died during hospital stay were done where parents consents could be obtained

A meticulous general and systematic examination was carried out by a consultant at the time of birth to detect any malformation. Ultrasound was employed whenever necessary to detect multiple congenital anomalies and to rule out majority of the internal congenital anomalies. Echocardiography with color Doppler was also used for all suspected congenital cardiac problems. Other investigations e.g intravenous urography was done when needed . C.T and MRI was also done for certain specific cases. Malformations were divided into major and minor ; major malformation (6) interfere considerably with the function of all or part of the infant , minor malformations give no serious medical or cosmetic consequences to the patients . The major malformations were divided into central nervous system (CNS), muscular skeletal, gastrointstenial , genitourinary, cardiovascular system (CVS), syndromes, associations , and miscellaneous disorders

Data analysis was done using SPSS 13. Rates and proportions were calculated with 95% confidence intervals. The proportions were compared using standerd T-test . Level of significance was set at $P<0.05$. Ethical approval of the study and consent to publish the clinical data derived from the study have been obtained from the Ethics Committee of BS Medical College, Bankura, West Bengal, India

IV. Results

Out of total 14079 deliveries 13791(98%) were livebirths and 288 (2%) were stillbirths. The number of babies with congenital malformations diagnosed at birth or within the first week of life was 328(2.3%). Table 1 gives the frequency and sex distribution of congenital malformations. Out of the 13893 singleton babies 305 (2.2%) were malformed whereas 23 of 93 pairs of twin has birth defect (12.4%). The sex wise distribution was 62 % males and 38% females giving a M: F ratio of 1.63:1.00, and there were 5 cases of ambiguous genitalia. Congenital malformations were seen more significantly in stillbirth, 2.7 times more frequently as compared to livebirth , the frequency being 5.9% and 2.3% respectively . 17 of the 328 malformed babies (5.18%) were stillborn. Table 2 shows the frequency of congenital malformations in relation to fetal and maternal factors. Women less than 20 years has 1.5 % babies with congenital anomalies and the mothers of babies with congenital anomalies were mostly between 20 and 30 years i.e. 90.2% , and 8.3% of the mothers were above 30 years .

History of parental consanguinity was present in 21 cases (0.15%) in our study. Babies with congenital anomalies were of the first order (34.6%) and second order to third order (51.9%)

More than four or, fourth birth order was associated with 12.5% of the anomalies. There was a history of oligohydramnios in 20/328(6.1%) cases and polyhydramios in 11/328(3.35%) cases. Also 31/328 mother (9.5%) had a history of previous abortions; 10/328(30%) where diabetic mothers and 8/328(2.4%) had a history of congenital heart disease in previous child or malformed babies

Table 3 shows the systemic distribution and the prevalence of individual congenital malformations. Cardiovascular malformations were most common among livebirth followed by musculoskeletal malformations. The CNS defect were most commonly seen in stillborn

V. Discussion

The prevalence of congenital malformation in the present study was 2.03% which is comparable with other studies (13, 14, 15, 16) from different parts of the country. The number of documented birth defects in infant is increasing antenatally and during neonatal period due to advanced diagnostic technology, especially USG and echocardiography. When autopsies are performed in Hospitals, the prevalence of birth defect is up to 3

times higher. Higher autopsy rates at Chandigarh and Pondichery centers reported a higher prevalence of congenital malformations (18, 19).

This study reported a higher prevalence of anomalies in still birth (5.9%), which is in accordance with some previous study (15, 16, 18, 20). Association of low birth weight with increased risk of congenital malformation was noted in this study which is in concordance with previous studies (17, 18, 19, 20)

The prevalence of congenital malformations was significantly higher in preterm babies as compared to full term neonates (21). Previous studies have reported male preponderence among congenitally malformed babies (19, 20), which was statistically insignificant in our study. Previous data showed a definite increase in prevalence of congenital malformation in babies born to consanguinous marriage (21). 21 cases had a history of consanguinity in our study. This study has statistically shown that mothers, above 30 years of age stand at a higher risk of producing malformed babies. Sagunabai et al (22) reported that mothers' age more than 35 years have a higher risk of giving birth to malformed babies where as Datta et al (18) documented statistically insignificant association of increased maternal age and congenital malformation.

Previous studies (19) have reported significantly higher prevalence of malformation among the mothers of gravida 4 or more and our results are consistent with that finding. This indicates that as the birth order increases there is an increased risk of congenital malformations. The previous studies evaluated the factors that significantly increase the risk of congenital malformations in newborn and those included the presence of hydramnios,maternal febrile illness in the first trimester , past history of abortions , diabetic mothers , eclampsia, previous abortions and history of congenital heart diseases in previous child or history of malformed babies ,.Certain maternal disease may occasionlly lead to increased risk of birth defects . According to Ordonez et al(23) diabetes mellitus, arterial hypertension and hypothyroidism show a positive association with congenital malformation. The main aim of this study was to plan measures for maternal and child health, with a main focus on prevention of congenital malformations, by health education, adequate prenatal care and organization of referral networks for major anomalies.

The annual report of Indian Council of Medical Research says that the commonest congenital malformations are cardiac (0.57%) in nature (24). Our study conforms to that. Low prevalence of cardiovascular defects at birth is reported by many studies in literature, given that this diagnosis is usually made after discharge from the hospital (25). But in our study Echocardiography with Color Doppler was done in neonates of mother having a history of high risk pregnancy. Kalra et al(26),reported that the CNS defects have the highest prevalence where as Sagunabai et al (22) reported gastrointestinal malformations to rank the highest .

Mathew et al (21) reported that musculoskeletal abnormalities were the commonest. The present study showed that cardiovascular, musculoskeletal and genitourinary were the most commonly affected system in a decending order of frequency. With regard to the cardiovascular system, ventricular septal defect was the most common lesion found in high risk mothers who had history of previous child with congenital heart disease, diabetics mothers or those with previous congenitally malformed babies.

Congenital talipes equinovarus was the commonest musculoskeletal abnormalities observed in the present study. Among the genitourinary tract anomalies, hypospadias, undescended testis and polycystic kidney were the most prevalent lesions . Regarding the central nervous system the most prevalent anomaly encountered was microcephaly , Dandy Walker malformations and meningomyelocele . With special reference to the neural tube defect (NTD), the prevalence of NTD has markedly reduced in the developed countries following mass promotion and mandatory prescription of folic acid for pregnant mother.

The present study helps us to know the pattern of congenital malformations prevalent in this part of West Bengal. Observations made in this study also help us to know the possible correlation of various factors as to the cause of congenital anomalies. Most of the observations are comparable with the similar studies undertaken in other parts of the country. However some of the observations differ which is expected given the nature of various studies like hospital versus community based , differences in geographical and environmental factors , differences in time period for follow up , criteria for classification used etc.

VI. Conclusion

The study definitely helps us to know the pattern of congenital anomalies and the relationship of various gestational and familial factors in relation to congenital anomalies and to plan future strategies for prevention, early diagnosis and timely management.

VII. Contributions

Dr. Pal actually planned, conducted and supervised the study along with contribution of important intellectual contents. He also drafted the manuscript.

Dr.Mukhopadhyay revised the manuscript and preformed the statistical analysis

Dr.Deoghuria and Dr.Patra added some important intellectual contents and provided all relevant helps needed for the study

Dr.Mandol conducted radiological studies and also added some intellectual contents
Dr.Murmu assisted Dr.Pal at all steps in the work alongwith contributing some important contents
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Finding – None
Competing interest – None

Table 1. Distribution of babies with congenital malformations according to birth outcome and sex

	No	Malformed babies		OR(95%C.I)
		No	%	
Live births	13791	311	2.3	1.00
Shill births	288	17	5.9	2.72(1.59-4.59)
Female	6854	125	1.8	1.00
Male	72220	203	2.8	1.56(1.24-1.96)
Ambiguous	5	0	0	NA
Total birth	14079	328	2.3	

Table 2- Distribution of babies with congenital malformations according to fetal and maternal factors

Factor	Category	Total No.	Malformed babies		Chi –square for linear trend (p value)
			No.	%	
Birth weight (Gms)	<1000	51	3	5.9	34.77(0.00000052)
	1000-1499	282	9	3.2	
	1500-1999	1021	48	4.7	
	2000-2499	4218	75	1.8	
	>2500	8507	193	2.3	
Period of gestation	Pre –term	585	36	6.2	24.895(<0.00001)
	Term	13014	283	2.2	
	Post –term	480	9	1.9	
No. of fetus	Single	13893	305	2.2	83.43# (<0.000001)
	Twin	186	23	12.4	
Total		14079	328		
Maternal age in years *	<21	841	4	0.5	80.145 (<0.00001)
	21-25	9198	200	2.2	
	26-30	3311	54	1.6	
	>30	636	70	11.0	
Parity *	Primi	4981	103	2.1	34.595 (<0.00001)
	2-3	8662	176	2.0	
	>3	343	49	14.3	
Total		13986	328		

*93 were twin deliveries
#chi-square test was applied.

Table 3:-Distribution and prevalence of individual congenital malformations

Type of defect	Total number	Rate /1000 Births
Cardiovascular system		
• Acyanotic CHD	44	3.08
• Cyanotic CHD	18	1.26
• Complex CHD	10	0.70
Central nervous system		
• Microcephaly	5	0.35
• Dandy walker malformation	5	0.35
• Hydrocephalus	4	0.28
• Meningoencephalocele	2	0.14
• Meningomyelocele	4	0.28
• Spina bifida	4	0.28
• Encephalocele	2	0.14
• Meningocele	2	0.14
Kidney		
• Polycystic Kidney	7	0.49
• Hydroureter	3	0.21
• Posterior urethral valve	6	0.42

Genital system		
• Hypospadias	10	0.70
• Micropenis	8	0.56
• Ambigious genitalia	6	0.42
• Congenital hydrocele	3	0.21
• Undescended testis	8	0.56
• Epispadias	4	0.28
Gastrointestinal system		
• Diaphragmatic hernia	6	0.42
• Duodenal atresia	3	0.21
• Omphalocele	2	0.14
• Extrophy of bladder	3	0.21
• Exomphalos	1	0.07
• Imperforate anus	4	0.28
• Gastroschisis	1	0.07
• Tracheo esophageal fistula	4	0.28
• Cleft lip/pala.te	16	1.12
Musculoskletal system		
• Craniosynostosis	6	0.42
• Talipes	15	1.05
• Hemimelia	5	0.35
• Polydactyly/syndactyly	24	1.68
• Osteogenesis imperfacta	4	0.28
• Hemivertebrae	3	0.21
Syndrome		
• TAR syndrome	2	0.14
• Pierre Robin Syndrome	1	0.07
• Prune Belly Syndrome	1	0.07
• Down syndrome	8	0.56
Respiratory system		
• Laryngomalacia	2	0.14
• Choanal atresia	1	0.07
• Pulmonary hypoplasia with congenital diaphragmatic hernia	2	0.14
Skin		
• Skin tag over face and hand	6	0.42
• Preauricular tag	9	0.63
• Hemangioma	8	0.56
• Giant hairy nevus	2	0.14
Eye		
• Anophthalmia	2	0.14
• Micropthalmia	3	0.21
• Congenital ptosis	2	0.14
Others		
• Sacrococcygeal sinus	2	0.14
• Single umbilical artery	12	0.84
• Miscellaneous	13	0.91

Table 3 Continues

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