

A hospital-based cross-sectional study on congenital anomalies in newborn babies of Uttarakhand

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ABSTRACT

Background: Congenital anomalies make an important contribution to infant mortality. They remain a leading cause of death in many countries of the world. The prevalence and pattern of presentation vary from place to place. **Objective:** The aims and objectives of this study were to determine the prevalence of congenital abnormalities among babies (inborn and outborn) admitted at the Himalayan Hospital, Dehradun, Uttarakhand, India. **Materials and Methods:** A cross-sectional retrospective study was conducted in a Medical College Hospital in Uttarakhand. The case files of these babies were retrieved from the hospital records department and examined individually by the investigators. Data collection was done with structured forms designed for the study. **Result:** Among total 2242 neonates included in the study, the prevalence of congenital malformations was 8.3%. Central nervous system and cardiovascular, musculoskeletal, and genitourinary systems were found to be most commonly involved. **Conclusion:** The present study provides us an idea regarding prevalence of cases of congenital malformations and factors affecting it. In Indian environment, this study helps us to know the pattern of congenital malformation in this part of the country.

Key words: *Congenital, Abnormalities, Newborns*

Congenital anomalies are also known as birth defects, congenital disorders, or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies (e.g., metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or later in life [1]. A malformation is the result of abnormal organogenesis. The primary malformations can be due to genetic factors e.g., congenital heart disease. Secondary malformations are caused by an external event (teratogen) interfering with the previously normal course of development [2].

Congenital anomalies can result in long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies. Some congenital anomalies can be prevented, for example, by vaccination, adequate intake of folic acid or iodine through fortification of staple foods or provision of supplements, and adequate antenatal care. The causative factors of birth defects are diverse – these include genetic abnormalities, teratogens in environment, maternal malnutrition, and intrauterine infections [1,3]. The birth prevalence of congenital anomalies in the developing world is underestimated by deficiencies in diagnostic capabilities and lack of reliability of medical records and health statistics. As a result, recorded diagnoses in vital statistics focus on overt acute illnesses, rather than on preexisting congenital conditions that increase vulnerability to infections and malnutrition (WHO, 1985).

There are more than 4000 different birth defects, in which some of the defect is treated and cured and other are not treated, leading to death in the first year of life. An estimated 276,000 babies die within 4 weeks of birth every year, worldwide, from congenital anomalies [3]. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India. Prevalence studies of congenital malformation are useful to establish baseline rates, to document changes over time, and to identify clues to etiology [3,4]. They are also important for health service planning and evaluating antenatal screening in population with high risk. The objective of this study was to determine the prevalence of congenital abnormalities among babies (inborn and outborn) admitted to Himalayan Hospital, Dehradun, Uttarakhand, India. It is hoped that this study will add to the knowledge available on the subject.

MATERIALS AND METHODS

A cross-sectional, retrospective study was conducted in the Department of Pediatrics, Himalayan Institute of Medical Sciences, Dehradun, a Medical College Hospital in Uttarakhand. The study was conducted in collaboration with the Department of Gynecology and Obstetrics. All the intramural deliveries and admitted newborns in neonatal intensive care unit of Pediatrics Department between May 2012 and April 2014 comprised the study material. The case

files of these babies were retrieved from the hospital records section and examined individually by the investigators. Data collection was done with structured forms designed for the study. A total of 2900 records were examined, out of which 2242 complete records were included in our study and rest of them was excluded from the study.

The diagnosis of congenital abnormality was based on clinical evaluation and relevant radiological investigations including ultrasound, X-rays, echocardiography, and neuroimaging. Patient's history, including antenatal history, history of exposure to teratogens, and family history of consanguinity were obtained from these records. Further information included maternal age, type of delivery, gestational age, and type of congenital abnormality. The prevalence rate was estimated as a percentage of the total number of babies admitted to the unit within the period of the study (number of babies with congenital abnormalities/total number of babies admitted to the hospital for the duration of the study).

Data analysis was done using SPSS 20. Rates and proportions were calculated with 95% confidence intervals. The proportions were compared using Z-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 has been obtained from the Ethics Committee of Himalayan Institute of Medical Sciences, Dehradun, Uttarakhand, India.

RESULTS

Out of 2900 records examined, 2242 neonates (inborn and outborn) with complete records were included in the study. 1296 (57.80%) were males and 946 (42.19%) were females. The prevalence of congenital anomalies was 8.39% (188/2242 total births). The most commonly detected congenital anomalies were those of the central nervous (2.32%) and gastrointestinal system (2.01%), followed by cardiovascular (1.74%), musculoskeletal (0.94%), genitourinary (0.76%), and respiratory system (0.54%), others (0.31%), and skin (0.04) as shown in Table 1.

Mode of delivery has a significant ($p=0.001$) association with congenital anomalies in this study, with cesarean section being more commonly associated than normal delivery (Table 2). Family history of birth deformity ($p=0.009$) and increasing parity ($p < 0.001$) were positively associated with congenital malformation. Congenital abnormalities were highest in para 1-3 mothers (103 cases) which were highly significant. Higher maternal age (above 35 years) was significantly ($p=0.009$) associated with congenital malformation. Birth weight ($p=0.001$) and period of gestation ($p=0.025$) were also positively associated with congenital malformation. Prematurity and low birth weight (LBW) were found to have a higher risk of congenital anomalies. However, gender ($p=0.587$) was not significantly associated with congenital malformation.

DISCUSSION

In our study, the prevalence of congenital anomalies among 2242 hospital live births was 8.39% (188/2242 total births). The

Table 1: Pattern of major congenital anomalies

Type of anomaly	Congenital anomalies		
	Male	Female	Total
Musculoskeletal system (talipes, spina bifida, polydactyly, syndactyly)	12 (0.93)	9 (0.95)	21 (0.94)
Central nervous system (meningomyelocele, anencephaly, hydrocephalus)	31 (2.39)	21 (2.22)	52 (2.32)
Gastrointestinal system (tracheoesophageal fistula, harelip, inguinal hernia imperforate anus, cleft palate)	26 (2.01)	19 (2.01)	45 (2.01)
Skin (preauricular tag, hemangioma, others)	1 (0.08)	0 (0)	1 (0.04)
Genitourinary system (coronal hypospadias, hydronephrosis)	13 (1)	4 (0.42)	17 (0.76)
Cardiovascular system (VSD, ASD, TOF, TGA)	21 (1.62)	12 (1.27)	33 (1.47)
Respiratory system (choanal atresia, adenomatoid, etc.)	7 (0.54)	5 (0.53)	12 (0.54)
Others	5 (0.39)	2 (0.21)	7 (0.31)
Total number of cases	116 (8.95)	72 (7.61)	188 (8.39)

VSD: Ventricular septal defect, ASD: Atrial septal defect, TGA: Transposition of great arteries, TOF: Tetralogy of Fallot

Table 2: Association of various maternal and fetal factors to the causation of congenital malformations

Parameters	Congenital malformation (N=2242)			p value
	Absent	Present	Total	
Maternal factors				
Maternal age (years)				
<25	832	72	904	0.009
25-35	741	54	795	
>35	481	62	543	
Mode of delivery				
Normal	1321	63	1384	0.001
Caesarean	733	125	858	
Family history of birth deformity				
No	1408	111	1519	0.009
Yes	646	77	723	
Parity				
Primi	732	24	756	0.001
1-3	1001	103	1106	
4 and more	319	61	380	
Fetal factors				
Gender				
Male	1180	116	1296	0.2804
Female	874	72	946	
Birth weight (kg)				
<2.5	730	103	833	0.001
>2.5	1324	85	1409	
Period of gestation				
Pre-term	621	72	693	0.025
Term	1433	116	1549	

frequency of congenital anomaly in our study was lower than that reported by Ghosh et al. [4] and the Birth Defect Registry of India [5]. However, it was higher than the frequencies reported in Maharashtra [4] and Shimla [6]. In our study, gender of the fetus had no effect on the prevalence of congenital anomalies, which concurs with the findings of Mishra and Baveja [7], Ali et al. [8], and Swain et al. [9]. However, many studies have demonstrated male predominance among congenital malformed babies [10,11]. Because of the high frequency of the central nervous system, gastrointestinal system, and cardiovascular system defects as shown in our study, we recommend their proper prenatal diagnosis both by abdominal sonography and echocardiography.

Association of LBW with increased risk of congenital malformations is very well documented [12]. Our finding is in accordance with that. The incidence of congenital anomalies was significantly higher in preterm babies as compared with the full-term babies, which is in conformity with the previous studies reported from this country [13]. Mode of delivery also showed a significant association with congenital anomalies in this study, with the cesarean section being more commonly associated than normal delivery. Higher prevalence of malformation in the babies born to mothers aged over 35 years reported by Suguna Bai et al. [14], whereas Dutta and Chaturvedi [15] documented statistically insignificant association of increased maternal age and congenital anomalies. In our study, a majority of malformed babies were born to mothers aged more than 35 years.

We thus recommend that all neonates should be thoroughly examined and investigated for congenital anomalies. In addition, a registry program of congenital malformations is needed; appropriate health education about consanguinity and genetic counseling for consanguineous couples should also be established before marriage. As it is a tertiary care hospital and referral center, prevalence calculated may be higher than the general population in this hospital-based study. Hence, the data cannot be projected to the general population, for which population-based studies are necessary. Second, we could not include the abortions and stillborns because often the abnormalities are not obvious or visible externally. In those cases, a pathological autopsy is warranted, and in most of the cases, parental consent is not available for pathological autopsy. In addition, we could not subclassify the diseases being a retrospective study.

CONCLUSION

Congenital anomalies make an important contribution to infant mortality. They remain a leading cause of death in many countries

in the world. The study definitely helps 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.

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