



# Prevalence and pattern of birth defects in a tertiary care hospital in Kashmir: A pilot study

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## ABSTRACT

### Research Question

What is the prevalence and types of birth defects in a Kashmiri population?

### Aims and Objectives

The present study was undertaken to determine proportion of structural birth defects occurring among institutional births in a tertiary care hospital.

### Materials and Methods

A cross-sectional descriptive study was undertaken in a tertiary care hospital, where outcome of all institutional deliveries was assessed in terms of birth defects detected within 24 hours of delivery. Diagnosis of congenital anomalies was based on clinical evaluation of a newborn by a **neonatologist**.

### Results

Out of 1146 women who delivered in the hospital, 1129 were live births, 12 still births and 5 intrauterine deaths; with 617 (53.8%) males and 529 (46.2%) females. Birth defects were identified in 17 (1.48%) newborns. Leading defects were related to central nervous system (29.41%), cardiovascular system (17.64%) and genitourinary system (17.64%) respectively. Female: male ratio of birth defects was 1.8:1.

### Conclusions

This study has highlighted the prevalence and types of birth defects seen in our region. Regular antenatal visits and prenatal diagnosis are recommended for prevention.

**Keywords:** Prevalence, Birth Defects, Newborn, Central Nervous System, Prevention

## INTRODUCTION

Congenital anomalies, also known as birth defects can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth. The term congenital malformation is restricted to structural defects at birth. Birth defects affect approximately 1 in 33 infants and result in 3.2 million birth defect related disabilities every year.

An estimated 270,000 newborns die during the first 28 days of life every year from birth defects.<sup>1</sup>

Presence of birth defects is a global problem, but their impact is particularly severe in middle and low-income countries where mortality due to serious birth defects approaches 95%.<sup>2</sup> Besides, birth defects may result in long-term disability, which may have

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significant impacts on individuals, families, health-care systems and societies.

Worldwide about 7.9 million children (6%) annually are born with a serious birth defect.<sup>3</sup> India, with its vast population of 1.2 billion and approximately 27 million births per year, possibly contributes to about one fifth of these defects.<sup>4,5</sup> Most of the available Indian studies, including the data available from Birth Defect Registry of India (BDRI)<sup>6</sup> show that the common systems involved in birth defects are central nervous system, musculoskeletal system and cardiovascular system, with neural tube defects being the commonest.<sup>7</sup>

Although birth defects may be genetic, infectious or environmental in origin, most often it is difficult to identify the exact causes. Many birth defects can be prevented. Penrose (1961) stressed that major advances in the prevention of defects may be achieved by attention to environmental factors rather than by attempting to improve heredity.<sup>8</sup> Vaccination, adequate intake of folic acid and iodine, and adequate antenatal care are keys for prevention.

Lack of epidemiological data prevents inclusion of birth defects under the scope of preventive strategies. Since there is paucity of data regarding the prevalence and type of birth defects in our region. So the present study was undertaken to have the baseline data of birth defects among institutional births in a tertiary care hospital.

#### OBJECTIVE

To determine the pattern and prevalence of structural birth defects in newborns in a tertiary care hospital.

#### MATERIAL AND METHODS

A cross-sectional descriptive study was conducted in a neonatal care unit of a tertiary care teaching hospital which provides specialist tertiary care services to patients largely belonging to lower and middle socio-economic strata of the society with both rural and urban background. Normal deliveries are discharged within 24-48 h while mothers with surgical interventions are discharged in about 5-7 days. The variables included in the study were type of

birth defect, residence, booked/un-booked antenatal status of mother, estimated period of gestation, gravida, sex of newborn, type of delivery and survival outcome at 24 h of birth. Diagnosis of congenital anomalies was based on clinical evaluation of a newborn by a neonatologist. Birth weights  $\geq 2.5$  kg was considered to be normal and birth weight  $< 2.5$  kg as low birth weight. Babies with birth weight less than 1.5 kg were labeled as very low birth weight and those with less than 1kg as extremely low birth weight. Babies born at  $< 37$  completed weeks (i.e.,  $< 259$  days), calculated from the 1<sup>st</sup> day of last menstrual period were considered as premature. Babies born less than 28 weeks gestational age were named as having extreme prematurity, 28 to 32 weeks as very preterm, 32 to 37 completed weeks as late preterm. Small for gestational age was defined as a newborn with birth weight less than 10<sup>th</sup> percentile for gestational age.

#### Statistical analysis

Data was entered into excel data sheet and appropriate statistical analysis was performed. Proportion was calculated and the association was tested with Chi-square test and Fisher's exact test.  $p < 0.05$  was considered to be statistically significant.

#### RESULTS

A total of 1146 women who were delivered in the tertiary care hospital were taken for the study. Out of this number there were 1129 live births, 12 still births and 5 intrauterine deaths; with 617(53.8%) males and 529(46.2%) females. Mode of delivery was by normal mode in 949(82.8%) of cases and by caesarian section in 197(17.2%) of cases. There were 43(3.75%) preterm deliveries conducted by either normal mode or caesarian section. Twenty three pairs of twins were delivered. Low birth weight babies constituted 55 (4.8%) of total cases.

Birth defects were identified in 17 (1.48%) newborns. Identified malformations are shown in Table 1. Leading defects were related to central nervous system (29.41%), cardiovascular system (17.64%) and genitourinary system (17.64%) respectively. Female: male ratio of birth defects was 1.8:1. Higher risk of malformed births were observed amongst pre-term (4.65%) vs. term (1.35%); cesarean section (3.55%) vs



vaginal delivery (1.05%); low birth weight( 7.27%) vs normal weighed babies( 1.19%). Details are shown in Table 2.

**Table 1 Different Type and Percentages of Birth Defects**

Birth defect	Number (N= 17)	Percentage (%)
CNS(Ancephaly, meningocele, microsomia)	5	29.41
Congenital Heart disease ( VSD,TPGV)	3	17.64
Ambiguous Genitalia	3	17.64
Multiple limb defect	2	11.76
Umbilical hernia	2	11.76
Cleft palate	1	5.88
Downs syndrome	1	5.88

**Table 2 Relation of Birth Defects with sex, gestational age, mode of delivery and birth weight**

	Birth Defect		Normal		Total	P Value
	N	%	N	%		
<b>Sex</b>						
Male	5	1.134	610	98.86	615	0.000
Female	9	1.89	519	98.12	528	
Unknown*	3	100	0	0	3	
<b>Preterm</b>	2	4.65	41	95.34	43	0.268
<b>Twins</b>	2	4.35	44	95.65	46	0.309
<b>Mode of Delivery</b>						
Normal	10	1.05	939	98.94	949	0.021
Caesarian	7	3.55	190	96.44	197	
<b>Birth Weight</b>						
≤2.5 Kg	4	7.27	51	92.72	55	0.002
>2.5 kg	13	1.19	1078	98.80	1091	

\*Unknown sex was because of ambiguous genitalia which was among the leading birth defects.

## DISCUSSION

Birth defects are important causes of still births and infant mortality, and are contributors to childhood morbidity. The actual prevalence is underestimated as intrauterine deaths and stillbirth rates are high. The prevalence of birth defects in the present study was 1.48%, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9%.<sup>9,10</sup> There are reports from different parts of the world representing different frequency of birth defects.<sup>11,12</sup> Tertiary care hospitals usually do not have definite catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Community based study should be ideal for true estimation of incidence of congenital anomalies in a population.

More female babies with birth defects than males were noted in the present study, which was in contrary to other studies.<sup>13</sup> With regard to pattern of birth defects in the study, the most common system involved was central nervous system (29.41%), followed by cardiovascular system (17.64%) and genitourinary system (17.64%) etc. This was comparable with studies conducted by others.<sup>11,14</sup> Some studies however recorded higher incidence of musculoskeletal malformations,<sup>15-20</sup> whereas Suguna Bai *et al*<sup>21</sup> reported GI malformations as the most common one. Under diagnosis is especially true for congenital heart diseases at birth even in developed countries, as some congenital heart diseases present late.<sup>22</sup>



Association of low birth weight with increased risk of congenital malformations was very well documented.<sup>23</sup> Our finding is in accordance with this. The prevalence of congenital anomalies was higher in preterm babies as compared to full term babies but the difference was not statistically significant.<sup>20</sup> LBW was found to have a higher risk of congenital anomalies. The occurrence was about 3.5 times more in case of preterm delivery as compared with the term ones. Mode of delivery was also significantly associated with congenital anomaly and it was more in case of cesarean deliveries.

In 2013, India launched a service for screening and referral of children with nine specific types of birth defects (neural tube defects, Down syndrome, orofacial clefts, talipes, developmental dysplasia of the hip, congenital cataract, congenital deafness, congenital heart diseases and retinopathy of prematurity) as a component of the national child health programme (Rashtriya Bal Swasthya Karyakram, RBSK).<sup>23</sup> This would be a step further to detect the actual load of structural deformities in the community.

#### LIMITATIONS

As it is a tertiary care hospital or referral centre, 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. Secondly, 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 needed which was again a limitation.

#### CONCLUSIONS

This study has highlighted the prevalence and types of congenital anomalies seen in our locality. Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed. Many congenital anomalies can be prevented. Vaccination, adequate intake of folic acid and iodine, and adequate antenatal care are important preventive measures.

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