Research Article

CONGENITAL ANOMALIES PREVALENCE IN KUMAUN REGION OF UTTARAKHAND, INDIA: A MULTICENTRE STUDY

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ABSTRACT

Congenital anomalies occur as a result of abnormalities in the newborn baby's structure, function or body metabolism. We conducted a descriptive study to know the incidences of congenital anomalies and its prevalence among children. This is a multicentre cross sectional study including Government and Private Hospitals of Haldwani city of Uttarakhand, India. The total numbers of hospital admissions were 5600 including 424 cases of congenital anomalies. This study was conducted for a period of five months duration. This survey was performed with the help of questionnaire. 424 cases of congenital anomalies showing the average incidence of 7.57%. The prevalence pattern of congenital anomalies shows cases of defects at birth 251, which includes congenital heart disease (36%), cleft lip and palate (14%), neural tube defects (12 %), clubfoot (10%), congenital deafness (8%) and others (16%). The cases of developmental delay and disability were 149 included vision impairment (45%), hearing impairment (17%), neuromotor impairment (23%), learning disorder (4%), behavior disorder (4%), language delay (3%), cognitive delay (3%) and motor delay (1%). 24 cases of intrauterine foetal death were observed. In order to reduce the chances of infant mortality and improving the health of the children, it is necessary for us to expand our study of the environment role in etiology of birth defects and better understanding of risk factors for birth defects.

Keywords: Congenital anomalies, birth defects, intrauterine foetal death, congenital heart disease.

INTRODUCTION

The term congenital anomaly refers to any morphological, biochemical, functional or molecular defects may occur in the embryo and foetus from conception period till birth, present at the time of birth whether detected or not. About 3 million infants born and undeveloped fetuses, have major malformations¹. Any abnormalities in newborn baby's structure function or body metabolism includes birth defects which leads to mental and physical disabilities and sometimes become fatal. However, there have been no studies showing incidences as well as factors responsible for causing congenital malformations. The main etiology behind birth defects involves wide range of factors-genetic and environmental interactions, chromosomal anomalies, environmental agents. India stands second position in population and large number of infants born annually². Birth defects increases economic burden and psychological toll on the parents of these children as there are no government health services for patients affected with this condition in India and these patients need lifelong care. The most common classes of birth defects include congenital heart defects, neural tube defects (NTDs), and cleft lip and palate³. These defects generally originate during the first trimester because of gene-environment interactions or polygenic inherited diseases⁴. Deficiency of folic acid during the preconceptional period, concomitant anticonvulsant drug exposure in case of maternal epilepsy, obesity, diabetes mellitus, and hyperthermia are all major risk factors for birth defects. Increased prevalence of birth defect (like NTD) has been observed in areas with maximum use of agricultural chemicals⁵. March of Dimes (MOD), a global report on birth defects shows worldwide 7.9 million births occur annually with serious birth defects⁶. Also joint report of World Health Organization and March of Dimes shows birth defects account for 7% of all neonatal mortality and 3.3 million under five deaths². In

Balrampur district, Uttar Pradesh, a door-to-door survey of mothers living in remote clusters of villages was conducted. This study shows that the incidence of NTDs was 6.57–8.21 per 1000 live births, which is among the highest worldwide⁷. In India, neural tube defect is the most common birth defect⁸. The types of NTDs vary from anencephaly to spina bifida occulta⁹. The risk of NTDs can be reduced by folic acid consumption during pregnancy⁸. Another set of birth defects in India are orofacial clefts, its prevalence rate was found to get reduced by intake of folic acid peri-conceptionally¹⁰. Cleft lip palate and cleft palate only are broader classification of orofacial clefts¹¹. Some studies show the relation between home proximity of pregnant females to agricultural land and risk of health problems of the infant¹²⁻¹⁴. Birth Defect Statistics shows that 2 to 3 per 100 children are born with birth defects globally, 1.5 per 1000 babies are born with neural tube defects, 2.7 per 1000 babies are born with club foot, gastrointestinal tract abnormalities and defective diaphragm, 1.9 per 1000 babies are born with cleft lip and palate and congenital heart disease. However incidences of congenital anomalies have not reduced in India over the last 8 years¹⁵.

The Study Area

Uttarakhand state is geographically, and socio-culturally diverse state spread over the thirteen districts among them there are two plain, two semi-plain and nine purely hilly districts, because of location and scattered population in both hilly and plain region (higher population density in plain region). This study was carried out at Government and Private Hospitals in Haldwani city of Nainital district Uttarakhand. Haldwani city is the largest city of Kumaun, population and population growth is higher compared with the other cities of Kumaun region. The total population of Haldwani city is 3, 64,129 (census- 2011)¹⁶. Hospitals in Haldwani are equipped with modern facilities and new

instruments. In emergency people from Kumaun region are referred to Haldwani.

MATERIAL AND METHODS

Sample selection

This study was conducted for a period of five months duration. The sample design for the survey used a multi stage random sampling approach. The inclusion criteria for the survey involve, newly born infant and children up to the age of 15 years having defects at birth and developmental delay and disability, intrauterine death cases, and infant deaths. The exclusion criteria involve deficiency disorders in children's like anemia, vitamin D deficiency, and childhood diseases like otitis media, dental caries, rheumatic heart disease. The information objectives of the survey were reflected in the content of the survey questionnaire included child age, gender, residential area, defects at birth, developmental delay and disability. Permission was obtained from Chief Medical officer of Nainital district prior to survey. This study was conducted as per GCP and volunteer consent was taken.

Questionnaire content

The questionnaire was designed in order to identify the incidences of birth defects, type of birth defects examined, age of the children, type of developmental delay and disability in the government and private hospitals. It also included the cases of intrauterine foetal death observed.

Diagnosis criteria

The diagnosis of congenital anomalies was done by the physicians, based upon the method of clinical observation and physical examination. Clinical observation focuses on the motor function, breathing function, and excretory functions. Assisted examination includes X-ray, Ultrasonic diagnosis, X-ray and pathological examination of still birth, dead birth and neonatal deaths.

Data analysis

The data entry was done by making numerical codes for various responses in excel sheet which was further analyzed by using IBM SPSS statistics 20 software. The results were obtained in tabular form, so pie charts were prepared for better understanding and representation of data.

RESULTS

This study contains total 5600 hospital admissions containing 424 patients of birth defects and developmental delay and disability, showing the average incidence of 7.57%. Among the children affected with anomalies 52% were males and 48% were females (Figure 1). The male: female ratio was 1.1:1. On the other hand, Annual Health survey, 2011 report shows that among the general population in Uttarakhand, males form only 50.94% and females form 49.06%¹⁷. Maximum number of cases of birth defects, developmental delay and disability were observed between the age group between 0 to 2 years. Figure 2 represents the age wise distribution of children's having birth defects, developmental delay and disability. The cases of defects at birth were 251, which includes congenital heart disease (36%), cleft lip and palate (14%), neural tube defects (12%), clubfoot (10%), congenital deafness (8%) and others (16%). The other cases of birth defects includes Cerebral palsy (4%), hydrocephalus (3%), hypospadias (2%), cerebral atrophy (0.8%), brachial nerve palsy (0.8%), hemangioma (0.8%), tartalis (0.4%), tissue overgrowth at genital organs (0.4%), mobile node at left hand with bluish discoloration (0.4%), pigeon chest (0.4%), congenital anomaly

both hands attached (0.4%), external ear absent (0.4%), intracranial mass on left side of head (0.4%), and neurofibromatosis in left eye (0.4%). The incidence rate of birth defect was 4.48%. Table No. 1 and Figure 3 represents the types of birth defects and number of cases observed in government and private hospitals of Haldwani city. The cases of developmental delay and disability were 149 included vision impairment (45%), hearing impairment (17%), neuromotor impairment (23%), learning disorder (4%), behavior disorder (4%), language delay (3%), cognitive delay (3%) and motor delay (1%).

Table 2 and Figure 4 shows the cases of developmental delay and disability observed in hospitals of Haldwani city.

The incidence rate of developmental delay and disability was 2.66%.

The majority of government referral hospitals are in Haldwani city, town at the base of the mountains. The study of Haldwani city includes the cases of Haldwani, Nainital, Bhimtal, Kashipur, Bazpur, Jaspur, Rudrapur, Sitarganj, Ramnagar, Gadarpur, Champawat, Khatima, Kichcha, Okhalkanda, Almora, and Tanakpur. Table No. 3 and 4 represent the cases of birth defects, developmental delay and disability according to their residential location. According to the residential location the data showed that maximum number of cases were from Haldwani, Bazpur, Sitarganj, Jaspur and Rudrapur. All these areas are tarai regions of Uttarakhand, where most of the people earnings depend upon the agricultural land. Also use of agricultural pesticides is more in these areas.

DISCUSSION

The data on the birth defects cases and their causes are very limited in the developing countries like India. Registration of perinatal deaths like still birth, intrauterine deaths is inappropriate¹⁸. There are a large number of studies on the association of birth defects and potential exposures using birth registry data. Garry *et al.*, 1996 conducted an ecological, birth registry-based study comparing the prevalence of specific birth defects between areas growing different crops. Some epidemiological studies show congenital anomalies as possible outcome of community exposure to chemical associated with environmental pollution, including studies of drinking water, contaminants like pesticides exposure, residence near waste disposal site and contaminated land, air pollution, industrial pollution, food contamination and disasters involving accidental, negligent or deliberate chemical releases of great magnitude¹⁹. There are some limitations of these studies and it is difficult to ascertain the possible causes of congenital anomalies because most of the cases go undetected until after the newborn child is discharged from the hospital. Detailed studies are required in order to identify the risk factors associated with maternal home proximity to agricultural land and high incidences of birth defects. A precautionary approach should be adopted at both community and individual level. One must reduce the exposure to potential teratogens before pregnancy is recognized. Environmental pollution is almost by definition a by-product of agricultural and industrial processes in which we engage to increase our benefits to both community and individual level. Environmental pollution causes congenital anomalies by these two mechanism i.e. preconceptional mutagenic action (maternal or paternal) or post conceptional teratogenic action (maternal). The mutagenic effects during preconception include chromosomal anomalies and syndromes caused by new mutations. During the first trimester of pregnancy the process of organogenesis occurs before pregnancy is even recognized by the females²⁰. Epidemiological studies related to environmental exposures are observational not experimental. There are various factors called cofounders, associated with health outcome (like congenital anomalies in this regard) and the exposures (environmental pollution), for example people with low socioeconomic status are more highly exposed to

pollution because of their lifestyle related with greater deprivation leading to higher exposure. Chromosomal abnormality like Down syndrome associated with maternal age, as maternal age increases with high socioeconomic status causing greater risk of this defect¹⁹. Environmental concern should be incorporated into both preconceptional and prenatal care. Important opportunities are currently being missed even in regard to well-known environmental hazards. Relevant prenatal services including screening and counseling must be provided to pregnant women. Service providers and counselors need to be aware of the uncertainties regarding environmental pollution when addressing parental concern.

A significant proportion of birth defects cases are preventable by proper antenatal care. Awareness about antenatal care will increase by female literacy and health education. Identification of high-risk cases and timely referral needs to be emphasized among health care professionals at the first time of contact with pregnant women. Recently some researchers have studied the association between home proximity of pregnant women to cultivated land and risk of health problems of her child. The reason behind cases of most birth defects remain unknown, but some of the literature shows environmental factors may cause genetic mutations

resulting in interaction with genetic factors causing birth defects²¹. Such scientific findings are of significant importance for the public health policy and practice as they highlight that birth defects may be largely preventable.

CONCLUSION

From this study, we conclude that congenital heart defects, cleft lip and palate, neural tube defects are the most prevalent birth defect cases in Kumaun region of Uttarakhand, India. The etiological factors responsible in causing birth defects remain unknown. However, genetic factors and environmental pollutants might be responsible for congenital anomalies. In order to reduce the chances of infant mortality and improving the health of the children, it is necessary for us to expand our study of the environment role in etiology of birth defects and better understanding of risk factors for birth defects will improve interventions, aimed at reducing birth defect prevalence. Future research is needed in assessing the role of environmental pollutants like agricultural pesticides and the risk of congenital anomalies.

Table 1: Birth defects cases observed in government and private hospitals of Haldwani city

S.No.	Types	Cases	Percentage
1	Congenital Heart Disease	90	36
2	Cleft lip and palate	36	14
3	Neural Tube Defect	29	12
4	Talipes (club foot)	26	10
5	Congenital deafness	18	7
6	Down's syndrome	14	5
7	Others	38	16
a)	Cerebral palsy	10	4
b)	Hydrocephalus	7	3
c)	Hypospadias	5	2
d)	Cerebral atrophy	2	0.8
e)	Hydronephrosis	2	0.8
f)	Brachial nerve palsy	2	0.8
g)	Hemangioma	2	0.8
h)	Tarticalis	1	0.4
i)	Tissue overgrowth at genital organs	1	0.4
j)	Mobile node at left hand with bluish discoloration	1	0.4
k)	Pigeon chest	1	0.4
l)	Congenital anomaly both hands attached	1	0.4
m)	External ear absent	1	0.4
n)	Intracranial mass on left side of head	1	0.4
o)	Neurofibromatosis in left eye	1	0.4
	Total	251	100

Table 2: Developmental delay and disability cases observed in government and private hospitals of Haldwani city

S. No.	Developmental delay and disability	No. of cases	Percentage
1	Vision impairment	67	45
2	Hearing impairment	25	17
3	Neuromotor impairment	34	23
4	Motor delay	2	1
5	Cognitive delay	4	3
6	Language delay	5	3
7	Behaviour disorder (Autism)	6	4
8	Learning disorder	6	4
	TOTAL	149	100

Table 3: Birth defects cases observed according to the residential location of the affected children

S. N	Defects	Residential location	Percentage
1.	Congenital Heart Disease(n=90)	Sitarganj	16.7%
		Bazpur	14.4%
		Haldwani	11.1%
		Ramnagar	10.0%
		Kashipur	10.0%
		Rudrapur	7.8%
		Gadarpur	7.8%
		Khatima	6.7%
		Bhimal	4.4%
		Tanakpur	3.3%
		Okhalkanda	2.2%
		Kotabagh	2.2%
		Betalghat	2.2%
		Champawat	1.2%
2	Cleft Lip and palate (n=36)	Haldwani	19.4%
		Bazpur	13.0%
		Kashipur	11.1%
		Sitarganj	8.0%
		Champawat	8.0%
		Rudrapur	8.0%
		Gadarpur	5.5%
		Bhimal	3.0%
		Barakot	3.0%
		Jaspur	3.0%
		Dhari	3.0%
		Nainital	3.0%
		Ramgarh	3.0%
		Ramnagar	3.0%
		Kotabagh	3.0%
		Okhalkanda	3.0%
3	Neural Tube Defect (n=29)	Haldwani	20.7%
		Champawat	13.9%
		Gadarpur	13.9%
		Jaspur	10.3%
		Rudrapur	6.9%
		Ramnagar	6.9%
		Khatima	6.9%
		Ramgarh	6.9%
		Kashipur	3.4%
		Kotabagh	3.4%
		Tanakpur	3.4%
		Bhimal	3.4%
4	Talipes (clubfoot) (n=26)	Haldwani	19.2%
		Sitarganj	15.4%
		Khatima	11.6%
		Ramnagar	11.6%
		Bazpur	7.8%
		Rudrapur	7.8%
		Kichcha	3.8%
		Mukteshwar	3.8%
		Almora	3.8%
		Jaspur	3.8%
		Ramgarh	3.8%
		Champawat	3.8%
		Dhari	3.8%
5	Congenital deafness (n=18)	Bazpur	27.7%
		Gadarpur	22.2%
		Sitarganj	16.6%
		Kashipur	11.1%
		Khatima	5.6%
		Kichcha	5.6%
		Rudrapur	5.6%
		Ramgarh	5.6%
6	Down Syndrome (n=14)	Haldwani	35.8%
		Sitarganj	21.5%
		Gadarpur	7.1%
		Bazpur	7.1%
		Jaspur	7.1%
		Ramnagar	7.1%
		Rudrapur	7.1%
		Nainital	7.1%
7	Others (n=38)	Haldwani	26.3%
		Jaspur	21.0%
		Bazpur	18.4%

		Rudrapur	13.1%
		Tanakpur	7.8%
		Khatima	5.2%
		Okhalkanda	2.5%
		Ramnagar	2.5%
		Gadarpur	2.5%

Table 4: Developmental delay and disability cases observed according to the residential location of the affected children

1.	Vision impairment (n=67)	Haldwani	67.1%
		Kashipur	15.1%
		Bazpur	8.9%
		Champawat	4.5%
		Gadarpur	2.9%
		Khatima	1.5%
2.	Hearing impairment (n=25)	Haldwani	24.0%
		Kotabagh	16.0%
		Sitarganj	16.0%
		Kashipur	12.0%
		Gadarpur	8.0%
		Tanakpur	8.0%
		Bazpur	4.0%
		Champawat	4.0%
		Rudrapur	4.0%
		Ramnagar	4.0%
3.	Neuromotor impairment (n=34)	Haldwani	29.4%
		Rudrapur	17.6%
		Champawat	14.8%
		Sitarganj	11.8%
		Bazpur	8.8%
		Gadarpur	5.9%
		Kashipur	5.9%
		Kotabagh	2.9%
		Ramnagar	2.9%
4.	Behaviour disorder (n=06)	Haldwani	33.3%
		Ramnagar	33.3%
		Jaspur	33.3%
5.	Learning disorder (n=06)	Haldwani	66.6%
		Rudrapur	33.4%
6.	Language delay (n=05)	Haldwani	60.0%
		Sitarganj	20.0%
		Gadarpur	20.0%
7.	Cognitive delay (n=04)	Haldwani	50.0%
		Jaspur	25.0%
		Bazpur	25.0%
8.	Motor delay (n=03)	Bazpur	33.3%
		Sitarganj	33.3%
		Rudrapur	33.3%

Figure 1: Gender wise distribution of children's having birth defect and developmental delay and disability

Figure 2: Age wise distribution of children's having birth defect and developmental delay and disability

Figure 3: Cases of Birth Defects observed in government and private hospitals of Haldwani city

Figure 4: Cases of Developmental delay and disability observed in government and private hospitals of Haldwani city

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