

# CONGENITAL MALFORMATIONS AT BIRTH

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## ABSTRACT

*Three thousand nine hundred and thirty-two consecutive newborns were examined at birth for the presence of congenital malformations. The overall incidence of malformations was 1.2%. Congenital malformations accounted for 9.2% of perinatal and 12.8% of neonatal deaths. The central nervous system (39.5%) was most commonly involved followed by musculoskeletal system (14.5%). Involvement of more than one system was observed in 18.8% cases. Though there was higher incidence of malformations in babies born to mothers of more than 35 years the difference was not statistically significant. However, the babies born to mothers of gravidity 4 or more had significantly higher incidence of malformation when compared to mothers of lower gravidity ( $\chi^2 = 4.67$ ,  $p < 0.05$ ). The incidence of congenital malformations at birth was higher in stillborn and low birthweight babies.*

**Key words:** Congenital malformations, Perinatal mortality.

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Congenital malformations affect 2.5% of infants at birth and are responsible for about 15% of perinatal mortality in India(1,2). In United States, among the newborn population 3 to 5% are likely to have a major congenital malformation and account for more than a quarter of a million affected children each year. The effect is not limited to a few affected individuals but extends to thousands of at-risk individuals as well as their families(3). The purpose of the present communication is to present the spectrum and course of congenital malformations in the fetus or the newborns, at birth, diagnosed clinically at a referral hospital in North India.

## Material and Methods

Three thousand nine hundred and thirty-two consecutive newborns (including 56 sets of twins) delivered in the Department of Obstetrics and Gynecology, Banaras Hindu University, Varanasi during January 1988 and December 1989 were examined for the presence of congenital malformations at birth. The details of maternal age, parity, obstetric history (particularly of drug intake or radiation exposure), complications in present pregnancy and labor were noted. The sex, birthweight, crown-heel length, head circumference, Apgar score at 1 and 5 minutes and morbidity and mortality were recorded. The results were analysed by simple statistical techniques and tests of significance including Chi-square tests were applied.

## Results

Of 3932 consecutive newborns, 48 had congenital anomalies giving an overall incidence of 1.2%. The incidence of congenital anomalies were higher among the stillbirths (6.9%) than among the liveborn babies (0.9%). The analysis of perinatal deaths

during January to December 1988 showed that 9.2% of perinatal deaths and 12.8% of neonatal deaths could be attributed to lethal congenital malformations (*Table I*).

*Table II* summarizes the congenital malformations seen in the patients. The central nervous system was the most commonly affected system (39.5%) followed by musculoskeletal system (14.5%). Of 48 cases, 39 (81.2%) had involvement of single system and 9 (18.8%) had involvement of more than one system.

*Table III* summarizes the maternal and fetal factors associated with congenital anomalies at birth. Though there was slightly higher incidence of malformations in the babies born to mothers of more than 35 years, the difference was not statistically significant. However, the babies born to mothers of gravida 4 or more had a significantly higher incidence of malformations

when compared to mothers of lower gravidity ( $\chi^2 = 4.67$ ,  $p < 0.05$ ). No significant sex difference could be observed among the congenitally malformed babies (male 24, female 23, ambiguous genitalia 1). The incidence of congenital malformation was significantly higher amongst low birth weight (<2500 g) babies and stillborn babies.

### Discussion

With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developed countries and would very soon be increasingly important determinants of perinatal mortality in developing countries like India(2,4). The congenital malformation was the third most important factor in the perinatal mortality determinants next to birth asphyxia and prematurity at our

**TABLE I-Causes of Perinatal Mortality in Relation to Neonatal Fetal Factors at University Hospital, Varanasi in Comparison with Other Institutions of India**

Sl. No. Causes	AIIMS(2) (n=291)	PGI(4) (n=134)	Hyderabad(5) (n=335)	Varanasi (n=195)
1. Perinatal hypoxia and injury	26.5	43.3	12.2	48.2
2. Immaturity (including HMD, IVH and weight less than 750 g)	18.9	114.2	3.9	21.5
3. Congenital malformations	13.4	10.5	4.2	9.2
4. Bacterial infections! septicemia	6.2	17.9	16.7	7.2
5. Others	8.2	6.4	63.0	5.1
6. Unknown (includes) macerated stillbirths)	26.8	7.5		8.7

HMD = Hyaline membrane disease.

IVH = Intraventricular hemorrhage.

**TABLE II-Type of Congenital Malformations Observed (n=48)**

System	Type	Malformations	No.	%
<i>Central nervous</i>			19	39.5
	Major	Anencephaly	6	12.5
		Hydrocephalus	8	16.6
		Meningomyelocele	3	6.2
		Microcephaly	1	2.0
		Encephalocele	1	2.0
<i>Musculoskeletal</i>			7	14.5
	Major	Osteogenesis imperfecta	1	2.0
	Minor	Talipes	5	10.0
		Congenital dislocation of hip	1	2.0
<i>Gastrointestinal</i>			5	10.4
	Major	Tracheo-esophageal fistula	1	2.0
		Exomphalos	3	6.2
		Diaphragmatic hernia	1	2.0
<i>Cardiovascular</i>			4	8.3
	Major	Patent ductus arteriosus	1	2.0
		Congenital cyanotic heart disease	1	2.0
		Congenital acyanotic heart disease	1	2.0
	Minor	Single umbilical artery	1	2.0
<i>Genitourinary</i>			5	10.4
	Major	Ambiguous genitalia	1	2.0
	Minor	Hypospadias	4	8.0
<i>Craniofacial</i>			3	6.5
	Major	Cleft palate	1	2.0
	Minor	Laryngeal web	1	2.0
		Ranula	1	2.0
<i>Others</i>			2	4.2
	Major	Down's syndrome	1	2.0
		Prune belly syndrome	1	2.0
<i>Miscellaneous</i>			3	6.3

centre. Congenital malformations accounted for 12.8% of early neonatal mortality. In our country, congenital malformations account for 10-15% neonatal deaths and 8-18% of perinatal mortality(2,4,5). A national collaborative community-based study by Indian Council of Medical Research(6) reported that congenital malformations accounted for 6.6% of neonatal deaths in the rural as well as urban slum communities.

The overall incidence of congenital malformations in the present series was 1.2%. One of the earlier studies(4) from this hospital reported an incidence of 1.6%. This is consistent with other reported studies from various parts of India(1,2,5,7). The variability in the incidence of malformations in various parts of the country could be due to inaccurate detection at birth, inclusion of minor anomalies, period of observation, au-

**TABLE III-***Congenital Malformations in Relation to Maternal and Fetal Factors*

Factors	Total	Malformations		p value
		No.	%	
<i>Maternal age</i>				
<20 yr	149	1	0.7	NS
20-35 yr	3666	46	1.2	
>35 yr	61	1	1.6	
<i>Gravida</i>				
<4	3186	33	1.04	<0.05
≥4	690	14	2.03	
<i>Sex</i>				
Male	2210	24	1.08	NS
Female	1721	23	1.33	
<i>Birth weight</i>				
<2500 g	1141	22	1.92	<0.01
≥ 2500 g	2791	26		
<i>Liveborn and stillborn</i>				
Liveborn	3729	34	0.91	<0.005
Stillborn	203	14	6.89	

NS = Not significant.

topsy rates, geographical and ethnic factors.

Analysis of the overall distribution of malformations in the present study showed that central nervous system was the commonest system involved (39.5%) followed by musculoskeletal system (14.5%). Similar observations have been reported by some of the studies reported earlier(7,8). On the contrary, musculoskeletal abnormalities has been observed as the commonest anomaly by various authors(4,9). Malformations involving multiple systems were seen among 18.8% babies. Mishra and Bhavaja(5) reported multiple anomalies in 37.6% of anomalies. An Institution Based Collaborative Study on High Risk Pregnancies(10) reported an incidence of 9.3/1000 deliveries; hydrocephalus and anencephaly being the

commonest major malformations whereas cleft lip or palate and talipes equinovarus were the commonest minor malformations.

#### **Maternal Factors**

The incidence of congenital malformations were slightly higher amongst the mothers of more than 35 years in comparison to younger mothers. Many authors have shown higher incidence of malformations in the babies born to mothers aged over 35 years(11), whereas others have associated 20-35 years maternal age group with higher incidence of congenital malformation(3). The observation in the present study regarding significantly higher incidence of malformation among the mothers of gravida 4 or more is consistent with earlier reported studies(4,12).

*Fetal Factors*

The incidence of congenital malformations was significantly higher among the low birth weight babies (less than 2500 g) in comparison to normal weight babies. This association of low birth weight and malformations has been well documented<sup>(4,5)</sup>. Many studies have documented male preponderance amongst congenital malformed babies<sup>(4,13)</sup>. However, in the present series, like some of the earlier studies, we could not observe any sex predilection of malformation. A significantly higher incidence of malformation observed among the stillbirths in the present study is consistent with earlier reports. Aiyar and Agrawal<sup>(12)</sup> observed that the highest incidence of malformations was among fullterm normal weight babies.

Thus, congenital malformations are emerging as important perinatal problem contributing sizeably to the perinatal mortality and morbidity with considerable repercussion on the mothers and the families affected. The life-threatening congenital malformations must be identified by thorough clinical examination because early diagnosis and surgical correction or palliation of these infants offer the best chance for survival.

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