



Incidence and Evaluation of Congenital Malformations in Victoria Govt. Hospital Visakhapatnam, Andhra Pradesh

Authors

Dr. R. Subhashini MD., Dr. N. Uma MD.DGO. Dr. Neeraja MD

Abstract

Retrospective analysis conducted in 150 bedded Government Victoria Hospital, a Maternity hospital at Visakhapatnam, Andhra Pradesh during the period between Aug. 2011 – Aug .2012. This paper was focused on incidence of structural congenital malformations detectable at birth among 6590 deliveries, evaluation of associated risk factors and the fetal outcome. In our study we found 134 fetal malformations, Incidence is 2.03%. Most commonly affected is cranio spinal system; risk factor is H/O Consanguinity, Malnutrition and previous h/o abortions.

Key words: Congenital malformations, craniospinal system, Malnutrition.

MATERIALS AND METHODS

Total 134 cases out of 6590 deliveries were retrospectively evaluated for structural congenital malformations and associated risk factors during one year period from Aug. 2011 – Aug .2012..Fetal outcome was assessed . Variables like Maternal age, Parity, Consanguinity, Abortions, Sibling with malformation, Nutrition, Smoking ,Alcoholism, Family H/O congenital anomalies , Conceived after infertility treatment, Maternal Diabetes,

Infections, Fever, Drugs, H/O Intra uterine deaths were critically evaluated.

Aim and Objectives:

1. To determine the frequency of different structural congenital anomalies in our hospital population as per WHO classification.
2. To identify the possible risk factors responsible for these anomalies.
3. To evaluate the fetal outcome.

INTRODUCTION

Congenital anomalies (CA) can be defined as structural or functional abnormalities including metabolic disorders, present at birth. These defects are of prenatal origin result from defective embryogenesis or intrinsic abnormalities in the development process. Birth defects can be isolated abnormalities or part of a syndrome and continue to be an important cause of neonatal and infant morbidity and mortality.

In many cases, the causes of congenital anomalies are unknown; however, several factors known to be associated are genetic factors, maternal infections like rubella, cytomegalovirus, toxoplasmosis and syphilis, drugs like thalidomide, streptomycin, tetracycline, phenytoin, smoking, irradiation, maternal age, health, geographical factors and dietary factors.

Fetal anomaly scanning is the most powerful approach available for reducing the birth prevalence of infants with serious congenital abnormalities and increasing the chances of survival for those who are born, the finding of a correctable abnormality can be an indication for delivery to take place a center with facilities for pediatric Surgery, the finding of a severe uncorrectable abnormality may lead to offer termination of pregnancy.

This study was conducted to evaluate the incidence of structural congenital anomalies and to predict the variables which contribute in the

incidence of congenital anomalies so that we can reduce the related perinatal morbidity and mortality.

RESULTS

Out of total 6590 deliveries from Aug 2011-Aug 2012, 134 babies with CA identified. Incidence being 2.03% (Table: 1). Commonest CA involving Craniospinal system (21.6%). Out of this most common is NTD (Table: 2) (12.7%). Most common CA involving musculoskeletal system is CTEV (Table: 3).

67% of cases are registered at our hospital (Table: 9). 77.6 % cases are in the age group of 20-29 yrs. 3.7% are in the age group of 35 yrs. (Table: 10). In 56% are cases H/o consanguinity was present (Table: 12), and about 40.1% cases are Primigravidae (Table: 13). In 34% of cases H/o abortions present (Table: 14). In 31% of cases malnutrition observed (Table: 15). About 31.4% CA are detected before 28 wks. 42% of the cases are diagnosed between 28-37 wks most of them have no previous scans due to infrequent antenatal visits (Table 15).

Most common perinatal risk factors are Preterm labor (31%), polyhydramnios and breech (25.4%) (Table: 16).

Congenital malformations contribute 46% of perinatal mortality

Table: 1 Distribution of anomalies according to W.H.O classification.

<i>Type of Malformation</i>	<i>Number of Malformations in my study</i>	<i>Percentage (%)</i>
Cranio spinal	29	21.6
Musculoskeletal	21	15.7
Facial	21	15.7
Cardiovascular	16	11.9
Renal	11	8.2
Respiratory	9	6.7
Gastrointestinal	7	5.2
Genital	5	3.8
Multisystem Defect	10	7.4
Others	5	3.8
<i>Total</i>	134	100%

Most commonly affected system was CRANIOSPINAL SYSTEM (21.6%). Second most common systems are MUSCULOSKELETAL AND FACIAL (15.7% each).

Table:2 Distribution of anomalies affecting CRANIOSPINAL SYSTEM.

<i>Type of Malformation</i>	<i>NO. Of cases</i>	<i>Incidence per 1000 births</i>
CRANIOSPINAL	29	
NTD	17	2.5
Anencephaly	9	
Meningomyelocele	3	
Meningocele	2	
HC+ Meningomyelocele	1	
HC+ Meningocele	1	
Occult Spina bifida	1	

Hydrocephalus	11	1.5
Posterior cerebellar cyst	1	

Neural Tube Defects are most common anomaly involving craniospinal system.

Table: 3 Distribution of type of anomalies affecting MUSCULOSKELETAL SYSTEM

<i>Type of Malformation</i>	<i>Number of Malformations in my study</i>	<i>Incidence per 1000 Births</i>
MUSCULOSKELETAL	21	
CTLV	5	0.86
Limb Abnormalities	5	0.86
Polydactyl	4	0.6
Chest wall Deformities	4	0.6
Diaphragmatic Hernia	2	0.3
Osteogenesis Imperfecta	1	0.17

CTLV and Limb abnormalities are most common anomalies affecting musculoskeletal system.

Table 4 : Distribution of type of anomalies affecting Cardiovascular System

<i>Malformation</i>	<i>Frequency</i>	<i>Incidence/1000 births</i>	<i>Normal</i>
Cardiovascular	12		
VSD	5	0.8	1-2
ASD	2	0.3	
Hypo plastic Ventricle	1	0.16	
Pericardial effusion Contracted heart	1	0.16	

Table 5: Distribution of type of anomalies affecting Renal System

<i>Malformation</i>	<i>Frequency</i>	<i>Incidence/1000 births</i>	<i>Normal</i>
Renal	11		
Multicystic Dysplastic Kidney	2	0.3	1/10000
Polycystic Kidney	7	1.16	1/1000
Hydronephrosis	2	0.3	1/3000-1/6000

Most common renal anomaly is Polycystic Kidney

Table 6: Distribution of type of anomalies affecting Respiratory System

<i>Malformation</i>	<i>Frequency</i>	<i>Incidence/1000 births</i>	<i>Normal</i>
Respiratory	9		
Pleural Effusion	6	0.30	1/10000
Pleural Effusion + Hypo plastic Lung	3	0.16	1/10000

Table 7: Distribution of type of anomalies affecting GenitalSystem

<i>Malformation</i>	<i>Frequency</i>	<i>Incidence/1000 births</i>	<i>Normal</i>
Genital	5		
Ambiguous Genitalia	3	0.46	1-2
Congenital Hydrocele	2	1.16	

Table 8: Distribution of other type of anomalies

MALFORMATION	FREQUENCY	INCIDENCE/1000 Births	NORMAL
Multipledfects	10		
OTHERS	5	3.7	
Abdominal wall defects EXOMPOLOS	2	1.5	1/4000-1/5000
Non immune hydros	1	0.7	1/1500-1/3500
Ellis van creveled syndrome	1	0.7	
Congenital Syphilis	1	0.7	

<i>Malformation</i>	<i>Frequency</i>	<i>Incidence/1000 births</i>	<i>Normal</i>
FACIAL	21		
Cleft Lip	3	0.46	1/10000
Cleft Palate	2	0.3	1/10000
Cleft Lip + Cleft Palate	5	0.83	1/1000
Cystic Hygroma	2	0.3	5-15/1000
Low set ears	2	0.3	
Microophlmos	1	0.16	

Table 9: Distribution of 134 cases according to antenatal visits.

BOOKED	89	67%
UNBOOKED	45	33%

67% cases are BOOKED with minimum two antenatal visits at our hospital

Table 10: Distribution Of 134 Cases According To Age Group

<i>Risk Factors</i>	<i>Number</i>	<i>%</i>
Age less than 19 yrs.	12	9
20-24 yrs.	56	41.8
25-29 yrs.	48	35.8
30-34 yrs.	13	9.7
35 yrs. and older	5	3.7

MOST COMMONLY AFFECTED AGE GROUP BEING 20-24 YRS (56%) FOLLOWED BY 25-29 YRS (48%)

Table 11: GESTATIONAL AGE AT THE TIME OF DIAGNOSIS OF CONGENITAL ANOMALY

<i>Age</i>	<i>Number</i>	<i>%</i>
< 28 Weeks	42	31.4
28 Weeks – 37 Weeks	57	42.5
>37 Weeks	22	16.4
After Birth	13	9.7

MOST ANOMALIES WERE DIAGNOSED AT THE GESTATIONAL AGE OF 28-37 Wks.

Table 12: Distribution Of 134 Cases According To History Of Consanguinity

H/O CONSANGUINITY	<i>Number</i>	%
NIL	59	44.0
I degree	17	12.7
II degree	20	14.9
III degree	38	28.4

56% CASES ARE CONSANGUINOUS MARRIAGES.

Table 13: Distribution of cases according to Parity

<i>PARITY</i>	<i>Number</i>	%
PrimiGravida	54	40.1
2 nd Gravida	28	20.9
3 rd Gravida	12	9.0
4 th Gravida	14	10.5
5 and above	26	19.5

Table 14: History Of Abortions

H/O ABORTIONS	<i>Number</i>	%
NIL	89	66.4
ONE	31	23.1
TWO	11	8.2
THREE AND ABOVE	3	2.3

IN 33.4% OF CASES THERE IS H/O ABORTIONS.

Table 15: Pattern of distribution of different risk factors

<i>Risk Factor</i>	<i>Number</i>	<i>%</i>
Consanguinity	75	56
Abortions	45	33.4
Low Nutritional Diet	42	31.3
H/O IUDS	18	13.4
Maternal Diabetes	11	8.2
Age > 35 years	5	3.7
Infections, Fever	5	3.7
Conceived after infertility treatment	2	1.4
Drugs (Anti-epileptic drugs, Misoprostol)	2	1.4
Sibling with malformation	2	1.4
Family H/O Malformations	1	0.7
Smoking, Alcoholism	-	

Table 16: distribution of perinatal risk factors

<i>Risk Factors</i>	<i>Number</i>	<i>%</i>
Preterm Labor	42	30.3
Polyhydramnios	34	25.4
Breech	34	25.4
IUGR	14	10.0
Oligohydramnios	12	8.9

Table 17: Fetal Outcome In Pregnancies With Ca

Abortions	42	32%
Vaginal delivery preterm	56	41%
vaginal delivery term	31	23%
CAESARIAN SECTION FOR OSTETRIC INDICATIONS	5	4%

Out of 152 Perinatal Deaths Congenital Anomalies Contributing about 46% OF DEATHS. Even though CA of minor degree, prematurity along with associated maternal contributing factors are responsible for the perinatal mortality.

DISCUSSION

We found the incidence of CA in our hospital is 2.03% in our study which is equal to the general incidence in developing countries [2,3,4,5]. With improvement in the standards of living prenatal and antenatal health awareness, the overall incidence of NTDs has come down markedly in developed countries, in our study 22% of cases involved Craniospinal system (Fig 1,2,3). Anencephaly amounting to 13% cases of NTDs and most common factor contributing to perinatal mortality. Second most common CA involved Facial and neck structures but most of them are non fatal but contributing to perinatal morbidity. (Fig 5,6). [6-20]. Though most of the anomalies are compatible with life the increase in perinatal mortality was mainly due to associated preterm labor, prematurity, polyhydramnios, maternal diabetes, IUGR, Consanguinity is single most

important factor which was found to increase the risk of CA in our study [22]. Half of the cases H/O 3rd degree consanguinity was noted. Even though considered as low risk factor compared to 1st degree. Appropriate health education about consanguinity and genetic counseling for consanguineous couples should also be established before marriage. In addition to this, there is a need for more extensive screening studies to determine the birth prevalence, types and distribution of congenital anomalies. In 1/3rd of cases there is H/O one or more abortions.

Maternal age is an important parameter in the birth of a congenitally malformed fetus. In our study 3.7% of the mothers are older mothers (35 years of age or older).

Mothers who have given birth to children with NTDs should take 4 mg of folic acid per day for subsequent pregnancies. This positive effect can

only be achieved when supplement is taken prior to conception.



Fig: 1 Anencephaly.



Fig: 3 Meningocele



Fig: 2 Meningomyelocele



Fig: 4 Osteogenesis Imperfecta



Fig: 5 Cleft lip and Cleft palate



Fig: 7 Exomphalos



Fig: 6 Cystic Hygroma



Fig: 8 Ellis van creveled syndrome

CONCLUSION

In the present study most of the mothers who had anomalous fetuses had risk factors like Consanguinity and previous H/O abortions. Hence the need for focused screening in this high risk category. Pre scan counseling with karyotyping, triple screen and relevant serology has to be done.

A level II targeted scan is done at 18-20 weeks and again at 24 weeks to exclude anomalies. Though the cost of routine screening even in low risk women is more the burden of a severely morbid and disabled child on the family and society is even more.

Hence, if a single ultrasound examination is allowed per pregnancy, the mid trimester scan at 18- 20 weeks clearly represents the best time to accomplish the most. Once an anomaly is detected, various management options are to be discussed with the patients in consultation with neonatologist, pediatric surgeon and neurosurgeon when necessary.

Lethal anomalies are terminated immediately after diagnosis irrespective of gestational age. Autopsy can be done in needed cases.

Careful monitoring and surveillance of fetuses with minor anomalies or those compatible with life is done and delivery is contemplated at term or after lung maturity is accomplished depending on type of anomaly in a tertiary center with an intensive neonatal care.

Adequate prenatal care to improve the preconception & prenatal nutrition along with periconceptional folic acid. Thanks to our JANANI SURAKSHA YOJANA to encourage all the pregnant mothers to attend health care center

from the first month of pregnancy for checkup and discover any abnormalities. Specialist services (genetic services) should be offered to women with high risk factors like diabetic Mellitus, Epileptic women, previous history with congenital anomalies and elderly gravid.

REFERENCES

1. Rosano A et al. Infant mortality and congenital anomalies from 1950 to 1994: an international perspective. *Journal of epidemiology and community health* 2000;54:660-6.
2. Kalter H et al. Congenital malformations: etiologic factors and their role in prevention (first of two parts). *The New England journal of medicine* 1983; 308:424-31.
3. Biri A et al. Birth prevalence and distribution of congenital anomalies in a university hospital. *Perinatol Dergisi* 2005; 13:86-90
4. Bittar Z. Major congenital malformations presenting in the first 24 hours of life in 3865 consecutive births in south of Beirut. Incidence and pattern. *Lebanese medical journal* 1998;46:256-60
5. Wen SW et al. Patterns of infant mortality caused by major congenital anomalies. *Teratology* 2000; 61:342-6.
6. Rajangam S et al. Consanguinity and chromosomal abnormality in mental retardation and or multiple congenital anomaly. *Journal of the Anatomical Society of India* 2007;56:30-3.
7. Mir NA et al. Easily identifiable congenital malformations in children: Survey of incidence

- and pattern in 32,332 live born neonates. *Annals of Saudi medicine* 1992;12:366-71.
8. Sawardekar KP. Profile of major congenital malformations at Nizwa Hospital, Oman: 10-year review. *Journal of paediatrics and child health* 2005;41:323-30.
 9. Verma M et al. Congenital malformations--a retrospective study of 10,000 cases. *Indian journal of pediatrics* 1991;58:245-52.
 10. Shafei A et al. Congenital malformations and consanguinity. *The Australian & New Zealand journal of obstetrics & gynaecology* 1986;26:168-72.
 11. Tayebi N et al. The prevalence of congenital malformations and its correlation with consanguineous marriages. *Oman medical journal* 2010;25:37-40.
 12. List of some birth defects related studies conducted in India. Study location No. Of Malformed Babies Risk Factors Most Predominant Anomalies
 13. Congenital malformations at birth in Central India: A rural medical college hospital based data. (Maharashtra January 2005 and 31 July 2007) 179
 14. A community-based survey of visible congenital anomalies in rural Tamil Nadu (Rural Areas of Tamil Nadu (2004-2005) 166,833
 15. Birth defects surveillance study. (Genetic Research Centre, National Institute for Research in Reproductive Health, Parel, Mumbai, India) 1694
 16. Chromosomal abnormalities: genetic disease burden in India (Guru Nanak Dev University, Amritsar, India, March 1991 -March 2005) 1950
 17. Congenital Malformations at Birth -A Prospective Study From South India. (Department of Pediatrics, Jawaharlal Institute of Post-Graduate Medical Education and Research, Pondicherry (September 1989 to December 1992) 469
 18. Pattern of distribution of congenital anomalies in stillborn: a hospital based prospective study. (Gandhi Medical College, Hyderabad (July 2007 to December 2009)
 19. The incidence of major congenital malformations in Mysore (1967 through 1969)
 20. Congenital Malformations at Birth (Department of Obstetrics and Gynecology, Banaras Hindu University, Varanasi, January 1988 to December 1989) 20
 21. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in Central India: A rural medical college hospital based data. *Indian J Hum Genetics* 2010;16:159-63
 22. Bhat BV, Ravikumara M. Perinatal mortality in India-Need for introspection. *Indian J Matern Child Health* 1996;7:31-3.
 23. Agarwal SS, Singh U, Singh PS, Singh SS, Das V, Sharma A, et al. Prevalence and spectrum of congenital malformations in a prospective study at a teaching hospital. *Indian J Med Res* 1991;94:413-9
 24. Amar Taksande, Krishna Vilhekar, Pushpa Chaturvedi, et al. Congenital malformations at birth in Central India: A rural medical college hospital based data. *Indian J Hum Genet.* 2010 Sep-Dec;3: 159-163
 25. Mathur BC, Karan S, Vijaya Devi KK. Congenital malformations in the newborn. *Indian Pediatr.* 1975 Feb;12:179-83

26. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G, et al. Congenital malformation in newborn: A study of 10,874 consecutive births. *J AnatSoc India*.1989; 38:101–11.
27. SugunaBai NS, Mascarene M, et al. An etiological study of congenital malformation in the newborn.*Indian Pediatr*.1982 Dec; 19:1003-7.
28. Dutta V, Chaturvedi P, et al. Congenital malformations in rural Maharashtra. *Indian Pediatr*.2000 Sep; 37:998-1001.
29. New Delhi: Reproductive health; Annual report 2002-03. Indian Council of Medical Research; p. 91
30. K. Sridhar, *et al*. A community-based survey of visible congenital anomalies in rural Tamil Nadu. *Indian J Plast Surg*. 2009; 42: S184–S191
31. Z.M. Patel and R.A. Adhia.Birth Defects Surveillance Study. *Indian J Pediatr* 2005; 72 : 489-49
32. Vishnu Bhat and LokeshBabu. Congenital Malformations at Birth – A Prospective Study From South India. *Indian J Pediatr* 1998; 65 : 873-881
33. Sunethripadma, Ramakrishna d ,etal.Pattern of distribution of congenital anomalies in stillborn: a hospital based prospectivestudy.*ijpbs* 2011;2:604-610.
34. P. DASH SHARMA. The incidence of major congenital malformationsinmysore. *Indian J. Pedlar* 1970; 37 : 1-2
35. P. Chaturvedi and K.S. Banerjee.Spectrum of Congenital Malformations in the newborns from Rural Maharashtra. *Indian J Pediatr* 1989; 56 :501-507