ORIGINAL ARTICLE

STUDY OF CONGENITAL MALFORMATIONS IN NEWBORN

K. Koteswara Rao¹, A. Krishna Prasad², D. Manikyamba³, K. Adi Reddy⁴, Solomon Saawan P⁵, S. Anusha⁶

HOW TO CITE THIS ARTICLE:

K. Koteswara Rao, A. Krishna Prasad, D. Manikyamba, K. Adi Reddy, Solomon Saawan P, S. Anusha. "Study of Congenital Malformations in Newborn". Journal of Evolution of Medical and Dental Sciences 2015; Vol. 4, Issue 78, September 28; Page: 13582-13595, DOI: 10.14260/jemds/2015/1943

ABSTRACT: The etiology of congenital malformations has not been clearly defined. It is interesting to note that certain congenital malformations are more prevalent in some areas¹. Neural tubal defects (NTD) are common in Punjab and cleft lip (CL) is common in south India. Gastrointestinal tract defects are detected from south i.e. Mysore, Trivandrum, Hyderabad and Pondicherry. Polydactyly was prevalent in south India and Chandigarh. Incidence of Talipes was highest in Delhi and next in order were Chennai, Pondicherry and Patna. Malformations are controlled by genetic and environmental factors and a thorough analysis would indicate the factors responsible for their genesis and thereby, their means of prevention². Thus it is imperative that in every region the prevalence and peculiarities of malformations should be studied. Therefore, the present study was carried out in Government General Hospital, Kakinada, where consanguineous marriages are very much prevalent. The aim is to study the spectrum, incidence and maternal risk factors associated with congenital malformations.

KEYWORDS: Congenital, Malformations, Birth Defects, Consanguinity, Anomalies.

INTRODUCTION: STUDY METHOD: The study was undertaken in the NICU, Department of Pediatrics, Govt. General Hospital, Kakinada in collaboration with Department of Gynecology and Obstetrics, Govt. General Hospital, Kakinada. This is a prevalence study. The present study was carried out from September 2014 to August 2015 and 10, 720 consecutive births were studied in Government General Hospital, Kakinada.

RESULTS:

- Out of 146 cases, CNS defects were seen in 29 cases and accounted for 19.8%, GIT defects in 25 cases (17.1%), Genitourinary malformations were 15 cases (10.3%), CVS were 14 cases (9.6%), musculoskeletal anomalies were 12 cases (8.2%), Oral clefts were 12 cases (8.2%), syndromes were 10 cases (6.8%), skin defects were 8 cases (5.5%), multiple anomalies were 9 cases (6.2%), others were 12 cases (8.2%).
- Incidence among male babies was 14.8/1000 total births, among females was 12.3/1000 total births. Incidence of malformations was 1.17% among live born babies and 8.6% among still born babies³.
- Out of 24 cases of stillborn babies, 8 cases (36%) were having CNS malformations, 7 cases (32%) were GI anomalies and 3 cases (14%) had multiple defects.
- History of Polyhydramnios was recorded among mothers of 10.3% malformed babies,⁴ prematurity in 8.2% and oligohydramnios in 6.8%. Maternal history of PIH was obtained in 6.2%. There was history of fever in 1st trimester in 4.1%, history of repeated abortions in 1.37% and maternal history of Diabetes and hypothyroidism was observed in 2.7% and 0.68% respectively.

ORIGINAL ARTICLE

- The parents of 8.3% of malformed babies were married to their first cousins.⁵ 13.6% were married to a relative further than first cousin. In the present study, parental consanguinity had been noted in 21.9% of all malformations and 33.4% of Musculoskeletal defects, 30% of syndromes, 20% of G.I defects & G.U defects, 13.8% of C.N.S defects, 33.3% of miscellaneous defects.

CONCLUSION: No definite environmental insult could be established as a cause in any of the cases. It was presumed that the abnormalities were mainly caused by genetic factors.⁶ A direct relation between the maternal age and occurrence of Down syndrome was noted.⁷ Maternal disorders complicating pregnancy like Polyhydramnios, oligohydramnios, maternal febrile illness, PIH were found to be one of the associated factors in the development of congenital malformations in newborns.

Good and thorough antenatal care regarding identification and treatment of maternal complications can prevent malformations.⁸ Whenever possible, a thorough screening of the mother and fetus is to be done to rule out intra uterine infections. People should be counseled about consanguinous marriages and about the relation between higher maternal age and Down syndrome. A good and sound genetic counseling will prevent the recurrence of another congenitally malformed baby.⁹ Antenatal diagnosis of congenital malformations by means of ultrasonography, Amniocentesis and chorionic villous biopsy is very essential as mother could terminate her pregnancy so that a lot of morbidity can be avoided.

MATERIALS AND METHODS:

Source of Data: The present study was carried out from September 2014 to August 2015 and 10,720 consecutive births were studied in Government General Hospital, Kakinada. All of the children are born at this hospital. Out of these, 146 cases were having congenital malformations. The cases represent random mixture of all categories of people as the hospital caters to the needs of people from all strata coming mainly from two Godavari districts. The population consists of people from all walks of life, though majority of them are poor and illiterate.

Inclusion Criteria: All of the children who were born at this hospital during the study period were included in this study.

Exclusion Criteria: Children born outside and brought to this institute were excluded from the study.

Method of Collection of Data: A detailed history of mother regarding risk factors, maternal investigations like ante natal scans along with relevant laboratory investigations and detailed family history was taken and the babies were thoroughly examined for congenital malformations and data was recorded as per proforma. Relevant available investigations were done whenever necessary.

RESULTS: In the present study, the incidence of congenital malformations in newborn was 13.6/1000 total births.¹⁰

ORIGINAL ARTICLE

	No. of Births	No. of Malformations	Incidence/ 1000 Births
Total	10, 720	146	13.6
Male	5, 585	83	14.8
Female	5, 135	63	12.3

Sex Distribution: Out of 10, 720 babies born in this period 5, 585 were male and 5, 135 were female and the incidence was 14.8/1000 births among males and 12.3/1000 among females.

Incidence in Live born and still born babies: Congenital malformations were found in 122 cases of 10440 live born (1.17%) and in 24 cases of 280 still born (8.6%)

Types	No. of Births	No. of Malformations	%
Live Born	10, 440	122	1.17
Still Born	280	24	8.6

ORIGINAL ARTICLE

Pattern of malformations in both live born and stillborn babies: In the present study, highest numbers of malformations were seen (in decreasing order) in CNS, GIT and Musculoskeletal system.

System	Still Born		Live Born		Total	
	No	%	No	%	No	%
C.N.S	8	36	21	16.9	29	19.9
G.I.T	7	32	18	14.5	25	17.1
M.S	0	0	12	9.7	12	8.7
G.U	0	0	15	12.1	15	10.4
C.V.S	4	18	10	8.1	14	9.6
Oral Clefts	0	0	12	9.7	12	8.2
Eye	0	0	5	4	5	3.2
Skin	0	0	8	6.5	8	5.5
Syndromes	0	0	10	8.1	10	6.8
Multiple	3	14	6	4.8	9	6.3
Miscellaneous	0	0	7	5.6	7	4.8
Total	22	100	124	100	146	100

Distribution of different types of CNS defects:

MENINGOMYLOCELE

ORIGINAL ARTICLE

CNS defects were seen in 29 cases (19.9%) out of 146. Meningomyelocele was the commonest CNS anomaly¹¹ seen in 10 cases (34.5%) followed by hydrocephalus in 9 cases (31%).

Malformation	No	%
Meningomyelocele	10	34.5
Anencephaly	4	13.8
Hydrocephalus	9	31
Spinabifida	1	3.5
Microcephaly	2	6.9
Sacroccygeal teratoma	1	3.5
Dandy Walker Malformation	2	6.8
Total	29	100

Distribution of different types of G.I defects:

ORIGINAL ARTICLE

GI defects were seen in 25 cases (17.1%) out of 146. Most common were Anorectal malformations and congenital diaphragmatic hernia¹² in 6 cases (24%) each.

Malformation	No	%
Anorectal Malformations	6	24
Exomphalos	3	12
Gastroschisis	3	12
Diaphragmatic hernia	6	24
Duodenal atresia	2	8
Tracheo-esophageal Fistula	5	20
Total	25	100

Distribution of various types of Musculoskeletal defects: Musculoskeletal defects were seen in 12 cases (8.2%) out of 146. TEV is found in 25% & Polydactyly, Genu recurvatum and CDH were found in 16.7% each. In many of these cases maternal history of Oligohydramnios was obtained.¹³

Malformation	No	%
Talipes equinovarus	3	25
Genu recurvatum	2	16.7
Absence of Depressor Angularis oris	1	8.4
Cong dislocation of hip	2	16.7
Polydactyly	2	16.7
Phocomelia	1	8.4
short neck	1	8.4
Total	12	100

ORIGINAL ARTICLE

Distrubution of various types of Genito Urinary defects: Genito urinary defects were seen in 15 cases (10.3%). Among the G.U defects one peculiar case of absence of penis was observed. There were 5 cases of hypospadias¹⁴, 2 cases of hydro uretronephrosis, one case of epispadias, ambiguous genitalia and dystrophic kidney each.

Malformation	No	%
Hypospadias	5	33.2
Exstropy Of Bladder +Epispadias	1	6.7
Congenital Hydrocele	3	20
Hydro uretronephrosis	2	13.3
Dystrophic Kidney	1	6.7
Absence of Penis	1	6.7
Ambiguous genitalia	1	6.7
Ovarian cyst	1	6.7
Total	15	100

Distribution of Cardiovascular Anomalies: Cardiovascular defects were seen in 14 cases (9.6%), consisting of 35.6% cases of Acyanotic Heart Disease and 21.4% cases of Complex Heart Disease and Cyanotic Heart Diseases each. One each case of atrial myxoma, Hypo plastic Heart, and Intra cardiac tumor were observed. The diagnosis was by antenatal ultrasonography and X-Ray, ECG and 2D ECHO of baby.

Malformation	No	%
Acyanotic Heart Disease	5	35.6
Cyanotic Heart Disease	3	21.4
Intra Cardiac Tumor	1	7.2
Hypo plastic Heart	1	7.2
Atrial Myxoma	1	7.2
Complex Heart Disease	3	21.4
Total	14	100

ORIGINAL ARTICLE

Distribution of Oral defects: Oral defects were seen in 11 cases¹⁵ (7.5%) out of 146. Among oral clefts 18.2% were cleft lip, 36.35% were cleft palate and 45.45% had both cleft lip + palate.

Malformation	No.	%
Cleft Lip	2	18.2
CL + CP	5	45.45
Cleft Palate	4	36.35
Total	11	100

Distribution of Eye defects: Eye defects were seen in 5 cases (3.4%) out of 146. Among eye defects, Anophthalmia, congenital cataract, corneal opacity, Buphthalmous, congenital retinoblastoma accounted for 20% each.

Malformation	No.	%
Cataract	1	20
Buphthalmous	1	20
Corneal opacity	1	20
Anophthalmia	1	20
Cong Retinoblastoma	1	20
Total	5	100

ORIGINAL ARTICLE

Distribution of Skin Defects: Skin defects were seen in 8 cases (5.5%) out of 146.

Malformation	No.	%
Capillary Haemangioma	3	37.5
Pre Auricular Tag	3	37.5
Ichthyosis	2	25
Total	8	100

Distribution of Syndromes: Syndromes were seen in 10 cases (6.8%) out of 146. Among these syndromes, 3 cases of Down syndrome had been noted, of which two are male and one female. In two cases maternal age was more than 30 years.¹⁶

Malformation	No.	%
Down Syndrome	3	30
Edward Syndrome	2	20
Pierre Robin Syndrome	2	20
Short Rib Polydactyl Syndrome	1	10
Arthrogryposis Multiplex Congenita	1	10
Cornelia de Lange Syndrome	1	10
Total	10	100

ORIGINAL ARTICLE

Distribution of Multiple Anomalies: There were 9 cases (6.2%) with multisystem involvement. Two cases were Dandy Walker Cysts with other anomalies, two cases were CHD with limb defects, two cases were ascites with microcephaly and others were cloacal extropy, corpus callosal agenesis and Anophthalmia one each.

Malformation	No.	%
Dandy walker cyst + Bladder Outlet Obstruction	1	11.10
Corpus Callosal Agenesis + Bilateral HDN	1	11.10
Dandy Walker Cyst + Haemangioma	1	11.10
Cloacal Extropy + CTEV	1	11.10
Anophthalmia + Bilateral HDN	1	11.10
Fetal Ascites + Microcephalus	2	22.25
CHD + Limb Defects	2	22.25
Total	9	100

Distribution of Miscellaneous Defects: Distribution of Miscellaneous defects seen in 7 cases (4.8%).

Malformation	No.	%
Cystic hygroma	2	28.55
Retro peritoneal teratoma	1	14.3
Posterior choanal atresia	2	28.55
Dentigerous cyst	1	14.3
Prenatal teeth	1	14.3
Total	7	100

ORIGINAL ARTICLE

Association of Maternal Factors in Congenital Malformations:

Maternal History	No.	%
Fever in 1 st Trimester	6	4.1
Polyhydramnios	15	10.3
Oligohydramnios	10	6.8
H/o Repeated Abortions	2	1.37
Diminished Fetal Movements	3	2.1
Preeclampsia	9	6.2
Prematurity	12	8.2
Gestational Diabetes	4	2.7
Hypothyroidism	1	0.68

Certain adverse maternal factors were associated with malformations especially fever during 1st trimester, Polyhydramnios, oligohydramnios and eclampsia. All cases of Anencephaly had maternal history of Hydramnios in this study. Maternal history of hydramnios, oligohydramnios and preeclampsia were present in 10.3%, 6.8% and 6.2% of cases respectively. There was no history of maternal smoking and alcoholism¹⁷ in this study.

Parental Consanguinity: In the present study, parental consanguinity had been noted in 21.9% of all malformations and 33.4% of Musculoskeletal defects, 30% of syndromes, 20% of G.I defects & G.U defects, 13.8% of C.N.S defects, 33.3% of miscellaneous defects. The parents of 8.3% of malformed babies were married to their first cousins. 13.6% were married to a relative further than first cousin.

DISCUSSION: Congenital malformations are prevalent throughout the world in all socio-economic strata in different ethnic groups. In the present study, the incidence of malformations in newborn is 13.6/1000 total births. The present study was carried out in children brought to Government General Hospital, Kakinada. Those delivered at home by midwives and not brought to this institute are not included. Thus, the figures might not be exact, still they give a picture of the incidence. The present study is a prospective study and gives a reliable picture.

ORIGINAL ARTICLE

It is possible that certain adverse environmental factors due to rapid industrialization and increased population flow over the area covered in the study might have been responsible for the higher incidence of malformations¹⁸. At present the high incidence in this area could probably be attributed to the high prevalence of consanguineous marriages. The rate of malformed babies was higher among stillborn. Incidence of congenital malformation was significantly higher among male babies, preterm babies, still births, LBW babies and in consanguineous marriages. In many of the cases of musculoskeletal malformations¹⁹, maternal history of Oligohydramnios was obtained and this might have caused the deformity in these cases.

CONCLUSION: 146 malformed babies were detected among 10, 720 newborns. Of these, CNS malformations were 29 (19.9%), Gastrointestinal were 25 (17.1%), Genitourinary malformations were 15 (10.3%), CVS were 14 (9.6%), musculoskeletal anomalies were 12 (8.2%), Oral clefts were 12 (8.2%), syndromes were 10 (6.8%), skin defects were 8 (5.5%), multiple anomalies were 9 (6.2%), others were 12 (8.2%). Parental consanguinity was noted 21.9%. No definite environmental insult could be established as a cause in any of these cases. It was presumed that these abnormalities were mainly caused by genetic factors. A direct relation between the maternal age and occurrence of Down syndrome was noted. Maternal disorders complicating pregnancy like Polyhydramnios, oligohydramnios, maternal febrile illness, PIH were found to be one of the associated factors in the development of congenital malformations in newborns.

Most of the congenital malformations are preventable. Good and thorough antenatal care regarding identification and treatment of maternal complications can prevent malformations. Administration of folic acid during the periconceptional period will go a long way in preventing NTD like Anencephaly²⁰. Scrupulous avoidance of teratogenic drugs and unnecessary exposure to radiation during antenatal period should be adhered to one and all²¹. Wherever possible a thorough screening of the mother and fetus to be done to rule out intra uterine infections.

People should be counseled about consanguineous marriages and about the relation between higher maternal age and Down syndrome. A good and sound genetic counseling will prevent the recurrence of another congenitally malformed baby. Antenatal diagnosis of congenital malformations by means of ultrasonography, Amniocentesis and chorionic villous biopsy is very essential as mother could terminate her pregnancy so that a lot of morbidity can be avoided.

REFERENCES:

1. Congenital malformations in India. ICMR Bull 1984; 14; 48-54.
2. Hudgins L, Cassidy S B. Congenital anomalies. In Martin RJ, Fanroff AA, Walsh MC (eds). Neonatal –Perinatal Medicine. 8th (edn), Philadelphia, Mosby- Elsevier. 2006. p561-81.
3. Ali A, Shafikhani Z, Abdulahi M. Congenital malformations among live births at Arvand Hospital Ahwaz Iran – A Prospective study. Pak J Med Sci 2008;24; 33-37.
4. Sagunabai NS, Mascarene M, Syamalan K and Nair PM. An etiological study of congenital malformations in the newborn. Indian Pediatr. 1982; 19: p1003.
5. Puri RK, Verma IC and Bhargava. Effects of consanguinity in a community in Pondicherry. In: Medical genetics in India, Vol 2, Ed. Verma, IC, Auroma enterprises, Pondichery 1998; p 176.
6. Sundara PSS, Inbreeding in Tamilnadu and its effects of human reproduction. In: Medical Genetics in India, Volll, Ed verma enterprises, Pondichery.1998; p.129.

ORIGINAL ARTICLE

7. Stoll B J. Congenital anomalies. In Kliegman R M, Jenson H B, Behrman R E, Stanton B F(eds) Nelson Textbook of paediatrics 18th (edn) Philadelphia. WB Saunders, 2008.pp.711-13.
8. Kalter H, Warkany J. 1983. (Medical Progress) Congenital malformations etiologic factors and their role in prevention. New England Journal of Medicine 308(8):424-431(Part I);491-497(Part II).
9. World Health Organization (WHO). 1999. Human Genetics: Services for the Prevention and Management of Genetic Disorders and Birth Defects in Developing Countries: Report of a Joint WHO/WOAPBD Meeting. Geneva: WHO.
10. Swain S. Agarwal A, Bhatia BD. Congenital malformations at birth. Ind Pediatr 1994; 31(10):1187-91.
11. Cherian A, Seena S, Bullock RK, Antony AC. Incidence of neural tube defects in the least developed area of India: A population-based study. Lancet 2005; 366:930-931.
12. Mishra PC, Baweja R. Congenital malformations in the newborns. A prospective study. Ind Pediatr 1989; 26(1): 32-35.
13. Verma M. Chhatwal J. Singh D. Congenital malformations. A prospective study of 10, 000 cases. Ind J Pediatr.1991; 58(2): 245-52.
14. Gupta R K, Singh A, Gupta R. Pattern of Congenital Anomalies In Newborn at Birth: A Hospital Based
a. Prospective Study. Scientific paper presented during the Proceedings at 42nd national conference of Indian academy of Pediatrics (Pedicon) at KolKatta, 2005.
15. Smith HW, Keen M, Edwards E. 1991. Cleft lip and palate surgery in La Ceiba, Honduras. Archives of Otolaryngology—Head and Neck Surgery 117(12):1356-1359.
16. Hook EB. 1982. The epidemiology of Down syndrome. In Pueschel SM (ed.). Down syndrome: Advances in Biomedicine and the Behavioral Sciences. Cambridge, MA: Ware Press. Pp. 11-18.
17. Gladstone J, Nulman I, Koren G. 1996. Reproductive risks of binge drinking during pregnancy. Reproductive Toxicology 10(1):3-13.
18. Kuliev AM, Modell B. 1990. Problems in the control of genetic disorders. Biomedical Science 1(1):3-17.
19. Gupta RK, Gupta CR, Singh D. Incidence of Congenital malformations of the musculoskeletal system in live borns in Jammu. JK Science 2003; 4: 157-60.
20. Berry RJ, Li Z, Erickson JD, Li S, Moore CA, Wang H, Mulinare J, Zhao P, Wong LC, Gindler J, Hong S, Correa A. 1999. Prevention of neural-tube defects with folic acid in China. New England Journal of Medicine 341(20):1485-1490.
21. Schull WJ, Otake M. 1999. Cognitive function and peri natal exposure to ionizing radiation. Teratology 59(4):222-226.

ORIGINAL ARTICLE

AUTHORS:

1. K. Koteswara Rao
2. A. Krishna Prasad
3. D. Manikyamba
4. K. Adi Reddy
5. Solomon Saawan P.
6. S. Anusha

PARTICULARS OF CONTRIBUTORS:

1. In charge Professor, Department of Pediatrics, Rangaraya Medical College, Kakinada.
2. Associate Professor, Department of Pediatrics, Rangaraya Medical College, Kakinada.
3. Professor & HOD, Department of Pediatrics, Rangaraya Medical College, Kakinada.
4. Junior Resident, Department of Pediatrics, Rangaraya Medical College, Kakinada.

FINANCIAL OR OTHER

COMPETING INTERESTS: None

5. Junior Resident, Department of ENT, Rangaraya Medical College, Kakinada.
6. Junior Resident, Department of Pediatrics, Rangaraya Medical College, Kakinada.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. K. Adi Reddy,
Junior Resident,
Department of Pediatrics,
Rangaraya Medical College,
Kakinada.
E-mail: adi9949844121@gmail.com

Date of Submission: 18/09/2015.
Date of Peer Review: 19/09/2015.
Date of Acceptance: 21/09/2015.
Date of Publishing: 28/09/2015.