



BMH Medical Journal 2016;3(4):85-93 **Research Article**

Prevalence of congenital anomalies in routine antenatal ultrasound

L Sreekala MS, DGO

Baby Memorial Hospital, Kozhikode, Kerala, India. PIN: 673004

Address for Correspondence: Dr. L Sreekala MS, DGO, Consultant Fetal Medicine, Baby Memorial Hospital, Kozhikode, Kerala, India. PIN: 673004. Email: kalasre@gmail.com

Abstract

Objectives: To evaluate the antenatal prevalence of major congenital anomalies in the hospital population.

Study design: Cross sectional observational.

Setting: Department of Fetal Medicine, Baby Memorial Hospital Calicut, India.

Duration: 10 months from November 2015 to August 2016.

Sample size: 2312

Material and methods: Retrospective analysis of all antenatal patients who underwent obstetric ultrasound was done. During the study period 5390 obstetric ultrasound examinations were performed for 2312 patients. Data was analyzed from all the antenatal ultrasound examinations to determine the prevalence of congenital anomalies. Data was entered into Excel data sheet and appropriate statistical analysis was performed.

Results: 38 cases of congenital anomalies were diagnosed. The antenatal prevalence of congenital anomalies was 16.43 per 1000 and 1.6%. The median maternal age at diagnosis was 26.7 years. The median gestational age at diagnosis was 21weeks+/- 3days. Central nervous system, cardiovascular and gastrointestinal anomalies were the most prevalent.

Conclusion: The prevalence of major congenital anomalies in the study population was 1.6%. Central nervous system anomalies were the most common, followed by cardiovascular and genitourinary system anomalies.

Keywords: congenital anomalies; antenatal ultrasound

Introduction

Congenital anomalies can be defined as structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth or later in life [1]. They caused 2.761 million deaths during the neonatal period in 2013, worldwide [2,3] and 2.5% in India [4]. Birth defects are present in about 3% of newborns in USA [5,6]. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India. It is not only a leading cause of fetal loss,

but also contributes significantly to preterm birth, childhood and adult morbidity along with considerable repercussion on the mothers and their families [7,8]. 2-3% of all newborns have at least one major abnormality, 10% newborns have minor abnormalities.

Congenital anomalies are often classified into four different types: malformations, deformations, disruption and dysplasia [9]. Malformation is a morphological defect of an organ, part of an organ or larger region of body that results from abnormal developmental process. Deformation refers to an abnormal form, shape or position of part of the body caused by mechanical forces antenatally, often as a result of intrauterine molding or constraint. Disruption is a morphological defect of an organ, part of an organ or larger region of the body resulting from breakdown of previously normal tissue. Dysplasia is an abnormal organization of cells in to tissues and its morphological results. The most common conditions include congenital heart defects [10] oro-facial clefts, Down syndrome [11], and neural tube defects [12].

Causes and risk factors

Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, there are some known causes or risk factors.

Socioeconomic and demographic factors

Although low income may be an indirect determinant, congenital anomalies are more frequent among resource-constrained families and countries. It is estimated that about 94% of severe congenital anomalies occur in low- and middle-income countries, where women often lack access to sufficient, nutritious food and may have increased exposure to agents or factors such as infection and alcohol that induce or increase the incidence of abnormal prenatal development. Further, advanced maternal age increases the risk of chromosomal abnormalities, including Down syndrome, while young maternal age increases the risk of some congenital anomalies.

Genetic factors

Consanguinity increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk for neonatal and childhood death, intellectual disability and other anomalies in first-cousin unions. Some ethnic communities (e.g. Ashkenazi Jews or Finns) have a comparatively high prevalence of rare genetic mutations, leading to a higher risk of congenital anomalies.

Infections

Maternal infections such as syphilis and rubella are a significant cause of congenital anomalies in low- and middle-income families.

Maternal nutritional status

Iodine deficiency, folate insufficiency, obesity and diabetes mellitus are linked to some congenital anomalies. For example, folate insufficiency increases the risk of having a baby with a neural tube defect. Also, excessive vitamin A intake may affect the normal development of an embryo or fetus.

Environmental factors

Maternal exposure to certain pesticides and other chemicals, as well as certain medications, alcohol, tobacco, psychoactive drugs and radiation during pregnancy, may increase the risk of having a fetus or neonate affected by congenital anomalies. Working or living near, or in, waste sites, smelters or mines may also be a risk factor, especially if the mother is exposed to other environmental risk factors or nutritional deficiencies.

Materials And Methods

This is a cross sectional observational study from the department of Fetal Medicine at Baby Memorial Hospital Calicut, India. It is a tertiary care hospital in private sector. We perform three antenatal scans routinely, at 11-14 weeks, 20-24 weeks and 32-36 weeks. Additional scans are done when indicated.

Ultrasound scan is an essential part of antenatal care. It is a safe, non-invasive method for evaluating the fetus. The purpose of this study was to evaluate the prevalence of congenital abnormalities in the general obstetric population of Baby Memorial Hospital, Calicut, India. Being an audit, IRB approval was waived as per our protocol.

During the study period from November 2015 to August 2016, 5390 obstetric scans were performed for 2312 patients. Scans were performed on Voluson 730pro machine either trans abdominally or transvaginally after obtaining written consent. Obstetric and medical history were noted, especially history of drug intake, viral infection and chronic disorders like Diabetes mellitus or hypertension. Type of congenital malformation, age of the mother and gestational age at the time of diagnosis were noted.

The following structures should be visualised at a routine second trimester morphology ultrasound:

Fetal head

Fetal skull: integrity and shape

Fetal brain:

- Ventricles and choroid plexus, Cavum septum pellucidum
- Posterior fossa, including measurements of trans cerebellar diameter and cisterna magna, Nuchal Fold Thickness

Fetal face

- Profile, Nasal bone, Orbits and lenses, Upper lip and palate, Mandible

Fetal heart and chest

- Fetal heart rate and rhythm, Cardiac situs, Four Chamber View, outflow tract views, aortic and ductal arches, diaphragm and lungs

Fetal abdomen

- Liver, stomach (including situs), kidneys and renal arteries, abdominal wall, umbilical cord insertion, bladder, umbilical arteries, presacral space

Fetal musculoskeletal system

- Spine - transverse, longitudinal +/- coronal views and skin line
- Upper limb - humera, including humeral length (HL), radius/ulna: both sides, Fingers and thumbs, including hand opening

- Lower limb - both femora, including femoral length (FL) as part of biometric assessment, both tibia/fibula: sagittal views to demonstrate orientation of the ankles to screen for talipes, both feet

Ancillary findings

- Fetal lie, cervical length, placenta, liquor volume, umbilical cord including the number of cord vessels and evaluation of knots

Results

During the study period, a total of 2312 antenatal cases were evaluated. Congenital abnormalities were identified in 38 cases. They form the study population.

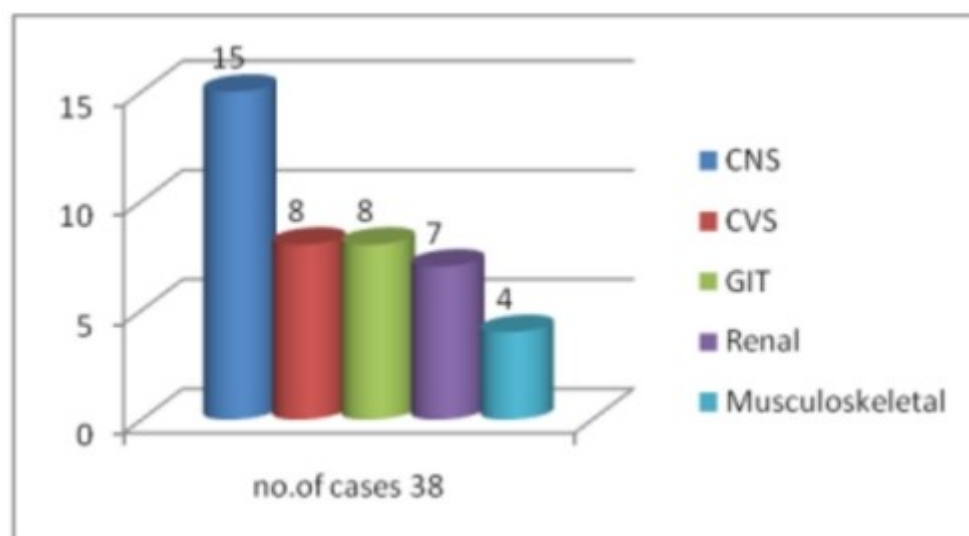
The antenatal prevalence of congenital anomalies was 1.6%. The median maternal age at diagnosis was 26.7 years.

Table 1: Maternal age wise evaluation

Maternal age (years)	No of cases
<20	1 (2.6%)
20-25	16 (42%)
26-30	15 (39.4%)
31-35	5 (13.1%)
36-40	1 (2.6%)
>40	0

Maternal age wise distribution showed that 81% of cases belonged to 20-30 years of age, reflecting the fact that maximum number of pregnancies occur in this age group (Table 1). Table 2 gives system wise distribution of anomalies.

Table 2: System wise distribution of anomalies



*Five foetuses had anomalies involving multiple systems.

The spectrum of anomalies noted is shown in **Table 3**.

Table 3: Spectrum of anomalies

Central nervous system 15(39%)	Hydrocephalus	8
	Inferior vermian agenesis	2
	Meningocele	2
	Arnold-Chiari	1
	Microcephaly	1
	Encephalocele	1
	Agenesis of corpus callosum	1
Cardiovascular system 8(21%)	Tetrology of Fallot	1
	Atrioventricular septal defect	5
	Situs inversus	2
	Ventricular septal defect	2
Genitourinary system 7(18%)	Pelvi-ureteric junction obstruction	3
	Multi cystic dysplastic kidneys	2
	Poly cystic kidneys	1
	Posterior urethral valve	1
	Unilateral renal agenesis	1
Gastrointestinal system 8(21%)	Diaphragmatic hernia	6
	Omphalocele	1
	Cleft lip and palate	1
	Bowel obstruction	2
Musculo- skeletal system 4(10%)	Mesomelia	2
	Talipes equinovarus	1
	Cystic hygroma	1

Some of the anomalies noted are illustrated in **Figures 1-4**.



Figure 1: Sonographic imaging demonstrates atrio-ventricular septal defect in the fetus of a 39 year old female at 24 weeks and 3 days gestation.

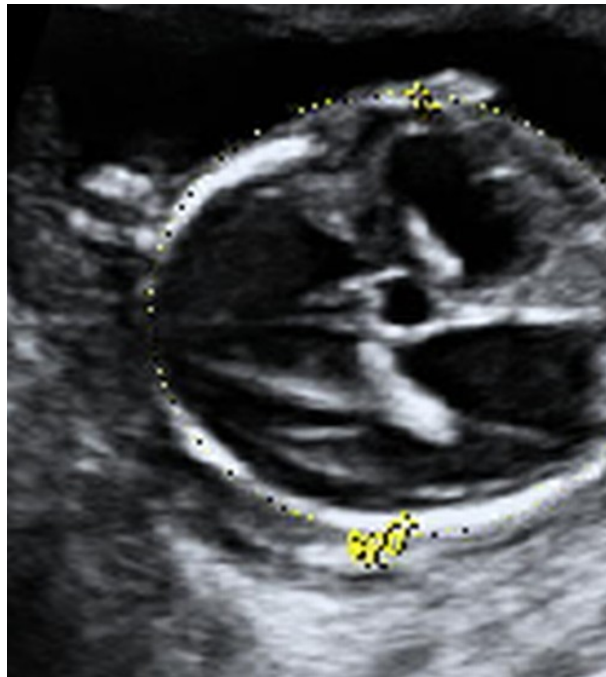


Figure 2: Sonographic imaging demonstrates dilated lateral ventricles and third ventricle – hydrocephalus in the fetus of a 26 year old female at 18 weeks and 5 days gestation.



Figure 3: Image demonstrates protrusion over occipital region of fetal head- encephalocele in the fetus of a 32 year old female at 14 weeks gestation.



Figure 4: Sonographic imaging demonstrates lumbosacral meningocele in the fetus of a 28 year old female at 22 weeks and 6 days gestation.

Discussion

The pattern and prevalence of congenital anomalies may vary over time or with geographical location [13]. With improved control of infections and nutritional deficiency diseases, congenital

malformations have become important causes of perinatal mortality in developing countries [14].

In the present study, the prevalence of congenital malformations in the ante natal period were 1.6 %, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9% [15,16].

With regard to pattern of congenital anomalies in the study, the most common system involved was central nervous system (39%), followed by gastro-intestinal tract (GIT) (21%), cardiovascular system (21%) and renal system (18%). This was comparable with studies conducted by others [17,18]. Some studies however recorded higher incidence of musculo skeletal and GIT defects [19-24], whereas Suguna Bai et al [25] reported GI malformations as the most common one.

The role of ultrasound in the detection of fetal anomalies is dependent on the prevalence of anomalies in a study population, the expertise of the examiner, the gestational age at scanning, the definition of anomaly-major and minor, and the postnatal ascertainment of anomalies. The skill and experience of the sonographers is a critical factor in the detection of fetal anomalies.

Even with a prevalence rate of 1.6%-2.7% of congenital malformations, there are no well-accepted preventive measures in most developing countries. It indicates that strong preventive measures for congenital anomalies in this region are needed. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

Conclusion

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.

References

1. Ruth A. Hannon (2010). *Porth pathophysiology: concepts of altered health states* (1st Canadian ed.). Philadelphia, PA: Wolters Kluwer Health/Lippincott Williams and Wilkins. p. 128. ISBN 9781605477817.
2. Kalter H, Warkany J. Congenital malformations (second of two parts) *N Engl J Med*. 1983;308:491-497.
3. Kalter H, Warkany J. Congenital malformations (first of two parts) *N Engl J Med*. 1983;308:424-431.
4. Patel ZM, Adhia RA, Birth defects surveillance study, year 2005. *Indian J Pediatr* 2005 Jun;72(6) :489-491.
5. Birth Defects. Available at: <http://www.cdc.gov/ncbddd/birthdefects/data.html>. Accessed on 28th September 2016.
6. Canfield MA, Honein MA, Yuskiv N, Xing J, Mai CT, Collins JS, Devine O, Petrini J, Ramadhani TA, Hobbs CA, Kirby RS, Neonatal estimates and race/ethnic specific variations of selected birth defects in the United States 1999-2001. *Birth defects Res A Clin Mol Teratol* 2006 Nov; 76(11) :747-756.

7. Bhat BV, Ravikumara M. Perinatal mortality in India-Need for introspection. *Indian J Matern Child Health.* 1996;7:31-3.
8. 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.
9. Moore KL, Persaud TVN: *The Developing Human: Clinically Oriented Embryology.* Philadelphia, WB Saunders, 2002.
10. Lin AE, Herring AH, Amstutz KS, Westgate MN, Lacro RV, Al-Jufan M, Ryan L, Holmes LB. Cardiovascular malformations :Changes in prevalence and birth status,1972-1990. *Am J Med Genet* 1999 May 21; 84(2) :102-110.
11. Centers for Disease Control and Prevention(CDC) Improved national prevalence estimates for 18 selected major birth defects -United States, 1999-2001. *MMWR Morb Mortal Wkly Rep* 2006 Jan 6 ; 54(51) :1301-1305.
12. WenSW, Liu S, Joseph KS, Rouleau J, Allen A, Patterns of infant mortality caused by major congenital anomalies. *Teratology* 2000 May; 6(5): 342-346.
13. Birch MR, Grayson N, Sullivan EA. AIHW Cat. No. PER 23. Birth Anomalies Series No. 1. Sydney: AIHW National Perinatal Statistics Unit; 2004. Recommendations for development of a new Australian birth anomalies system: A review of the congenital malformations and birth defects data collection.
14. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G. Congenital malformations in newborns: A study of 10,874 consecutive births. *J Anat Soc India.* 1989;38:101-11.
15. Chaturvedi P, Banerjee KS. Spectrum of congenital malformations in the newborns from rural Maharashtra. *Indian J Pediatr.* 1989;56:501-7.
16. 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 Genet.* 2010;16:159-63.
17. Khatemi F, Mamoori GA. Survey of congenital major malformations in 10/000 newborns. *Iran J Pediatr.*2005;15:315-20.
18. Dutta V, Chaturvedi P. Congenital malformations in rural Maharashtra. *Indian Pediatr.* 2000;37:998-1001.
19. Gupta RK, Singh A, Gupta R. Pattern of congenital anomalies in newborn at birth: A hospital based prospective study. *Proceedings of the 42nd National Conference of Indian Academy of Pediatrics (Pedicon);* Jan 6-9; Kolkata, India. 2005.
20. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. *Indian Pediatr.* 1994;31:1187-91.
21. Tibrewala NS, Pai PM. Congenital malformations in the newborn period. *Indian Pediatr.* 1974;11:403-7.
22. Mishra PC, Baweja R. Congenital malformations in the newborns. A prospective study. *Indian Pediatr.*1989;26:32-5.

23. Verma M, Chhatwal J, Singh D. Congenital malformations - A retrospective study of 10,000 cases. *Indian J Pediatr.* 1991;58:245-52.
24. Mathur BC, Karan S, Vijaya Devi KK. Congenital malformations in the newborn. *Indian Pediatr.* 1975;12:179.
25. Suguna Bai NS, Mascarene M, Syamalan K, Nair PM. An etiological study of congenital malformation in the newborn. *Indian Pediatr.* 1982;19:1003-7.