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Original Article

Congenital Anomaly Frequency, Risk Factor and Trends among Antenatal Patients Presenting at Tertiary Care Hospital in Pakistan

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Abstract

Background: Congenital anomaly is known for its multifactorial origin and trends among people across the world. Karachi is a hub of people belongs to different ethnic groups from all over Pakistan.
Objective: The study was conducted to determine the frequency of congenital anomalies among antenatal patients.
Study type, settings & duration: This descriptive cross sectional study was conducted in the outpatient Gynae department of Civil Hospital, Karachi from January 2017 to December 2018.
Methodology: The antenatal patients both booked and un-booked, diagnosed antenatal with congenitally abnormal fetus or delivered congenitally abnormal neonate were included in the study.
Results: During this tenure, a total of 8908 pregnant ladies were admitted in labor room and among those 198 were diagnosed with congenitally malformed babies. The most commonly identified abnormality was of central nervous system 60.1%, followed by renal 11.61%, gastrointestinal 4.54% and others 17.6%. Various risk factors like parental age, history of previous congenital anomaly and consanguinity showed association with these anomalies.
Conclusion: Early detection of these anomalies and identification of the associated risk factor will help in reducing morbidity and mortality and creating awareness to solve the issue.
Key words: Congenital anomalies, neural tube defects, risk factors.

Introduction

Congenitally anomalous babies are not only a concern for health care professionals but also source of emotional, psychological trauma for parents. Congenital anomalies can be defined as structural or functional anomalies including metabolic disorders that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later, as child grows.[1] These anomalies are further divided into minor and major anomalies. A minor abnormality although more common than major anomaly present at birth, having minimal effect on clinical function, but may have cosmetic and social effect, while major congenital anomalies are conditions that have significant medical, social and cosmetic consequences for the effected individual and require medical intervention.[2] Good antenatal care and recent advances in technology have improved the detection rates of anomalies, while fetus is in utero. Lack of these availabilities in developing countries, result in increased prevalence of these conditions in developing world.

According to the WHO Fact Sheet 2016, an estimated 303,000 neonatal deaths occur every year worldwide due to congenital anomalies.[3] The prevalence and trends of congenital anomalies varies across different regions of the world. European Surveillance of Congenital Anomalies (EUROCAT) is a network of population based registries to conduct epidemiological surveillance of congenital anomalies in Europe. Twenty five population based EUROCAT registries over a span of almost 30 years (1980-2012) showed 250,000 congenital anomalies among 11.5 million births

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LA conceptualized the project and did the editing of the manuscript. TA did the literature search, statistical analysis and drafting, revision & writing of manuscript. Data collection was done by SB & TS.

across Europe over 30 year.[4] The study also shows increasing trends in congenital heart disease and the anomalies associated with GIT and decreasing trends in limb defects. Another study conducted in five different regions of UK shows the prevalence of 129 per 10,000 births with increasing trend of CHD.[5] In Pakistan, mostly the studies conducted were hospital based and these studies show the prevalence as low as 1.4% to as high as 7% with preponderance of neural tube defects.[6-13]

The causes associated with congenital anomalies are multifactorial in origin in 20 to 25% of cases and remain unexplained in approximately 40 to 60% of cases.[14,15] Genetic disorders are found to be responsible in 15 to 25% of cases and environmental factors in the remaining 8 to 12%.[16] Role of Consanguinity in causing congenital anomaly has been evident by a number of studies.[11,12,17] Parental consanguinity especially the first cousin marriages are common in various part of the Middle East, Africa and Indian subcontinent has emerged as risk factor for various congenital anomalies as well as Mendelian condition such as inborn error of metabolism.[18-24] Other risk factors include folic acid deficiency, advanced maternal age, alcohol, smoking, diabetes and thyroid disorder.[25] Despite having a number of studies conducted previously there is still a need to determine the increasing prevalence and changing trends of the risk factors associated with congenital anomalies in Pakistani population.

Methodology

This cross sectional study was performed on the antenatal patients (outdoor and indoor) in Gynae unit II at Civil hospital, Karachi over a period of two years. Those patients who had congenital anomaly diagnosed antenatally or delivered with a congenitally anomalous baby were included in the study. Total 198 antenatal patients were found to have congenital anomalies. Detailed history including maternal age, trends towards cousin marriage, gestational age and antenatal detection of congenital anomaly was analyzed along with examination details of the delivered babies done at the time of delivery.

The ethical approval was obtained from Internal Review Board (IRB) of Dow University of Health Sciences, Karachi.

Results

Over a period of 2 years, total 8908 pregnant ladies admitted in the labor room were included, among these women 198 were diagnosed with congenital anomalies 2.22% in the given population. Abnormalities related to central nervous system were the most common in 119 patients (60.1%). Within those hydrocephalus was most common 44 (22.2%), anencephaly was 39 (19.69%) in number. The second most commonly identified birth defects were renal anomalies followed by GI anomalies. Most of the women who were identified with congenital anomalies aged between 26 to 30 years and majority of men were found between 26 to 35 years. Only 19 (9.5%) women had history of congenital anomaly in previous pregnancies and most of them were diagnosed between 14 to 26 weeks of gestational age and 134 (67.6%) participants showed consanguinity, among them 100 participants were found to be first cousins. Out of 198 participants, 15 (7.57%) were found diabetic and 17 (8.58%) were diagnosed as hypothyroid (Table-1).

Table 1: Distribution of maternal factors in relation to congenital anomalies. n=198

Maternal Age	*No of patients*	*%*
>18 years	21	10.60
21 to 25 years	55	27.77
26 to 30 years	81	40.90
31 to 35 years	34	17.17
36 to 40 years	3	1.51
>40 years	4	2.02
Paternal Age		
>18 years	3	1.51
21 to 25 years	22	11.11
26 to 30 years	63	31.81
31 to 35 years	51	25.75
36 to 40 years	47	23.73
>40 years	12	6.06
Parity		
Primigravida	42	21.21
Multiparous	119	60.10
Grandmultiparous	37	18.68
History of anomalies in previous pregnancies		
Yes	19	9.59
No	179	90.40
Consanguinity		
Yes	134	67.67
No	64	32.32
First cousin	100	50.50
Second cousin	34	17.17
Gestational Age		
≤13 weeks	4	2.02
14 to 26 weeks	111	56,06
27 to 39 weeks	80	40.40
≥40 weeks	3	1.51
GDM	15	7.57
Hypothyroidism	17	8.58

It is our hospital protocol to fully inform the patient about the nature, prognosis and outcome of the pregnancy before taking consent for any intervention as such we did in all these patients with congenital anomaly. As per protocol, informed consent was taken after giving full information to

those patients who opted for termination of pregnancy with congenital anomaly (Table-2).

Table 2: Distribution of congenital anomalies in relation to system involved. (n=198)

	Type	*No of Patients*	*%*
CNS	Anencephaly	36	18.18
	Hydrocephalus	59	29.79
	Hydrocephalus & spina bifida	3	1.51
	Spinabifida	5	2.52
	Meningiocele	16	8.08
Renal	Polycystic kidneys	22	11.11
	Hypospadias	1	0.5
GIT	Omphalocele	9	4.54
Skeletal	Dysplasia	1	0.5
Cystic Hygroma		5	2.52
Hydrops		6	3.03
Multiple anomalies		35	17.67
Fetal outcome n=198			
Sex of babies		*No of Babies*	*%*
Male		117	59.09
Female		81	40.90
Alive babies		97	45.45
IUD		101	54.54

Table 3: Maternal outcome among patient. (n=198)

	No of Patients	*%*
Method Of Termination		
Mechanical intracervical Foleys alone	45	22.72
Medication (PGE1 or PGE2)	99	50.00
Mechanical intracervical Foleys +Medication	16	8.00
None	38	19.19
PGE1	96	48.48
PGE2	19	9.59
No of Cycle		
First	90	45.45
Second	25	12.62
Complications		
bleeding	1	0.50
Infection	2	1.01
Transfusion	1	0.50

For termination of pregnancies we used intra cervical foley's catheter or prostaglandin E1 or Prostaglandin E2 alone and intra cervical foley's catheter followed by prostaglandin E1 or prostaglandin E2. Total 45 (22.72%) patients were induced with intra cervical Foleys', 99 (50%) with prostaglandin alone and 16 (8%) induced initially with intra cervical foleys followed by prostaglandin. Remaining 38 (19.19%) didn't need induction they delivered or aborted spontaneously. Majority of the participants 90 (45.45%) were terminated with single cycle of prostaglandin. Few of the participants developed complications like bleeding and infection. Mostly male babies 117 (59.09%) were delivered (Table-3).

Discussion

In this study, the prevalence of congenital anomaly was found to be 2.22% in our Pakistani population which is in comparable with other local studies.[17,18] Butt et al conducted a study in a similar pattern including still birth and intrauterine deaths along with live births which shows similar results (2.15%).[11] Somehow similar results found in the study conducted in Europe (EUROCAT) 2.4%.[26] Mohammed A et al from Egypt and Al media et AL from Brazilian study showed prevalence of 2.5 % & 2.4% respectively but these studies are on live births only.[21, 27] Recent Nigerian study showed the incidence of 6.2% in a study conducting in a similar pattern as our study but with differences in environmental or geographical factors responsible for congenital anomalies.[28] Our results were also found comparable with neighboring countries like India by Sarkar S et al with incidence of 2.2%. Previous Indian studies also showed similar results of 1.9% to 2.72% and studies in Iran also showed similar results of 2.8%.[22, 29]

As far as the maternal age is concerned, majority of women with congenital anomaly in our study were belonged to 26-30 years age group. The results were quite similar to the studies conducted by Singh and khan et al respectively which contradict the belief that congenital anomaly found in women with extreme age groups.[24,30] Role of consanguinity for congenital anomaly has been evident by number of previous studies.[12,13,31] This study also supports the association of consanguinity with congenital anomaly especially the first cousin marriage 50.5%.

Most of the congenital anomalies identified during our study were found between 14-26 weeks of gestational age, a time when a detailed anomaly scan is routinely carried out for structural anomalies where 60 to 90 % anomalies could be detected by an experienced sonologist as its evident by several studies.[28,32]

Only fifteen patients (7.57%) were diagnosed with GDM which is similar (8.1%) with one of the local study.[13] On the other hand, local studies showed higher incidence of diabetes (25%) in mothers delivering congenitally abnormal babies.[6] The frequencies of congenital anomalies were observed more in multiparous patients, the results are comparable with previous studies.[13, 22.]

Congenital anomalies were more frequent in male fetuses, 59% similar to the result of Nigerian and Brazilian study and a previous study

by shabbir et al in Pakistan.[12,31,33] In contrast, some studies showed female preponderance.[11, 34]

The most commonly identified congenital anomaly in our study was neural tube defects followed by renal anomalies and GI anomalies as a single anomaly. The results were comparable to other studies conducted in Brazil, Pakistan and the other neighboring countries of the region which showed highest prevalence of neural tube defect among other congenital anomalies.[9,12,13,31] The most common CNS abnormality found among these patients were hydrocephalus followed by anencephaly and meningocele respectively. Previous local studies support these trends in neural tube defects.[9,10,13]

Multiple other anomalies were identified in 17.6% of cases in our study and the results are quite similar to the study conducted in Nigeria by Singh S et al (18%) but contradict to Egyptian study which showed the incidence of multiple anomalies to be 28%.[31,21]

Our results are in accordance with the previous studies showing the preponderance of neural tube defect among pregnant woman having anomalous babies. Consanguinity and male sex of babies were found as the most common identifiable risk factors. Though, the study was conducted at tertiary care center which provided us much information about the pattern and risk associated with congenital anomalies As one knows tertiary care is a referral center so there is still a need of community based study with large number of women to enlighten the risk and pattern associated with this problem and need to address with better preconception and antenatal care.

Conflict of interest: None declared.

References

1. Connolly CO, Donoghue KO, Mahony M. A review of congenital anomalies in the Cork and Kery region from 1996-20100. Arch Dis child Fetal Neonatal ed.2014: 99.
2. Czeizel AE. Birth defects are preventable. Int J Med Sci 2005; 2(3): 91-2.
3. World Health O. Congenital Anomalies. World Health Organization Fact Sheet, 2014.
4. Loane M, Dolk H, Garne E. Paper 3: EUROCAT data quality indicators for population based registries of congenital anomalies. Birth Defects Res A Clin Mol Teratol 2011; 91(suppl-1): S23-30.
5. Rankin J, Pattenden S, Abramsky L, Boyd P, Jordan H, Stone D, et al. Prevalence of congenital anomalies in five British regions (1991-99). Arch Dis Child Fetal Neonatal Ed 2005; 374-9.
6. Parveen F, Tayyab S. Frequency and pattern of distribution of congenital anomalies in the newborn and associated maternal risk factors. J Coll Physician Surg Pak2007; 17(6): 340-3.
7. Waqas J, Farooq A, Taimoor J, Sajjad MM. Prevalence of Gross congenital malformations at birth in the neonates in a tertiary care hospital. APMC 2009; 3(1): 47-50.
8. Gulrukh Q. Relationship of selected prenatal factors to pregnancy out and congenital anomalies. J Ayub Med Coll Abbottabad 2010; 22(4): 41-5.
9. Saima G, Kazmi NH Najeeb S, Hussain S, Raza A. Frequencies of congenital anomalies among new borns admitted in nursery of Ayub teaching hospital Abbottabad Pakistan. J Ayub Med Coll Abbottabad 2011; 23(1): 117-21.
10. Khan AA, Khattak TA, Shah SH, Roshan E, Haq AU. Pattern of congenital anomalies in newborn. JRMC2012; 16(2): 171-3.
11. Butt F, Shahzad R, Pasha I. Pattern and outcomes of congenital anomalies and maternal risk factor association. Biomedica 2013; 29: 234-40.
12. Hussain S, Imran A, Sabir MU, Chatta MN, Tarar SH, Mushtaq R. Prevalence and pattern of congenital malformations among neonates in the neonatal unit of teaching hospital. J Pak Med Assoc2014; 64(6): 629-34.
13. Karim R, Wahab S, Akhtar R, Jamaal F, Jabeen S. Frequency and pattern of distribution of antenatally diagnosed congenital anomalies and the associated risk factor. J Postgrad Med Inst 2014; 28(2): 184-8.
14. Kumar MR, Bhat BV, Oumachigui A. Perinatal mortality trends in a referral hospital. Indian J Pediatr 1996; 63: 357-61.
15. Akinmoladun JA, Ogbole GI, O Oluwasola TA. Pattern and outcome of prenatally diagnosed major congenital anomalies at a Nigerian tertiary hospital. Niger J Clin Pract 2018: 21; 560-5.
16. Rajangam S, Devi R. Consangunity and chromosomal abnormality in mental retardation and or multiple congenital anomalies. J Anat Soc India 2007; 56: 30-3.
17. Jehangir W, Ali F, Jahangir T, Massod MS. Prevalence of gross congenital malformations at birth in the neonates in a tertiary care hospital.APMC 2009; 1: 47-50.
18. Gillani S. Kazmi NH, Najeeb S, Hussain S, Raza S. Frequencies of congenital anomalies among newborns admitted in nursery of Ayub teaching hospital Abbottabad. Pakistan J Ayub Med Coll Abbottabad 2011; 23: 117-21.
19. Salloum Al A, El Mouzan MI, Herbish Al A, Omer Al A, Qurashi M. Prevalence of selected congenital anomalies in Saudi children: a community –based study. Ann Saudi Med 2015; 35(2):107-10.
20. Sallout B, Obedat N, Shakeel F. Prevalence of major congenital anomalies at King Fahad Medical City in Saudia Arabia: a tertiary care center based study. Ann Saudi Med 2015; 35(5): 343-51.
21. El Koumi MA, Al Banna EA, Lebda I. Pattern of congenital anomalies in newborn: a hospital based study. Pediatr Rep 2013; 5: e5.
22. Sarkar S, Patra C, Dasgupta MK, Nayek K, Karmakar PR. Prevalence of congenital anomalies in neonates

and associated risk factors in a tertiary care hospital in Eastern India. J Clin Neonatol 2013; 2: 131-4.
23. Borhany M, Shamsi T, Naz A, Khan A, Parveen K, Ansari S, et al. Congenital bleeding disorders in Karachi, Pakistan. Clin Appl Thrombosis/ Haemostasis 2011; 1796): 131-7.
24. Moammar H, Cheriyan G, Mathew R, Al-Sannaa N. Incidence and patterns of inborn errors of metabolism in the eastern Province of Saudi Arabia, 1983-2008. Ann Saudi Med 2010; 30: 271-7.
25. World Health O. Birth defects. Executive Board, 126th session, provisional agenda, December 2009; EB 126/10, 3. (Accessed on 10h June 2021) Available from URL:http://apps.who.int/gb/ebwha /pdf_files/EB1 26/B126_10-en.pdf.
26. Boyd PA, Haeusler M, BaristicI, Loane M, Garne E, Dolk H. Paper 1: the EUROCAT network organization and processes. Birth Defects Res A Clin Mol Teratol 2011; 91 (suppl-1): S2-15.
27. Almeida LFG, Junior EA, Crott GC, Okido MM, Berezowski AT, Duarte G, et al. Epidemiological risk factors and perinatal outcomes of congenital anomalies. Rev Ginecol Obstet 2016; 38: 348-355.
28. Akinmoladun JA, Ogbole GI, O Oluwasola TA. Pattern and outcome of prenatally diagnosed major congenital anomalies at a Nigerian tertiary hospital. Niger J ClinPract 2018; 21: 560-5.
29. Samadirad B, Khamnian Z, Hosseini MB, Dastgiri S. Congenital anomalies and termination of pregnancy in Iran. J Pregnancy 2012: 574513..
30. Singh A, Gupta RK. Pattern of congenital anomalies in new born: a hospital based prospective study. JK Science 2009; 2: 34-36.
31. Singh S, Chukwunyere DN, Omembelede J, Onankpa B. Getal congenital anomalies: An experience from a tertiary health institution in North west Nigeria (2011-2013).Niger Postgrad Med J 2015; 22: 174-8.
32. Stumpflen I, Stumpflen A, Wimmer M, Bernaschek G. Effect of detailed fetal echocardiography as part of routine prenatal ultrasonographic screening on detection of congenital heart disease. Lancet 1996; 348: 854-7.
33. Willian CH, Silva LL, Garcia LB. Prevalence of congenital anomalies and their associated factors in newborns in the city of Sao Paulo from 2010-2014. Rev Paul Pediatr 2017; 35(1): 33-8.
34. Kondo A, Kamihira O, Ozawa H. Neural tube defects: prevalence, etiology and prevention. Int J Uro 2009; 16: 49-57.