Original Article

Birth Defects in Newborns

Frequency of Birth Defects in Newborns Admitted in Neonatal Unit of Pediatric Department in Tertiary Care Hospital Nawabshah, Pakistan

Ali Akbar Siyal, Naseer Ahmed Memon and Juverya Shah

ABSTRACT

Objective: To determine the frequency of birth defects in newborns presented in Neonatal unit of tertiary hospital Nawabshah.

Study Design: Observational / analytical study

Place and Duration of Study: This study was conducted at the Department of Pediatric Medicine, Peoples University of Medical & Health Sciences, Nawabshah from January 2015 to December 2017.

Materials and Methods: Total of 10388 neonates were received in neonatal unit pediatric ward of tertiary care hospital Nawabshah during study period. Files of all newborns were filtered to look for recorded birth defects. Physical defects detected by naked eye were registered; CNS, GIT and Cardiac defects were confirmed by Ultrasound and Echocardiography respectively.

Results: Out of 10388 neonates who were admitted in neonatal unit of our hospital,519 patients were found to have birth defects (5.09%), of these babies 260 (50.09%) were males, 239 (46.05%) were females and 20 (3.85%) presented with ambiguous genitelia. Most common defects seen in our study were from CNS (17.2%), next common defect was congenital heart disease detected in (13.5%), syndromic babies like Down syndrome, Turner syndrome, Noonan syndrome and Treacher –Collin syndrome and other syndromes. Skeletal defects were telipes deformity, polydactyly, cleft lip/palate, trachea-esophageal fistula, congenital diaphragmatic hernia and miscellaneous.

Conclusion: Birth defects are quite common in our community and impose a greater burden on health care system. The most common system involved is CNS. There is an increased need for preventive measures to be implemented in pregnant women.

Key Words: Birth defects, Congenital, Spina bifida, CNS

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INTRODUCTION

A birth defect is any deformity or deviation from the normal structure/functionthat is present since birth; it can be a physical deformity, something that we can see in a baby with naked eyes, or other birth defects like biochemical or metabolic that may need specific tests to be detected as all of these may not be apparent at birth. Congenital birth defects may be minor or major. Minor defect is defined as structural anomaly present at birth which has slight effect on clinical function, but may have a cosmetic effect e.g. pre-auricular tag. Major malformation has a noteworthy effect on function or on social acceptability e.g. ventricular septal defect (VSD) and cleft lip 1. Whatever the type of birth defect is there, it's sure a very stressful situation for family as in most cases parents has been expecting a normal newborn. In 2015 World Health Organization stated that around 303000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies[2]. The problem with congenital anomalies/ birth defects is that these are lifelong disabilities, and have a lifelong impact on baby as well as his/her family. The risk factors or causative agents have been studied extensively and are so many, but in about half of the cases no associated causative risk factors are identified[2]. Genetic susceptibility or interfamily marriages also play a significant role in birth anomalies[2, 3]. Incidence and prevalence of birth defects varies from population to population as well as different ethnic background has variable prevalence of birth defects[4]. In our country 6-9% of perinatal deaths are attributable to birth defects[5]. Globally the range for some birth defects like cleft lip/palate is different in different in low socioeconomic regions its 82/1000 live births and in high income regions its 39.7/1000 live

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births[6]. The trend has been increasing regarding the prevalence of birth defects as new threats such as Zika virus is emerging[7,8].In China a surveillance system has been created to detect/identify birth defects, no such surveillance system are working in our country but there is a high need of doing that[9]. This study is conducted in hope to look at the frequency and recognize the most common varieties of birth defects in study population so that the health authorities can design the preventive strategies accordingly.

MATERIALS AND METHODS

This study was done from the period of January 2015 to December 2017, a total of 10388 neonates were admitted in neonatal unit pediatric ward of tertiary care hospital Nawabshah during study period. Files of all newborns were filtered to look for recorded birth defects. Physical defects detected by naked eye were registered; CNS, GIT and Cardiac defects were confirmed by Ultrasound and Echocardiography respectively in the files and were taken as such.

RESULTS

Out of 10388 neonates who were admitted in neonatal unit of our hospital, 519 patients were found to have birth defects (5.09%), of these babies 260 (50.09%) were males, 239 (46.05%) were females and 20 (3.85%) presented with ambiguous genitelia (Figure 1). Most common defects seen in our study were from CNS (19.8%), next common defect was congenital heart disease detected in (18.5%), Skeletal defects like telipes deformity, polydactyly and developmental dysplasia of hip(DDH) were seen in 16.6%, syndromic presentation like Down syndrome, Turner syndrome,

Table No.1: Distribution of birth defects system wise (n=519)

S.No	System	Malformation Type	Frequency	Percentage
1	Central Nervous System		109	21%
		Meningomyelocele with/without hydrocephalus	65	
		Microcephaly Encephalocoele Anencephaly	17	
			22	
			05	
2	Cardiovascular system		94	18.5%
		Acyanotic	58	
		Cyanotic	32	
		Complex	4	
3	Skeletal system		84	16.6%
		Telipes deformity	44	
		Polydactyly	28	
		DDH	08	
		Arthrogryposis multiplex	04	
4	Syndromic babies		68	13.2%
		Down syndrome,	42	
		Edward/Patau syndrome	11	
		Turner syndrome/ Noonan syndrome	09	
		Treacher–Collin syndrome	03	
		Pierre- Robin syndrome	02	
		Prune belly syndrome	01	
5	Gastrointestinal		52	10.5%
		Cleft lip/palate,	30	
		Trachea-esophageal fistula,	05	
		Exomphalos	07	
		Imperforate anus	10	
6	Respiratory system		47	9.0%
		Choanal atresia unilateral/bilateral	17	
		congenital diaphragmatic hernia	16	
		Congenital sequestrations	14	
7	Urogenital system		25	4.8%
		Ambiguous Genitalia	20	
		Bladder exstrophy	05	
8	Miscellaneous	Multi-system involvement	40	7.7%

Noonan syndrome and Teacher–Collin syndrome and other syndromes were present in (13.2%), gastrointestinal defects like cleft lip/palate, trachea-esophageal fistula, exomphalos and intestinal atresia were present in 10.5%, respiratory problems like Choanal atresia and congenital diaphragmatic hernia were seen in 9.0% and other miscellaneous defects like a combination of multiple defects were seen in 12.4% of neonates.

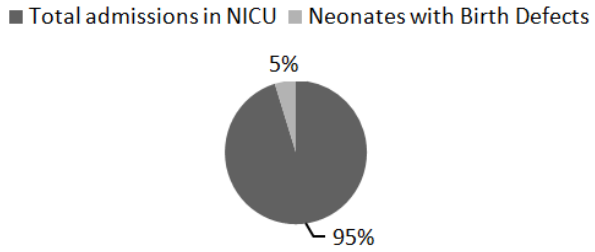

Figure No.1: Frequency of birth defects

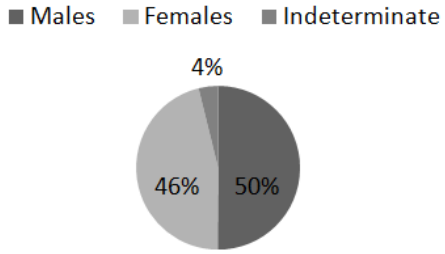

Figure No.2: Gender distribution

DISCUSSION

The frequency of birth defects in our study was 5.09%.The actual occurrence of birth defects depends on multiple factors and we know that not all newborns that have birth defects are presented to hospitals so obviously the prevalence varies among various studies even in same population and setting.The global prevalence of birth defects is 3-7% but fluctuates from country to country[10].Even few studies from Pakistan show a higher prevalence than our study[11,12,13]. Our study's frequency was comparable with a hospital based study done in Ayub Medical College and Hospital (4.23%) [14]. When we compare our prevalence rate to some international studies, our rate is somewhat higher compared with Prevalence from Nigeria[15] which has been reported as 2.7%, Taiwan[16] 4.3%, Oman[16] 2.46%, Bahrain[16] 2.7% and India[17]1.5%.

The gender difference in our study shows the prevalence of male babies affected with birth defects was 50% of study population and females were 46%. This gender difference is evident in other studies too[18], [19], but another study from Pakistan shows female predominance in their study[13].

In our study the most commonly involved system was central nervous system followed by congenital heart defects on second place and skeletal deformities were third most common defects. When we compare this data with international studies, we see in a study from Saudi Arabia the most common birth defect detected was also CNS related[20,26]. Another study from Iran and India also showed CNS malformations to be on top[21, 22]all of these support the finding of our study.But few other studiesfrom Iran and India show some contrasting results like urogenital and musculoskeletal deformities to be the most common systems to be involved[22, 23]. Some other Local studies[25,26] done in Pakistan however also show some contrasting evidence from our study like they showed that most common system involved was gastrointestinal.These differences could be due to the availability of facility regarding subspecialties which are not present in our setup. This study has its limitations so it should not be considered a true representative of the whole community, as not all babies are delivered in hospital and some parents avoid seeking medical care for neonates with birth defects.

CONCLUSION

Birth defects are definitely nota rare condition in our set-up and CNS was the most commonly affected system in our study. Knowledge of incidence and pattern of birth defects will be helpful to design preventive strategies at multiple levels by healthcare providers.

Author's Contribution:

Concept & Design of Study:	Ali Akbar Siyal
Drafting:	Naseer Ahmed Memon
Data Analysis:	Juverya Shah
Revisiting Critically:	Ali Akbar Siyal, Naseer Ahmed Memon
Final Approval of version:	Ali Akbar Siyal

Conflict of Interest: The study has no conflict of interest to declare by any author.

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