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Patterns of occurrence and management abilities of birth defects: A study from a highly urbanized coastal district of India

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ABSTRACT

Background: The present study was aimed to find out the patterns of occurrence and management abilities of Birth Defects (BDs) in Visakhapatnam, one of the north coastal district of Andhra Pradesh, India during a span of five years.

Methods: A cross-sectional investigation was held at District Early Intervention Center (DEIC), Visakhapatnam from 2016 to 2020. To identify the pattern and trend of different BDs including seasonal variations, a retrospective analysis of the health center's inpatient database for the past 5 years was done. Male and female children aged 2 months-18 years are included in the study with the prior permission of the concerned medical officer. The screening tool developed by the Ministry of Health and Family Welfare, India, was used for the study.

Results: Among 26,423 cases, children with birth defects (BDs) are 962, 2229 with deficiencies, 7516 with diseases, and 15716 with Developmental Delays (DDs) & Disabilities were admitted during the study period. From birth defects, congenital deafness occurred in large numbers with 22.66%, and neural tube defect observed in a small number of cases with 0.83% during the period. From the side of deficiencies, severe acute malnutrition has mostly occurred (66.80%) and a small number of children were affected with goiter (1.70%).

Conclusion: Through this study, it is observed that the incidence of birth defects, as well as genetic disease burden, is high in the Visakhapatnam district. Hence there is a need for strengthening of management services for these diseases in this region.

1. Introduction

Birth Defects (BDs) are one of the most frequent conditions that pediatricians experience in clinical practice. The major clinical conditions can be grouped as genetic disorders and congenital anomalies.[1] When the child fails to meet developmental milestones related to daily living, it is considered as Developmental Delays (DDs).[2] Over the past 12 years, the occurrence of DD's has increased by three percent in children under the age group of five and has reached up to 15%.[3] Worldwide, 1.5%–19.8% of children have DDs. In India, the children who are discharged from the sick newborn unit have a high prevalence of DDs.

Rashtriya Bal Swasthya Karyakram (RBSK) has been dispatched in 2013 by the Government of India focused on early intercession administrations in child health screening is to be accomplished through District Early Intervention Centers (DEICs) which are being built up in all areas the nation over for the treatment and support of these challenged children.[4] Network location achieved by Mobile health groups and Accredited Social Health Activists (ASHA's) and alluded to DEIC central command for the complete screen, analyze and treat youngsters, for this inside needs to build up institutional limits like expert labor, led preparing programs at cutting edge level for viable operationalization and diverse kind of gear. The inside offered various types of assistance like vision, intellectual, discourse, language, and dietary treatments separated from lab tests. Youngsters treated with the 30 recorded wellbeing conditions including medical procedures at tertiary level, liberated from cost.[5]

For ensuring proper health care for children, it is necessary for early detection and in time management of disorders.[6] If the DD's are not intervened in time, it can lead to manifested functional disabilities in children. In such cases, the children are being subjected to treatment after the identification of these disabilities.[7]

DEIC is engaged with a team consisting of medical officers, Pediatrician, Paramedics, and staff nurses and also provides referral support to children (1 month-18 years) diagnosed with health conditions during the screening period.

The present study investigates the prevalence of the conditions

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screened among the children attending the DEIC center at Visakhapatnam for the period of 5 years (January-2016 to December-2020) and also to find out the adequacy of institutional capacities in child health screening.

2. Methods

The present study was a cross-sectional investigation, aimed to assess the burden of DDs and their trend in the DEIC, Visakhapatnam, of south India and also to analyze the availability of manpower as well as infrastructure, so as to suggest possible improvements in the management of such diseases.

The investigation populace was shaped by the children with any of the 30 conditions alluded to DEIC from the fringe RBSK units, essential medical services units, network medical care units, region emergency clinics, children distinguished at conveyance point screening in the emergency clinic, children alluded structure the pediatric out-tolerant center and furthermore children who please their own alongside their folks/parental information. In this center, BD's were diagnosed by the pediatrician through a medical examination. As part of the study, the registers of the patients admitted at the health center with DD's during 2016–2020 were reviewed on the lines of patient sex, age and health condition including genetic disorder. The children were isolated into four age gatherings (<6 weeks, 6 Weeks to 3 years, 3–6 years, and 6–18 years) and the disparity among male and female was additionally watched. Pattern of referral, 4D Conditions screened, and an investigation of the institutional amenities like manpower and diagnostic services accessible during the examination time frame was classified by the facility checklist and semi-structured interview schedule for service providers. Present study was done taking into cognizance of the Rashtriya Bal Swasthya Karyakram (RBSK) norms which were initiated by the government of India.

Necessary permission was obtained from the institutional ethics committee, Andhra University, and also from the Andhra Medical College in this regard. During and after the survey privacy and confidentiality were ensured. SPSS Software Version 19 was used for data entry and analysis (SPSS South Asia Pvt. Ltd, Banglore, Karnataka).

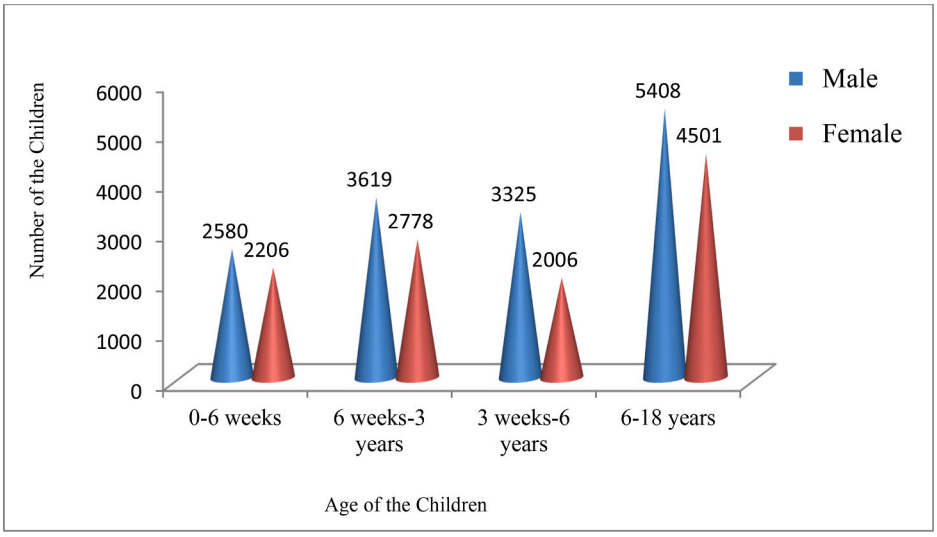
3. Results

Total 26423 children with DDs consulted the DEIC during the investigation time frame. These children were classified under the 4D's (i.e., defects, deficiencies, diseases, and disabilities) approach. From the results tabulated in graph-1, it can be observed that children below 6 weeks age group are 4786 (18.11%) numbers, of which 2508 are male and 2206 are female. The age group of one and a half month old to 3 years old, total 6397 (24.20%) kids was there of which 3619 are male and 2778 are female; and in 3 years–6 years category, 3325 are male and 2006 are female totaling to 5331 (20.17%) numbers. Children between 6 years to 18 years old are 9909 (37.50%) numbers involving 5408 males and 4501 females.

The children having conditions under each of the 4Ds are tabulated under Tables 1–4 respectively.

Table-1 shows the distribution of children according to the presence of BDs for the last five years. The different birth defects screened by the center are neural tube defect, down's syndrome, cleft lip & palate, club foot, developmental dysplasia of the hip, congenital cataract, congenital deafness, congenital heart diseases, and retinopathy of prematurity. Out of 135 BDs cases identified in 2016, the majority of cases were cleft lip & palate cases with 86 (63.70%) numbers followed by congenital heart disease cases with 33 (24.44%) numbers. In 2017, the total BDs are 157 out of which club foot cases were maximum with 57 (36.30%) numbers and congenital heart diseases cases come next with 47 (29.93%) numbers. In 2018 total number of these cases are 246, of which club foot cases were maximum with 73 (29.67%) numbers followed by congenital deafness cases with 66 (26.82%) numbers. During the year 2019, a total of 298 cases were identified with congenital deafness in a maximum of 95 (31.87%) numbers followed by cleft lip & palate cases in 66 (22.14%) numbers. For the year 2020, a total of 126 cases was found, comprising the highest number of club foot cases in 45 (35.71%) children and congenital deafness cases in 36 (28.57%) children. From all the above it is observed that the highest number of BDs occurred in 2019 and dropped to their lowest among children in 2020. However, while studying the trends in BDs data for a selected five-year period, it was observed that the prevalence rate of cleft lip & palate, club foot, and congenital deafness cases is much higher.

Table-2 shows the distribution of children according to the presence of deficiencies. These are severe anemia, vitamin- A deficiency (bigot's spot), vitamin-D deficiency, severe acute malnutrition, and goiter. The number of deficiencies is 1496, 296, 116, 261, and 60 for the years 2016, 2017, 2018, 2019, and 2020, respectively from the center. The deficiencies have diminished from a maximum of 1496 cases during the year 2016 to a minimum of 60 cases in the year 2020. During the total period of the study, it is observed that most of the children are suffering from severe acute malnutrition followed by severe anemia. To overcome this problem, a nutritional rehabilitation center was attached to this institute and is located in King George hospital, Visakhapatnam. With the help of this rehabilitation center, the nutritional status of the needy



Graph-1. Profile of the children attending at DEIC.

Table 1
Distribution of children according to presence of Birth Defects (BDs).

S.No	Name Of the BD	Prevalence total number of BDs in the study period	Year wise % of affected BDs				
			2016	2017	2018	2019	2020
1	Neural Tube Defect	8	50	25	12.5	12.5	0
2	Down's Syndrome	128	3.12	4.68	31.25	50.78	10.15
3	Cleft Lip & Palate	212	40.56	13.20	8.01	31.13	7.07
4	Club Foot	211	1.89	27.01	34.59	15.16	21.32
5	Developmental Dysplasia of the hip	0	0	0	0	0	0
6	Congenital Cataract	1	0	0	0	0	100
7	Congenital Deafness	218	1.83	7.79	30.27	43.57	16.51
8	Congenital Heart Diseases	170	19.41	27.64	22.94	21.76	8.23
9	Retinopathy of Prematurity	14	0	0	71.42	14.28	14.28

Table 2
Distribution of children according to presence of deficiencies.

S.No	Name of the deficiency	Prevalence total number of deficiencies in the study period	Year wise % of affected deficiencies				
			2016	2017	2018	2019	2020
1	Severe Anemia	430	70	23.02	5.34	1.62	0
2	Vitamin A Deficiency (Bitot's Spot)	88	78.40	18.18	0	3.40	0
3	Vitamin-D Deficiency	154	92.20	3.89	2.59	1.29	0
4	Severe acute malnutrition	1519	62.40	11.45	5.85	16.39	3.88
5	Goiter	38	94.73	2.63	0	0	2.63

Table 3
Distribution of children according to presence of diseases.

S.No	Name of the diseases.	Prevalence total number of diseases in the study period	Year wise % of affected diseases				
			2016	2017	2018	2019	2020
1	Skin Conditions	740	37.56	49.59	10.81	1.62	0.40
2	Otitis Media	771	93.12	6.87	0	0	0
3	Rheumatic Heart Disease	39	92.30	7.69	0	0	0
4	Reactive Airway Disease	577	54.24	36.04	7.79	1.90	0
5	Dental Caries	5109	36.75	7.84	20.70	28.36	6.32
6	Convulsive Disorders	280	**26.42**	58.92	2.5	0.35	11.78

Table 4
Distribution of children according to presence of developmental delays and disabilities (DDs & Disabilities). Others: growing up concerns, substance abuse, feel depressed, delay in period cycles, torment during period, agony or copying sensation while peeing and release/Foul smelling release from the genitourinary zone.

S.No	Name of DDs and disabilities.	Prevalence total number of DDs and disabilities in the study period.	Year wise % of affected DDs & disabilities				
			2016	2017	2018	2019	2020
1	Vision impairment	3231	47.91	9.16	12.53	28.93	1.45
2	Hearing impairment	2228	17.81	14	30.02	26.61	11.53
3	Neuro Motor impairment	2263	10.69	29.47	30	27.37	2.47
4	Motor Delay	1659	5	35.86	30.50	27.30	1.32
5	Cognitive Delay	965	51.70	14.81	15.54	17.92	0
6	Language Delay	2438	15.87	21.32	32.03	27.92	2.78
7	Behavior Disorder(Autism)	127	29.13	16.53	14.17	24.40	15.74
8	Learning Disorder	2115	25.62	4.63	29.88	33.09	6.76
9	Attention Deficit Hyperactivity Disorder	59	47.45	18.64	15.25	11.86	6.77
10	Others	631	3.48	19.65	12.67	62.91	1.26

children is being improved.

Table-3 indicates the distribution of children according to the presence of diseases. Among the list of diseases, skin conditions, otitis media, rheumatic heart disease, reactive airway disease, dental caries, and convulsive disorders were present. The number of diseases is 3297, 1197, 1190, 1473, and 359 from the center for the years 2016, 2017, 2018, 2019, and 2020, respectively. During the total period of the study dental caries were found in a maximum number of children and the second most common disease found was otitis media. Year by year there is a gradual decline in the number of cases with diseases and this is due to public awareness and stringent steps taken by the government towards diseases.

Table-4 demonstrates the distribution of children according to the presence of disabilities. Among these vision impairment, hearing impairment, neuromotor impairment, motor delay, cognitive delay, language delay, behavior disorder (autism), learning disorder, attention deficit hyperactivity disorder, and other inabilities like growing up concerns, substance abuse, feel depression, delay in period cycles, torment during the period, agony or copying sensation while peeing and release/Foul-smelling release from the genitourinary zone. 3785 disability cases were found in 2016, of which there were a maximum of 1548 (40.89%) vision impairment cases followed by common learning disorder cases with 542 (14.31%) numbers. In 2017 the total cases coming under disability are 2787 of which neuromotor impairment

cases were the highest with 667 (23.93%) numbers and motor delay cases were the next with 595 (21.34%) numbers. In 2018 the total cases under this section are 3929, in which language delay cases are topping the list with 781 (19.87%) numbers followed by neuromotor impairment cases with 679 (17.28%) numbers. During the years 2019 and 2020 the total number of cases under this category are 4590 and 625 respectively of which vision impairment, hearing impairment, and learning disorders have mostly occurred. While studying the data, it is observed from the trends in disabilities for the selected period that the occurrence rate of vision impairment and language delay disorders was much higher.

On investigation of the institutional amenities like manpower and diagnostic services accessible in the DEIC, it is observed that the posts of a pediatrician, dental specialist, physiotherapist, optometrist, audiologist cum speech therapist, early interventionist cum exceptional instructor, lab technician, staff nurse-1, staff nurse-2, and social worker were filled as per the sanctioned strength and they were working throughout the study period. The post of a medical officer, psychologist, and manager stayed empty all through the examination time frame. Pediatricians and 40% of supporting staff are having knowledge of genetic testing and awareness towards genetic disease management. Regular record maintenance is taking place in this center; however, much information related to genetic disorders is missing. Also observed that the center does not have genetic counselors. It was found that there are no investigation facilities for genetic diseases. Related diagnostic tests were virtually non-existent in the center with the exception of basic blood, serum, and urine diagnostic tests. The needy children are being referred to KGH or private diagnostic laboratories for specific genetic tests and for confirmation.

4. Discussion

BDs are persisting throughout the world. Due to the high mortality rate of affected infants in low-income countries, it can be admitted that the impact of birth defects is higher in these countries. Even in the children who have BDs and still survive, due to no timely intervention, these disorders are causing irreversible lifetime complications with mental or physical disabilities and these children are about 3.2 million in number around the world per each year.[8]

The present study found a significantly increased prevalence of BDs in children in the selected region. Our study revealed that a total of 26,423 cases were admitted through the 5 years period. This prevalence is higher than that reported by Prajna Bhide and Anita Kar wherein it was stated that the affected births with surveillance of congenital anomalies are as many as 472,177 in India each year.[9]

The findings of the present study reveal that the gender distribution of admitted patients is 14932 (56.51%) male and 11491 (43.48%) female. Such a study is also made in the past by Valla et al. and it was identified that the male sex is associated with a high risk of having DD's.[10] Another previous study by Dabar et al. illustrates that there is no relationship between gender and DDs.[11]

With regard to BDs, our findings provide evidence of the rise of these defects over time. Majority children had congenital deafness (218; 22.66%) followed by cleft lip & palate (212, 22.03%). Our study confirms the findings of several other studies which reported that the prevalence of congenital deafness is more in India and 63 million people suffer from significant auditory loss, due to a lack of skilled manpower and human resources for the management of this defects.[12] The second most common defect was cleft lip & palate. Our estimates however have to be considered as best-available data, as previous analysis on the cleft lip in south India also reported similar findings.[13,14]

In respect of deficiencies, during the selected study period the most common deficiency was severe acute malnutrition with 1519 (68.14%) cases followed by severe anemia with 430 (19.29%) cases. Our findings are also supported by one of the previous surveys conducted by measuring weight for height during a ten-year period which states that children under the age of five years are mostly suffering from severe acute malnutrition.[15] Regarding anemia, similar findings reported by Avina Sarna et al., who identified that the prevalence of anemia is higher from newborns to adolescents in India.[16]

Regarding diseases, it is observed that most of the children had dental caries. The admission trend of these cases is high in 2016 and then declined in the next year and again rises from 2017 to 2019. Previous findings by Abhishek Mehta et al., are supporting these trends, wherein it was reported that a large number of Indian children have been affected by dental caries.[17] In the present study, we also observed a gradual declining trend of otitis media, skin conditions, and reactive airway disease. Similar findings were observed in the previous studies that the prevalence of otitis media,[18] skin conditions,[19] and reactive airway disease[20] are low during the past years. This is probably due to the reason that the people are now much aware of these diseases and they approach the health care personnel in time.

From the side of disabilities, vision impairment has occurred in most of the children in about 3231 (20.55%) numbers and the occurrence was alternatively rising and declining through the years. Similar findings were observed by Murthy et al., that the incidence and prevalence of loss of sight in children is varying during their study period in India.[21] This was followed by language delay with (2438, 15.51%) cases, and their occurrence has risen from 2016 to 2018 and then declined. Overall, these results are slightly higher when compared to the prevalence in developed countries as reported by Wren.[22] The present study reveals that the trend of admission of neuromotor impairment cases was being consistent with a large number up to 2019 and then diminished. In the past, population-based studies reported that the neurological disorders in rural India are higher and were found in about 6–8 million people.[23] The present study divulges that the peak incidence of deafness was during 2018; and this finding corroborates with the study findings of Nagapoornima et al. wherein it was reported that due to failure of timely screening of newborns, most of them are facing hearing impairment in India.[24]

Institutional facilities are human resource and infrastructural facilities and are important in offering quality assistance to children regarding genetic disease management. In the present study, on examining the manpower we observed that the medical officer, psychologist, and manager were not available throughout the study period. Due to the non-availability of manpower in such key posts, there is a devastating effect on the treatment of the kids with DDs. The rest of the working staff from DEIC, Visakhapatnam is well aware of their respective duties in the management of these disorders. Further, only basic biochemical tests are being conducted in the center and there are no specific genetic testing facilities available here. Private diagnostic laboratories in the city are being referred to for genetic tests. With regard to public health concerns, Institutional facilities form the foundation for improving public health. The entire public health services are dependent on the availability of basic infrastructural facilities and manpower capacities.[25]

There was a lack of advanced equipment required for identication of the genetic diseases. The total infrastructural amenities are important for top-notch health frameworks. Past examinations likewise detailed that the helpless accessibility of laboratory amenities and diagnostic apparatus is additionally deterrents to quiet appraisal and find, in any event, when suppliers know about the fundamental tests.[26] High-quality care is determined by thorough assessment, detection of asymptomatic and parallel conditions, accurate diagnosis and suitable and appropriate treatment. To build up DEIC infrastructural facilities, it is essential to replace the damaged apparatus for enrichment of the program. The findings of the present study will be useful input for early intervention centers to improve action plans in child disease management programmes. They can also serve as outlook for the RBSK operations in their future programs to face the challenges identified by early Intervention centers.

5. Conclusion

Through this study, the institutional facilities at DEIC, Visakhapatnam as well as the high incidence of birth defects in Visakhapatnam district were observed. The study discloses the need for strengthening management services for these disorders in this region so that the prevalence of birth defects can be minimized. National health agencies use such data for the design and evaluation of birth defects. The proposals for better management of birth defects in the selected area are; increasing the number of genetic testing units, improving the skills and expertise of the health care personnel with respect to birth defects, and developing national policies for reinforcing related services. In addition, more extensive studies are needed across the nation to determine the distribution of birth defects and their causes for overall understanding and management of the same.

Declaration of competing interest

None.

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