HOSPITAL BASED BIRTH DEFECTS SURVEILLANCE WITH SPECIAL REFERENCE TO NEURAL TUBE DEFECTS: A STUDY OF ONE YEAR STATISTICS FROM A TERTIARY CARE GOVERNMENT TEACHING HOSPITAL OF NORTH COASTAL ANDHRA PRADESH

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ABSTRACT

INTRODUCTION

Birth defects (also called congenital anomalies) are defined as abnormalities of both structures or functions that are present at birth and are of prenatal origin. They contribute to a significant proportion of perinatal, neonatal & childhood mortality as well as morbidity.

AIM

To study at birth, prevalence of selected birth defects among babies delivered at King George Hospital, Visakhapatnam, from Jan 2015 to December 2015, appropriate management strategy of the most common neural tube defects.

RESULTS

Among the total deliveries of 6088 in the above period, the incidences of birth defects were 88 (1.44%), the most common among them were neural tube defects.

CONCLUSION

Birth defect surveillance and standardisation of the data is essential to understand the public health burden and to design appropriate prenatal prevention programs.

KEYWORDS

Birth Defects, Neural Tube Defects, Prevention, Surveillance, TIFFA scan.

HOW TO CITE THIS ARTICLE: Vijayasekhar MV, Vani I, Manoja V. Hospital based birth defects surveillance with special reference to neural tube defects: A study of one year statistics from a tertiary care government teaching hospital of North Coastal Andhra Pradesh. J. Evid. Based Med. Healthc. 2016; 3(38), 1873-1876. DOI: 10.18410/jebmh/2016/416

INTRODUCTION: Globally 7.9 million children are born annually with serious birth defects of genetic or partially genetic origin. WHO estimated that out of one million neonatal deaths in 2012 in South East Asia region, about 46,000 (94.6%) are attributed to birth defects. The prevalence and spectrum of birth defects varies among different communities and regions depending on the presence or absence of these risk factors.[1,2]

Since many factors are preventable, a wide range of prevention and intervention approaches like legislation controlling, management of toxic chemicals, vaccination against rubella, folic acid supplementation and fortification of foods with micronutrients, prevention and management of syphilis and timely identification of a family risk of inherited diseases and carrier screening with genetic counselling are the important strategies in prevention.[3,4] Early identification of at-risk pregnancies is the next most important step.

Financial or Other, Competing Interest: None.
Submission 23-03-2016, Peer Review 04-04-2016,
Acceptance 14-04-2016, Published 12-05-2016.
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DOI: 10.18410/jebmh/2016/416

All high-risk birth defects should have a mandatory anomaly scan at 18-22 weeks of gestation wherein all major externally visible major defects can be identified by an expert sonologist. For those with risk to cardiovascular anomalies, another repeat scan at 26-28 weeks' gestation at which time the 4-chambered cardiac anatomy can be best visualised should be done, if possible with foetal echocardiography.[4,5,6]

Sl. No.	External visible	Others
1.	Neural Tube Defects	Down syndrome
2.	Orofacial defects Cleft lip/ palate	Congenital diaphragmatic hernia
3.	Talipes equinovarus	TEF
4.	Limb reduction defects	Exstrophy of bladder
5.	Hypospadias	Congenital Heart Disease
6.	Exomphalos/ omphalocoele	Others
7.	Gastroschisis	
8.	Imperforate anus	

Table 1: Major Birth Defects in Babies.[1,7]

To improve the data and information, there need to be a detailed surveillance mechanism. All hospitals should identify and report the listed externally visible major birth defects at the earliest.

MATERIALS & METHODS:

OBJECTIVE: To study the birth prevalence & distribution of selected birth defects among live and still births in KGH from Jan 2015 to Dec 2015 – one-year period. To discuss management option of the most common birth defects of neural tube. To formalise preventive and to reduce recurrence of these birth defects in future pregnancies.

Inclusion Criteria:

1. All the foetuses or babies live birth or still birth, delivered with birth defects recognised either at birth or before discharge are included in our study.
2. All babies delivered after 28 weeks of gestation or birth weight more than 1000 g only were included.
3. Case definitions of the birth defects were in accordance to WHO & CDC catalogue.

Exclusion Criteria: Babies less than 28 weeks of gestation or less than 1000 g were excluded, all the defects were notified to appropriate authorities and data analysed.

RESULTS: Out of 6088 deliveries in the study period, birth defects were 88, amounting to an incidence of 1.4%. Out of 88 cases, 69 were externally visible and 19 were internal defects, categorised under others group.

Sl. No.	Category	No. of cases	Percentage
1	Neural tube defects	48	54.54 %
2	Orofacial clefts	06	6.81%
3	Hypospadias	04	4.54%
4.	Congenital talipes equinovarus	04	4.54%
5.	Limb reduction defects	01	1.1%
6.	Exomphalos or omphalocoeles	03	33.405%
7.	Gastroschisis	01	1.1%
8.	Imperforate anus	02	2.27%

Table 2: Classification of Externally Visible Birth Defects N=69

Sl. No.		No. of Cases	Percentage
1.	Anencephaly	12	13.63%
2.	Craniostenosis	2	2.27%
3.	Iniencephaly	2	2.27%
4.	Encephalocoele	10	11%
5.	Spina Bifida Meningocoele, Meningomyelocoele, Myelomeningocoele, Myelocoele, Rachischisis	12	13.63%

Table 3: Sub Classification of Neural Tube Defects: N=48

Fig. 1: Encephalocoele

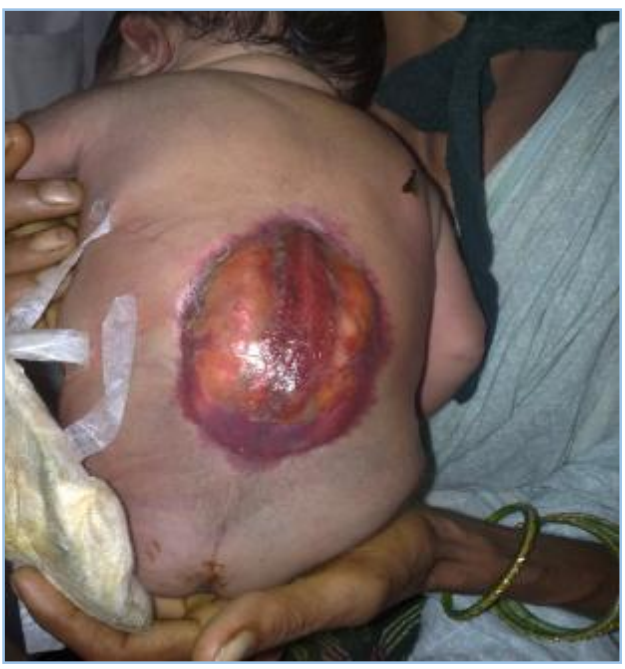

Fig. 2: Rachischisis

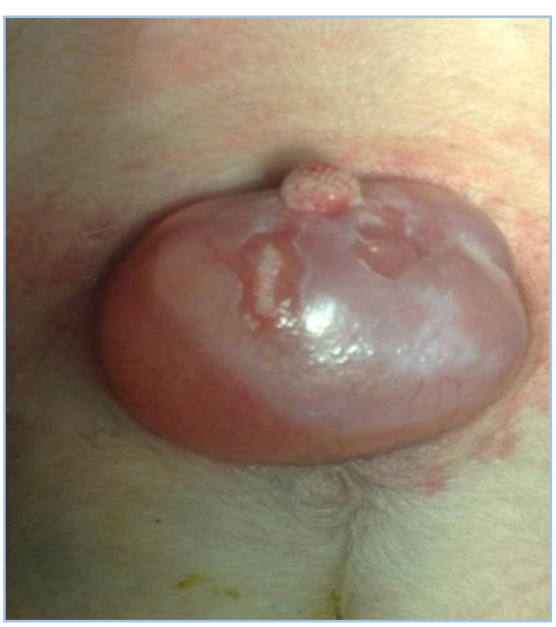

Fig. 3: Myelomeningocoele

Fig. 4: Anencephaly

Type	Number	Percentage
Down Syndrome	2	2.27%
Congenital Diaphragm Hernia	4	4.54
Congenital Heart Diseases	8	9.08%
Trachea Oesophageal Fistula	2	2.27%
Exstrophy of Bladder	1	1.1%
Others	2	2.27%

Table 4: Classification of Internal Birth Defects N=19

(All the percentages in Tables 2, 3, 4 were calculated among total birth defects N=88). Among the total 88 birth defects, majority were neural tube defects. (48)

Encephalocoeles, spina bifida, meningocoele and its variants constituted 22 cases and such defects can be easily identified by a sonologist, if a timely anomaly scan was performed in those cases. Similarly, orofacial clefts, exomphalos, and gastroschisis contributed to 10 cases which can also be recognised easily on an ultrasound scan antenatally.

Congenital diaphragmatic hernias, heart diseases, alimentary fistulae, and urological defects which were the most common internal birth defects are easily identifiable antenatally by an expert sonologist.

NTDs	7
Orofacial clefts	4
Hypospadias	1
CTEV	1
Imperforate anus	1
Congenital heart diseases	4
Tracheo-oesophageal fistulae	1

Table 5: Birth Defects Missed Antenatally on USG

Majority of cases who delivered babies or foetuses of birth defects had at least one ultrasound in 2nd or 3rd trimesters, either by a sonologist or qualified gynaecologist. The percentage of missing the anomalies in the present study was 11%.

DISCUSSION: Primary prevention especially for neural tube defects is by folic acid supplementation. In prenatal preventive medicine, this is the only entity which is partially preventable. As a primary prevention, the women with previously defective child with NTDs, subsequent pregnancies can be maintained from pre-conception to delivery by supplementation of folic acid, 5 mg/day.[8,9]

Secondary prevention of NTD is by early diagnosis. Most of them can be easily recognised by 18-22 weeks of pregnancy. Anencephaly was the most common NTD in our series and can be diagnosed as early as 11 weeks of pregnancy. The accuracy of ultrasound in detecting congenital anomalies is 80% and 98% for NTDs. It is possible to terminate the pregnancies before 20 weeks if they are timely diagnosed. This can avoid burden on the society in general and psychological trauma to parents in specific.[10,11]

1.	Expertise of observer
2.	Prevalence of congenital anomalies in the population
3.	Timing of USG
4.	Quality of US machine
5.	3D and 4D reconstruction
6.	Number of scans

Table 6: Factors Influencing Accuracy of Detection of Anomalies on USG.[4.5]

1.	Maternal age at conception
2.	Proportion of unplanned pregnancies
3.	No antenatal care
4.	Medical conditions of mother
5.	Consanguineous marriages
6.	Carrier status of genetic disorder
7.	Nutrition status

Table 7: Risk Factors for Occurrence of Congenital Birth Defects in Indian Scenario.[8,10]

The screening should be performed by a sonologist adequately trained in antenatal scans and foetal medicine. Sufficient time should be allocated to each scan. In high risk cases with previous anomalies, multiple scans periodically done will improve the detection rates. However, the screening for anomalies should start at sufficiently early gestation so that the decision for terminating such pregnancies if at all any defects are detected can be taken within MTP rules limits.

1.	Increase in the minimal age for marriage
2.	Pre-conception folic acid
3.	Genetic counselling
4.	Regular antenatal care
5.	Screening for hereditary disorders
6.	Anomaly scans at 18–22 weeks of pregnancy
7.	Preventing exposure to teratogenic agents in pregnancy
8.	Preventing congenital rubella syndrome by immunisation
9.	Avoiding smoking, alcohol and industrial teratogens
10.	Screening and treating maternal diabetes in pregnancy

Table 8: Strategies for Prevention of Birth Defects in Indian context.[8,10,12]

CONCLUSION: Birth defects surveillance and standardisation of the data is essential and prompt reporting to appropriate authorities is mandatory. This will ensure proper understanding of the logistics of the present problem of public health burden contributed by defective new born and helps the authorities to design appropriate prenatal preventive strategies and management strategies of those children.

Correctable anomalies should be taken up for surgeries if possible by the institutes free of cost. Timely antenatal scans by experienced members will improve the detection rates of these anomalies in the antenatal period itself and provide option of MTP if detected before 20 weeks.

Providing folic acid supplementation to all antenatal mothers from preconception to delivery is excellent primary prevention strategy.

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