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Original Research Article

A STUDY ON CONGENITAL ANOMALIES OF CENTRAL NERVOUS SYSTEM

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ABSTRACT

Background: CNS malformations are one of the most common among congenital malformations rating to 5-10% of total malformations. Therefore, the present study was performed to evaluate the congenital anomalies in the central nervous system.

Materials and Methods: Patients of paediatric age group (1-12) years suspect to have congenital anomalies of CNS were followed up. Data on imaging studies were collected and tabulated. Antenatally diagnosed cases of CNS anomalies were followed up. Examination for associated anomalies of head, eyes, ears, nose, face, palate, thorax, abdomen and genitalia, back, upper limb, lower limb and additional points if any was also recorded. The Ultrasound, CT scan, MRI scan and other relevant investigations if any was also performed.

Results: In the present study 30 cases of congenital malformations were identified. Out of 30 cases 14 of them were neural tube defects, 13 microcephaly, 2 Dandy Walker alformations and one holoprosencephaly. Out of 14 neural tube defects 9 of them are lumbar myelomeningocoele, two encephalocoeles in the occipital area, one cephalocoele in the parietal area and one cephalocoele in the frontoethmoidal region and one anencephaly. Maximum number of cases of CNS anomaly were of neural tube defects 14/30 (47%). Microcephaly was found to be 13/30 (43.3%). Neural tube defects were found to be more in primigravida and microcephaly in multigravida.

Conclusion: Recognition of aetiological factors permits implementation of preventive measures in the society to decrease the incidence of this dreadful condition. In all cases of microcephaly, associated anomalies of eyes or ears or facial dysmorphism were noticed.

KEY WORDS: Dandy Walker Malformations, Holo-prosencephaly, Lumbar Myelomeningocoele, Cncephalocoeles, Consanguinity.

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INTRODUCTION

Malformations were known to exist from time immemorial. About 2500 BC gross malformations were recognized in ancient Egypt. In fourth century BC Aristotle recognized malformations. To begin with structural abnormalities which were readily visible were recognized as malformation. There are a lot of such characters in Indian mythology like Kabanthan who had his head low positioned and Ashtavakram with kypho-scoliosis [1]. In 18th and 19th century embryology developed as a science. Embryologic research flourished and causations of malformations were understood and they were given a scientific basis. In early 20th century more explorations were done and the mechanism of malformations was more clearly understood.

Environmental causations of malformations particularly of central nervous system (CNS) were first described by Aschenheim in 1920 [2].

He published a child with microcephaly whose mother was exposed to radiation during pregnancy. Gregg- I discovered that rubella infection can cause serious malformation [3]. Lenz described chemical induction of malformation by thalidomide [4]. By the end of 19th century X-rays were discovered which started a new era in the research methodology of anatomy. We were able to identify the anomalies of internal organs with imaging techniques. By the mid 20th century cross sectional imaging was produced which helped a lot in understanding and characterizing the lesions.

CNS malformations are one of the most common among congenital malformations rating to 5-10% of total malformations. Most of CNS malformations lead to death of foetus in utero or death in infancy or early childhood. They cause severe disability and patients have a short life span. Some of them are caused by gene mutations and some of them by environmental factors. But majority of malformations are of unknown aetiology. National Institute of Health of United States in 1975 a panel of experts from Bethesda Maryland, discussed and suggested the following definitions - "A malformation is a primary structural defect that results from an error in morphogenesis". Deformation is an alteration in shape and structure of a previously formed part [5].

There are several aetiological factors thought to be responsible for congenital anomalies of CNS. They include drugs, alcohol, viral infection, exposure to radiation, exposure to chemicals.

CNS develops from ectoderm. Knowledge of the development of brain is essential for understanding the congenital anomalies.

In this study anomalies resulting from disturbance of CNS development upto stage 3 were included. Disorders arising from stage IV, V and VI are beyond the scope of this work, as those cases constitute part of cliniteria of neurophysicians. Therefore, the present study was performed to evaluate the congenital anomalies in the central nervous system.

MATERIALS AND METHODS

Patients of paediatric age group (1-12) years admitted to Pediatrics and Neurology wards during the period of 2002 April to 2003 April suspect to have congenital anomalies of CNS were followed up. Imaging studies were collected and tabulated. Other relevant investigations were collected to arrive at meaningful conclusions. Antenatally diagnosed cases of CNS anomalies were followed up. The anthopometric and aetiological parameters like gender, age, district, community, socio- economic status, consanguinity, family history of congenital anomalies, gravida, abortion, still birth, maternal diabetes, drug intake, maternal age, exposure to radiation,exposure to chemotherapy, febrile illness in 1st trimester, obesity, maternal malnutrition, folic acid supplementation, zinc supplementation, psychological condition of mother and contraceptive failure was recorded.

Examination for associated anomalies of head, eyes, ears, nose, face, palate, thorax, abdomen and genitalia, back, upper limb, lower limb and additional points if any was also recorded. The Ultrasound, CT scan, MRI scan and other relevant investigations if any was also performed.

RESULTS

In the present study 30 cases of congenital malformations were identified. Out of 30 cases 14 of them were neural tube defects, 13 microcephaly, 2 Dandy Walker malformations and one holoprosencephaly (Fig-1).

Out of 14 neural tube defects 9 of them are lumbar myelomeningocoele, two encephalocoeles in the occipital area, one cephalocoele in the parietal area and one cephalocoele in the frontoethmoidal region and one anencephaly (Fig-2). Distribution among Hindus and Muslims was almost equal with a slight increase among Hindus. The Hindu patients were 53.33% and Muslim patients were 46.67% (Table 1). Only 3 belong to middle class. Rest of 27 are from low socioeconomic class. They are educationally and financially backward. They have low nutritional status (Table 2). 19 patients were males and 10 of them females. In one, sex was not determined because foetus was terminated at 26 weeks. The Male patients were 65.51% and female patients were 34.48% (Table 3).

The number of patients belongs to family history of nervous disorders, consanguinity,gravida, maternal age, abortion, drugs,

febrile illness, folic acid supplementation, Zinc supplementation was shown in table-4. Fourteen patients showed abnormalities of eyes (Table-5). Eleven patients showed abnormalities of ears. Eight patients had anomalies of both eyes and ears. Six of them are microcephalics and two of them with neural tube defects (Table-6). In the present study conducted, maximum number of cases of CNS anomaly were of neural tube defects 14/30 (47%). Microcephaly was found to be 13/30 (43.3%). Neural tube defects were found to be more in primigravida and microcephaly in multigravida (Table-7).

Fig. 1: Different types of congenital malformations identified in the study.

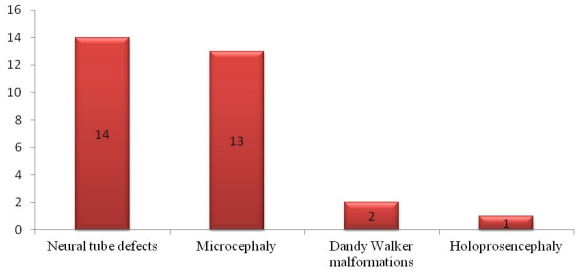

Fig. 2: Different types of neural tube anomalies identified in the study.

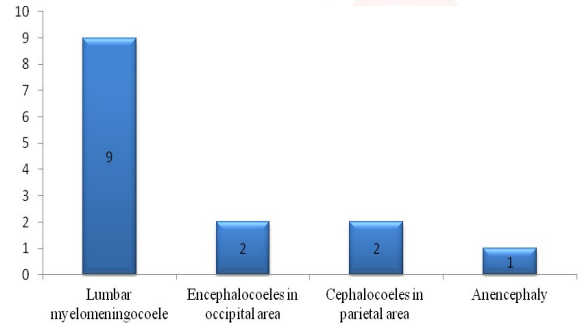

Table 1: Community wise distribution of patients in the study.

Community	No. of cases	Percentage
Hindus	16	53.33
Muslims	14	46.67
Total	30	100

Table 2: Socioeconomic status of patients in the study.

Socioeconomic status	No. of cases	Percentage
Middles class	3	10
Low class	27	90
Total	30	100

Table 3: Gender wise distribution of patients in the study.

Sex	No. of cases	Percentage
Males	19	63.33
Females	10	33.33
Sex not determined	1	3.33
Total	30	100

Table 4: Number of patients depending on various parameters.

Parameters	Number
Family history	4
Consanguinity	8
Gravida	11- primigravida 12- Second gravida 7- Multigravida
Maternal age	Mothers age between; 20 years—3 babies 20-30 years—11 babies 30-35 years—5 babies 38 years----1 baby
Abortion:	4
Drugs	02 mothers- antiepileptic drugs 02 mothers- antidiabetic drugs
Febrile illness	02 mothers in first trimester
Folic acid supplementation	13 mothers- ignorant about regular intake of folic acid during pregnancy.
Zinc supplementation	No mothers give a positive history of regular intake of zinc tablets.

Table 5: Number of patients belongs to associated anomalies with eyes.

	No. of cases	Percentage
Microcephalics	**2**	**66.7**
Neural tube defects	**1**	**33.3**
Bilateral proptosis	8	50
Hypertelorism	4	25
Optic atrophy	1	6.25
Bilateral papilloedema	1	6.25
Sunken eyes	1	6.25
Diminished vision	1	6.25
Total	16	100

Table 6: Number of patients belongs to associated anomalies with ears.

	No. of cases	Percentage
Large prominent ears	7	63.63
Low set ears	2	18.18
Malformed right pinna	1	9.09
Diminished hearing	1	9.09
Total	11	100

Table 7: Number of patients belongs to other associated anomalies.

	No. of cases	Percentage
Neural tube defects	14	46.7
Microcephaly	13	43.3
Dandy Walker malformations	2	6.7
Holoprosencephaly	1	3.3
Total	30	100

DISCUSSION

The aetiological factors which are having important roles are found to be intake of drugs like glibenclamide and sodium valproate, consanguinity, irregular intake of folic acid, low

socio economic conditions and febrile illness during first trimester. Low socioeconomic condition denotes nutritional deficiency during pregnancy, when the cells are undergoing active proliferation in the embryo [6]. Recognition of aetiological factors permits implementation of preventive measures in the society to decrease the incidence of this dreadful condition. In all cases of microcephaly, associated anomalies of eyes or ears or facial dysmorphism were noticed. Ultrasound screening of pregnant ladies is a very useful and harmless method of investigation for the early diagnosis of anomalies, so that a foetus with a gross anomaly can be terminated. Early diagnosis also helps in intrauterine correction of anomalies like spina bifida.

Increased risk of neural tube defects among people in low socio economic group has offered a clue to the factors that poor families are different form affluent families. Poor nutrition is the obvious reason. Effect of poor nutrition is magnified in the developing embryo where active cell proliferation occurs [7]. Risk factor for microcephaly includes alcohol use, inadequate weight gain during pregnancy, inadequate prenatal care, black race and low education.

In this study 27 out of 30 patients belong to low socioeconomic class. They are educationally backward. Their nutritional status is poor. They cannot afford to take balanced diet with plenty of fruits and vegetables in their diet. Fruits and vegetables are good source of folic acid. They do not know the importance of prenatal care. Family studies show multiple affected siblings' in microcephaly. Consanguinity provides strong evidence for an autosomal recessive mode of inheritance in microcephaly (Penrose 1956, Koch 1959) [8,9]. It was also noted that abnormalities of the neural tube were common among siblings. 6.5% in anencephaly and 4.4% in spina bifida [10].

Though maternal age was found to be associated with various anomalies, there was no association of maternal age and congenital anomalies in the present study. Abortion is the way of nature to get rid of nonviable embryos. In this study 4 cases out of 30 (13.3%) give history of previous abortions. Products of conception of 25 cases were analysed and no causes of malformations were detected and the incidence of malformations in offsprings of mothers taking anticonvulsants drugs was 6%.

In the present study two epileptic mothers taking valproic acid and two diabetic mothers taking glibenclamide gave birth to offsprings with CNS malformation. Two epileptic mothers are having babies with neural tube defects. Out of two diabetic mothers, one gave birth to baby with encephalocoele and one to baby with microcephaly. Fever and hyperthermia in early pregnancy increased the risk of neural tube defects [11]. It was reported that the viruses interfere with production of neurons as well as their migration [12]. In the present study mothers of two babies gave history of febrile illness in the first trimester. One gave birth to baby with neural tube defect and one to baby with microcephaly.

The importance of folic acid supplementation in the prevention of neural tube defects is given in much literature. As a method of primary prevention of neural tube defect, daily intake of 400 micrograms of folic acid is advised to women of reproductive age group by Public Health Service in 1992. In other countries like Australia and Netherlands women are advised to take 400 micrograms of folic acid every day. Grain flour is fortified with folic acid and women are advised to take plenty of fruits and vegetables.

CONCLUSION

Aetiological factors having important roles are found to be intake of drugs like glibenclamide and sodium valproate, consanguinity, irregular intake of folic acid, low socio economic conditions and febrile illness during first trimester. Low socioeconomic condition denotes nutritional deficiency during pregnancy, when the cells are undergoing active proliferation in the embryo. Recognition of aetiological factors permits implementation of preventive measures in the society to decrease the incidence of this dreadful condition. In all cases of microcephaly, associated anomalies of eyes or ears or facial dysmorphism were noticed.

Conflicts of Interests: **None**

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