Original article

Study of anomalous babies of neural tubal defects and their mothers

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Abstract

Introduction: Neural tube defects occur due to failure of closure of neural tube at its cephalic end .If failure takes place at cranial region, then anencephaly occurs. If failure of closure occurs from cervical region onwards caudally then spina bifida occurs. It usually involves the lumbo-sacral region. Commonly anencephaly, spinabifida, myelocele myelomeningocele occurs as neural tube defects. Environmental and genetic factors give rise to Neural tube defects.

Methods: Patients presenting to the Pediatrics department as well as from the Labor ward and antenatal ward of Obstetrics and Gynecology department in the 'Teaching Hospital' of 'Sree Raja Rajeswari Medical College', Bangalore were included in the study. It was conducted from 2007 to 2008 for a period of fifteen months.

They were studied in detail with proper family history, personal history, drug history, Obstetrics history and were later examined in detail.

Results: Sixteen patients were observed out of these 5 were live births. Of them, two were males and three females. Five were still births and seven cases were born to consanguious parents. Eleven cases were first child to their parents. Mothers of three anomalous babies were diabetic, two were epileptic and the rest were normal.

Key words: Anencephaly, craniospinal anomalies, myelocele, myelomeningocele, encephalocele, rachischisis

Introduction

Neural tube defects are the defects that occur due to abnormal closure of neural fold in the 3rd and 4th week of intra uterine life. The common structures involving in the formation of neural tube defects are meninges, vertebrae, muscles, and the skin. The incidence of neural tube defects is 1 in 1000 in USA; 1 in 500 in north and South Carolina and 1 in 200 in Northern China. Environmental and genetic factors give rise to Neural Tube Defects.[1] In the past only external anomalies were detected but after the invent of ultrasonography there has been a revolution in the diagnosis of congenital anomalies of Central Nervous System (CNS).[2] Anomalies of CNS is frequently observed now a days. Any failure of fusion at more than one or more sites during the above period will give rise to Neural tube defects.[2]Numerious studies have been conducted on Neural tube defects in India and abroad.

Campbell conducted a study on Anencephaly and concluded that it is due to failure of closure of anterior neuropore which normally closes by the completion of 46 days of conception. This anomaly can be diagnosed by 11-12 weeks of pregnancy.[3] Ultrasound has been used to determine the condition of placenta and to check prenatal mortality and outcome of the foetus.[4]. By application of alpha protein screening, a study conducted in India, detected 27 (1.55%) cases of anomalies whereas only 8.48%

anomalies were diagnosed by ultrasonography during the antenatal period.[2] In another study, false diagnosis was made in three cases (0.17%).[5] Another study on anomalies with neural tube defects conducted in the East Delhi Hospital identified only seven patients of anencephaly out of thousand births. Patients with hydrocephalous were also diagnosed along with neural tube defects in the later part of pregnancy.[6]

Methods

This study was carried out in the Pediatrics and Obstetrics and Gynecology department of the Teaching Hospital of Sree Raja Rajeswari Medical College, Bangalore. It was conducted from 13.02.2007 to 12.05.2008 for a period of fifteen months.

Neonates and their mothers admitted in the respective departments were examined in detail for the presence of any congenital Neural Tubal Defects. Congenital Anomaly was detected in three stages; Congenital Anomaly Detected in the antenatal check up during routine ultrasound, Congenital Anomaly defected in a stillbirth child following normal delivery or an aborted child, Congenital Anomaly defected after live births delivered either normally or after Lower Segment Caesarean Section (LSCS).

All the pregnant women were subjected to a detailed history that included;

1) Date of last Menstrual Period (LMP).
2) Drug intake during the first trimester;
3) History of
   Epilepsy,
   Systemic Diseases like tuberculosis, diabetes, fits, etc;
   Any exposure to Radiations during first trimester;
   Bad Obstetrics History (BOH); (Repeated Abortions in the previous deliveries; Stillbirths; Presence of any Congenital Anomaly)
   Any history of similar Congenital Anomaly in the family;
   Any history of chromosomal anomalies like Down's Syndromes etc.

Along with routine investigations like haemogram and urine routine, special investigation like scanning / ultrasound of the involved systems and were also studied in detail. If deemed necessary, MRI was also done.

Scanning was done by explaining the procedure to the patient and written consent of the patient was taken. The procedure was done in full bladder in supine position with jelly smeared over the abdomen. It was carried out in longitudinal, transverse and oblique positions. The aim of the scanning was to identify twin pregnancy if any; cardiac activity of the fetus; presentations and positions of the fetus; age of gestation and correlating it with Last menstrual period (LMP); weight of the fetus; the location and length of placenta; the quantity of liquor and the presence of any congenital anomalies. Ultrasonography of the abdomen was repeated at least three or four times till the later part of pregnancy.

After delivery, local examination of the new born baby was carried to identify the presence of any congenital anomaly and the type if any present. A post natal scan was performed to see the integrity of the skeletal architecture as well as internal organs. In case of still births, after a written consent, autopsy was performed.

Results

There were 8 types of craniospinal anomalies mainly found in 12 males & 4 females. Anomalies were found in 4 still births, 8 new borns, 2 in preterm babies and in one 15 days old female child. There were 13 children who were first born, two were gravid-2 and one was gravid-4 (Table 1).

Table 1: List of congenital anomalies with age, sex, parity and type of marriage

S.No	Anomalies	sex	age	Parity	Consanguoious/ nonconsanguious
1	Hydrocephalus with cervical Meningocele:-	male,	New born	Primi	Consanguoious
2	Fetal Hydrocephalus & lumbosacral meningomyelocele	male	New born	Gravida-2	Non consanguious
3	Fetal Occipital Encephalocele with Rochischisis	male	28 weeks	Gravida-4	Consanguoious
4	Encephalocele.	male	Still birth	Primi	Non Consanguoious
5	Anencephaly	male	New born	Primi	consanguoious
6	Open spina bifida(RACHISCHISIS)	male	Still birth	Primi	consanguious
7	Fetal Hydrocephalus with lumbo sacral meningioma	male	Preterm	Primi	Non consanguious
8	Gross hydrocephalus associated with cervical Meningocele	male	New born	Primi	consanguoious
9	Arnold Chiary Syndrome of type-II	female	24 weeks	Primi	consanguious
10	Hydraencephaly	Male	New born	Primi	Non consanguious
11	Spina Bifida at lumbar level and lumbar meningocele	female	New born	Gravida-2	consanguious marriage
12	Lumbar Myelomeningocele with Paraplegia of lower limbs.	female	New born	Primi	Non consanguious
13	Infected Myelomeninmgocele	female	15 days	First child	consanguious parents
14	encephalocele:-	male	Aborted fetus	Primi	Non consanguious
15	Anencephaly	male	Aborted fetus	Primi	Non consanguious
16	Complete Spina bifida with Anencephaly	male	New born	primi	consanguious

Five had Hydrocephalous associated with Cervical and Lumbar Meningocele and Myelomeninigocele; one with Lumbar Myelomeninmgocele with Paraplegia of lower limbs; one had Occipital Encephalocele with Rochischisis. Three had Fetal Encephalocele associated with Rochischisis and Complete Spina bifida. Seven were born to consangious

parents and eleven were first child (Fig. 1). One patient had Arnold Chiary Syndrome (type-II) and Hydraencephaly and two had Anencephaly.

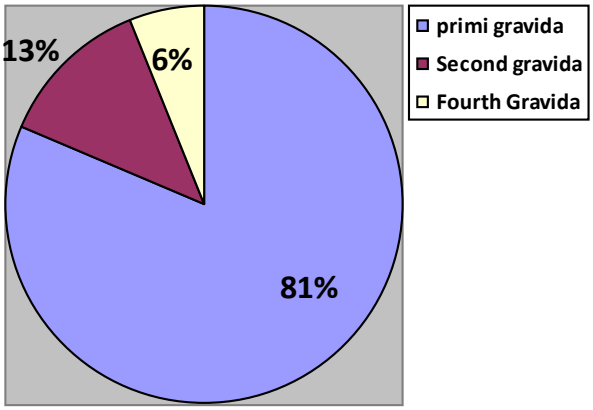

Fig. 1: Parity of the patients

There were eight types of Cranio-spinal anomalies. Of them, Meningomyelocele and Rachischiasis were found to be the most common (18.75%). Hydraencephaly and Arnold Chiary Syndrome of type-II were found to be the least common (6.25%) (Table 2).

Table 2: Various Cranio-spinal anomalies

S.No	Types of Anomalies	Number	Percentages
1	Hydrocephalus	2	12.5%
2	Meningomyelocele	3	18.75%
3	Meningocele	2	12.5%
4	Anencephaly	2	12.5%
5	Rachischisis	3	18.75%
6	Hydraencephaly	1	6.25%
7	Encephalocele	2	12.5%
8	Arnold Chiary Syndrome of type-I I	1	6.25%

Discussion

Incidences of congenital malformation vary from countries to countries. The incidences of anomalies in Afghanistan is 5.5%, in Michigan 3.4% , in Northampton shire it is 0.9%.[7-9] In India, incidence of anomalies varies from 0.3 to 3.6%.[10] In Chandigarh & Pondicherry, incidence of anomalies is quite high may due to higher rate of autopsies that reveal the malformations.[10] In Nepal, overall incidences of malformations is 2.8%.[11]

The studies on anomalies by various workers in India & abroad have shown higher incidences in one of the above systems. A study in India showed musculo-skeletal anomalies as the commonest in live births followed by gastrointestinal and central nervous system.[12] Amongst defects in central nervous system seen in still births in their studies, the frequency of neural tubes defects was 1.68/ 1000 & Anencephaly was 1.34/1000 births.[12] Neural defects are said to be higher in Northern India and congenital malformations are said to be six times more in still births.[12] For the occurrences of congenital malformations, consanguineous marriages seems to play a prominent role.[13] In the other parts of India, incidence of neural tubes defects ranged from 0.5 to11.8/1000.[14] Davangere of Karnataka has reported the highest incidences of neural tube defect.[15] Central nervous system defects have been reported to be highest in the Indian subcontinent.[16,]

A study conducted in China on 150 normal pregnant women for calcium and pregnancy induced hypertension revealed that supplementation of calcium during pregnancy may bring about incidence of pregnancy induced Hypertension.[17]

An epidemiological study on hypertension in a non pregnant patients suggested that abnormal calcium metabolism may be the contributing factors for the onset of hypertension. They also studied serum ionized calcium, total calcium, phosphorus magnesium, total protein and albumin in 16 non pregnant women, 31 pregnant women with pregnant induced hypertension (PIH) and 12 patients with chronic hypertension. They found no differences in serum ionized calcium between the non pregnant normal women and those with hypertension.[18]

Lower levels of serum calcium and higher levels urea and uric acid was observed in patients with pre-eclampsia at 28 weeks of pregnancy in a study conducted in Pakistan and hence, they suggested that this could be used as an early diagnostic tool for pre-eclampsia.[19]

In our study, fifteen cases were observed out of these five were live births, two males and three females. Five were still births–aborted babies and seven were born to consanguious parents.

Mothers of three anomalous babies were hypertensive (20%); four had pregnancy induced hypertension (PIH)

(26.66%); three were diabetic (20%) and two were epileptic (13.33%). The mothers were not taking any drugs nor were exposed to radiation but seven of them had history of consanguinity of second degree (inbreeding). This might be an important contributing factor for the occurrence of anomalies in children.

Conclusions

Frequency of congenital anomalies vary from race to race and region to region. With advancement of investigations, abnormalities can be detected early during the antenatal period. Consanguious marriage also is a major contributor for the occurrence of congenital anomalies hence, social awareness regarding marriage practices with in a community should also be addressed.

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