Craniospinal Anomalies and their Management

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Abstract

Background: To find out the frequency of various craniospinal anomalies and their management in Rawalpindi

Methods: In this cross sectional study patients with congenital gross craniospinal anomalies, were included. All patients were assessed from head to toe for any other associated anomalies. Patients with high neural tube defects, cases associated with other gross anomalies and children below 15 days of age were deferred for operation. Patients with craniospinal anomalies were advised for CT scan brain and Spinal X ray. In Hydrocephalus patient’s ventricular tap was performed and CSF was sent for routine examination before inserting ventriculoperitoneal shunt. Patients who were expected to have adequate quality of life and with amenable neural tube defects were operated after proper investigations.

Results: A total number of 160 cases of craniospinal anomalies were presented. Majority of the cases were males (57%). The most common anomaly observed was of hydrocephalus (66.25%) and second most common anomaly was of myelomeningocele (14.37%). There were 2 cases of anencephaly, which were managed conservatively because of their incompatibility with life. Majority of the cases of hydrocephalus were operated on priority basis to restrict their head size. Cases of hydrocephalus which presented in patients above 6 months of age and with enlarged 3rd ventricle underwent 3rd ventriculostomy while others underwent shunting procedure.

Conclusion: Patients presenting with high neural tube defects and have systemic congenital defects should be treated by non-operative measures. Patients presenting with amenable neural tube defect and who are expected to have adequate quality of life should be given proper surgical treatment along with proper counseling.

Key Words: Craniospinal anomalies, Hydrocephalus, Neural tube defects

Introduction

Neural tube defects (NTDs) are a group of complex congenital malformations of the brain and spinal cord that arise due to failure of closure of neural tube during embryogenesis. During this malformation there is abnormal closure of neural fold in the 3rd and 4th week of intra uterine life and the structures commonly involved in its formation are meninges, vertebrae, skin and muscles. NTDs are found in about 1:1000 pregnancies in the US. In UK the prevalence of NTDs in absence of selective abortions or antenatal diagnosis, is 3-4 per 1000 births. In Pakistan the incidence found was higher that is 13.9 per 1000 births. The commonest NTDs found in Pakistan are hydrocephalus and anencephaly. After the congenital heart diseases, NTDs are second in frequency among the major public health problems. There are various causes of NTDs, the commonest causes include genetic susceptibility, deficiency of folate concentration, environmental factors, defect in metabolic pathways that play an important role in closure of neural tube during fetal development and even in utero drug exposure. Prenatal supplementation with folic acid decreases the incidence of birth defects due to folate deficiency.

The neural tube is a neuro-ectodermal derivative and it forms the brain vesicles, i.e., prosencephalon, mesencephalon and rhombencephalon by enlarging at the cranial end. There may be defects of morphogenesis due to deformation, disruption or malformation and the cause of malformations is mainly due to genetic disturbance. One of the risk factors for chromosomal defects is increasing age of the mother. If there is some uterine or pelvic abnormality of the mother or any other mechanical distortions, it can lead to deformations. The cause of disruption of fetal tissues is mainly ischemia, anoxia, infections (TORCH) or even radiations.

NTDs can be of various types. These can be due to defects of neural folding (meningoceles and myelomeningoceles), can be due to incomplete dysjunction such as in dermal sinus and associated dermoid and epidermoid tumors, can be due to...
premature dysjunction such as in spinal lipomas. There can be disorders of gastrulation seen in combined spina bifida, split cord and neurenteric cysts. Another cause is disordered secondary neurulation (thickened filum, myelocystocele) or disordered postneurulation development seen in encephaloceles. There can also be failure of caudal neuraxial development leading to caudal agenesis.

The cases having NTDs are sometimes associated with other abnormalities such as vertebral, anorectal, cardiac, tracheo-esophageal fistula, renal and limb abnormalities (VACTERL). There is another syndrome called CHARGE syndrome which is named for its six major clinical features that include; coloboma of the eye, heart defects, atresia of the choanae, retarded growth and developmental anomalies. These developmental abnormalities can be CNS anomalies, genital hypoplasia, urinary tract anomalies, ear anomalies or even hearing loss.

Treatment for NTDs mainly include either termination of pregnancy or in utero intervention. For postnatal cases, conservative or operative procedures are done. According to Lorrbre criteria in cases with high neural tube defect, gross hydrocephalus and associated other abnormalities only conservative management should be done. Higher incidence of NTDs in Pakistan can be ascribed to lack of balanced diet leading to deficiency of folic acid. It is observed that the serum B12 levels are lower in pregnancies with NTDs and it is also observed that there is reduction of recurrence rate of NTDs to about 85-100% in women who took folic acid prior to their subsequent pregnancies.

Patients and Methods

In this cross sectional study patients with congenital gross craniospinal anomalies, admitted in the Neurosurgery department of District Head Quarter Hospital Rawalpindi were included, over a period of 1 year from January 2013 to December 2013. All patients were assessed from head to toe for any other associated anomalies. Patients with high NTDs, cases associated with other gross anomalies and children below 15 days of age were deferred for operation. The families were informed about the condition and prognosis of the disease and about the treatment option available. Patients with craniospinal anomalies were advised for CT scan brain and Spinal X ray. In Hydrocephalus patient’s ventricular tap was performed and CSF was sent for routine examination before inserting ventriculo-peritoneal Shunt. Patients that were expected to have adequate quality of life and with amenable neural tube defects were operated after proper investigations.

Results

A total number of 160 cases of Craniospinal anomalies were presented in Neurosurgery department. Majority of the cases were males (57%). The most common anomaly observed was of hydrocephalus (66.25%) and second most common anomaly was of myelomeningocele (14.37%) (Table 1; Figure 1&2). There were 2 cases of anencephaly that were managed conservatively because of their incompatibility with life. Majority of the cases of hydrocephalus were operated on priority bases to restrict their head size. Cases of hydrocephalus that presented in patients above 6 months of age and with enlarged 3rd ventricle underwent 3rd ventriculostomy while others underwent shunting procedure. Mode of treatment for other craniospinal anomalies(Table 2).

Table 1: Frequency of Various Craniospinal anomalies (n=160)

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>No (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hydrocephalus</td>
<td>106 (66.25)</td>
</tr>
<tr>
<td>Anencephaly</td>
<td>2 (1.25)</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>9 (5.6)</td>
</tr>
<tr>
<td>Meningocele</td>
<td>8 (5)</td>
</tr>
<tr>
<td>Lipomeningocele</td>
<td>5 (3.12)</td>
</tr>
<tr>
<td>Myelomeningocele</td>
<td>23 (14.37)</td>
</tr>
<tr>
<td>Craniosynostosis</td>
<td>5 (3.12)</td>
</tr>
<tr>
<td>Diastematomyelia</td>
<td>2 (1.25)</td>
</tr>
</tbody>
</table>

Discussion

The incidence of NTDs is variable depending upon the ethnicity, geographical location and gender. It is usually more common in areas where maternal folic
acid intake is very low and there is also an increased incidence in female fetus and Hispanic patients.\textsuperscript{12}

### Table 2: Craniospinal anomalies-Mode of treatment

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>Mode of Treatment</th>
<th>No. of Cases with Conservative management</th>
<th>No. of Cases with Operative management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hydrocephalus</td>
<td>Nil</td>
<td>1</td>
<td>VA Shunt =15</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>VP Shunt = 80</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>3\textsuperscript{rd} Ventriculostomy = 10</td>
</tr>
<tr>
<td>Anencephaly</td>
<td>2</td>
<td>Nil</td>
<td></td>
</tr>
<tr>
<td>Encephalocele</td>
<td>Nil</td>
<td>9</td>
<td>Excision and repair</td>
</tr>
<tr>
<td>Meningocele</td>
<td>Nil</td>
<td>8</td>
<td>Excision and repair</td>
</tr>
<tr>
<td>Lipomeningocele</td>
<td>Nil</td>
<td>5</td>
<td>Excision and repair</td>
</tr>
<tr>
<td>Myelomeningocele</td>
<td>Nil</td>
<td>23</td>
<td>Excision and repair</td>
</tr>
<tr>
<td>Craniosynostosis</td>
<td>Nil</td>
<td>5</td>
<td>Strip cranectomy</td>
</tr>
<tr>
<td>Diastematomyelia</td>
<td>Nil</td>
<td>2</td>
<td>Excision and repair</td>
</tr>
</tbody>
</table>

There are many other risk factors such as females who have insulin dependent diabetes, obesity, those taking anti convulsants and females having vitamin B-12 deficiency.\textsuperscript{4}

In our country there is a lack of balanced diet in most of the areas therefore the incidence of NTDs is quite higher although no reliable statistics are available. By the help of ultrasonography NTDs can be usually identified as early as first trimester.\textsuperscript{13} There is a lack of such antenatal screening in many areas by which those females cannot be identified who are at high risk of having such abnormalities.\textsuperscript{14} Due to lack of such facilities most of the patients are diagnosed near term or at the time of labor. There is also lack of preconception counseling by the healthcare providers especially in cases where there is chance of recurrence of NTDs in successive pregnancies. Routine screening by alpha fetoprotein is important in cases where no defect is visualized on ultrasonography but the alpha fetoprotein levels are raised.\textsuperscript{15}

The treatment mainly given in such cases can either be termination of pregnancy, in utero intervention or postnatal management. In case of central nervous system anomalies there is a chance of spontaneous abortion. Termination of pregnancy is dependent upon the maternal age and severity of such abnormalities.\textsuperscript{16} In certain cases in utero intervention is required such as ventriculo-amniotic shunt and meningocele repair. There is lack of proper screening and intrauterine intervention during pregnancy in order to detect CNS anomalies in our setup so we rely mainly on postnatal measures.

In this study most of the cases were of male children. Similar results were seen in a study by Raza A et al in which 62.5% of cases were of males.\textsuperscript{10} In another study by Bilodi AK there were total 8 types of craniospinal anomalies which were found in 12 males and 4 females.\textsuperscript{2} In a study by Nawaz F et al out of 3,350 deliveries 18 fetuses presented with neural tube defects and the incidence was of 5.3 cases per 1000 deliveries.\textsuperscript{17}

Out of all the craniospinal anomalies, the commonest anomaly noted in most of the studies is hydrocephalus\textsuperscript{18,19}. In our study the most common anomaly noted was also hydrocephalus (66.25%). The patients who presented early were treated by VP (Ventriculoperitoneal) shunting and the patients who presented late in life were treated by 3\textsuperscript{rd} ventriculostomy or VA (Ventriculoatrial shunting). In one study most of the cases were of hydrocephalus, present in 27 cases (42.18%). In another study by Kumar R et al there were 58.8% cases of hydrocephalus and out of this 63.3% of children presenting with overt hydrocephalus had shunt surgery prior to definitive surgery whereas 23.3% of cases required a shunt after the MMC was closed.\textsuperscript{20}

Similar results were seen in a study by Moin et al, reported hydrocephalus in 42.1% cases, anencephaly in 31.6% cases, spina bifida with meningocele in 15.8% cases, Meningomyelocele in 5.3% case and Dandy Walker syndrome in 5.3% case.\textsuperscript{21} In Pakistan, Perveen F et al\textsuperscript{2} reported hydrocephalus and anencephaly as the most common NTDs in Pakistan. In another study conducted in Peshawar by Khattak ST et al maximum number of cases (45.60%) were hydrocephalic whereas 17.39% were of Meningocele and Spina bifida.\textsuperscript{5}

In present study Anencephalic fetuses were very few (1.25%) but in a study conducted by Behrooz A in Iran, it was the most frequent anomaly i.e 53.6% and they reported 5.4% cases of meningomyelocele.\textsuperscript{22}

In present study there were 14.37% cases of meningomyelocele but in some studies it was the most frequent NTD such as in Switzerland (70%) and in Jordan (85%). In a study conducted by Raza A et al there were 2 cases of anencephaly which were not compatible with life and died within 12 hours. There were seven patients that presented with encephaloceles out of which occipital were 6 and only 1 case of frontonasal encephalocele.\textsuperscript{10} Whereas in our study there were 9 cases of encephalocele.
It is important to differentiate syndromic NTDs from isolated forms in terms of its pathogenesis, genetic counseling and prognosis. During the last 30 years there has been improvement regarding prenatal diagnosis and now a days there is availability of various methods for prenatal diagnosis. The risk of NTDs is reduced by 35-70% by consistent supplementation but the recommendations are not effective enough to decline birth prevalence in our country and also in other countries world-wide. The females that are at risk should have folic acid fortified food. In countries like Canada and USA, food fortification is already being done due to which folate status has been considerably improved. The prevalence of NTD has further reduced to 25-50% due to food fortification by folic acid.

Conclusion

1. Methods of prenatal diagnosis of craniospinal anomalies needs to be explored.
2. The utility of folic acid administration in preventing neural tube defects requires to be highlighted.
3. Patients that present with high neural tube defects and have systemic congenital defects should be treated by giving non-operative measures.
4. Patients presenting with amenable neural tube defect and who are expected to have adequate quality of life should be given proper surgical treatment along with proper counseling.

References