

PATTERN AND FREQUENCY OF CRANIO-SPINAL ANOMALIES

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ABSTRACT

Objective To find out number and type of cranio-spinal anomalies and management given.

Study design Observational study

Place & Duration of study Department of Paediatric Surgery and Neurosurgery at Nawabshah Medical College Nawabshah, from January 2006 to December 2007.

Patients and Methods All patients with gross CNS anomalies were included in this study. Data related to type of anomaly, gender, age, location etc were recorded on a pre-designed proforma. Parents were counseled regarding prospects of survival, operative procedure and the quality of life. Genetic and family planning advice was given. Those considered candidate for surgery were investigated using ultrasound and CT scan according to the type of the lesion.

Results A total of sixty four patients were received during the study period. Males were in dominance ($n = 40-62.5\%$) and most of them belonged to rural areas. Fifty two cases (81.25%) were operated and 12 cases (19.75%) managed conservatively. The most common anomaly was isolated hydrocephalus ($n=27$) followed by meningocele and myelomeningocele ($n=17$). Both lesions combined found in 7 patients. None of the mothers had taken folic acid supplementation before conception.

Conclusions Patients with gross major CNS defects and associated with systemic problems do not need active surgical intervention. Patients with amenable neural tube defect and expected of adequate quality of life require operation after proper counseling. There is a need of public awareness of folic acid supplementation in the antenatal period and of antenatal screening to detect anomalies.

Key words Neural tube defects, CNS anomalies, Spina bifida, Hydrocephalus, Encephalocele.

INTRODUCTION:

The neural tube, a neurectodermal derivative, closes in zipper fashion in caudocranial direction. At the cranial end neural tube enlarges and form the brain vesicles i.e. prosencephalon, mesencephalon and rhombencephalon. The problem of morphogenesis may be due to malformation, deformation and disruption. Malformations may be due to genetic disturbance. Increasing age of the mother (and, to a lesser extent, of

the father) may increase the risk of chromosomal defects. Deformations may be due to mechanical distortions resulting from uterine or pelvic abnormality. Disruption of the fetal tissues occurs due to anoxia, ischemia, radiation or infections (TORCHS: Toxoplasmosis, Rubella, Cytomegalovirus, Herpes and Syphilis).¹

Anomalies may be due to defects of neural folding (neural tube defects such as meningoceles and myelomeningoceles), incomplete dysjunction (dermal sinus and associated dermoid and epidermoid tumors), premature dysjunction. (spinal lipomas), disorders of gastrulation (combined spina bifida, split cord, neurenteric cysts), disordered secondary neurulation (thickened,

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filum, myelocystocele), failure of caudal neuraxial development (caudal agenesis) and disordered postneurulation development (encephaloceles). Periconceptual usage of folic acid reduces the incidence of neural tube defects.² Mahadevan and Bhat have found increasing frequency of neural tube defects in Southern India.³

Associated other abnormalities may be vertebral, anorectal, cardiac, tracheo-esophageal fistula, renal and limb abnormalities (VACTERL). CHARGE syndrome was first described by Pagon, and named for its six major clinical features, which are; coloboma of the eye, heart defects, atresia of the choanae, retarded growth and developmental anomalies, which include CNS anomalies, genital hypoplasia and/or urinary tract anomalies, and ear anomalies and/or hearing loss.

Treatment options include termination of pregnancy, in utero intervention or postnatal conservative or operative procedure. According to Lorbrer criteria if patient has high neural tube defect, gross hydrocephalus and associated other abnormalities, such cases may be left as such and managed conservatively. Pattern and frequency of CNS anomalies presented in the Neurosurgery and Pediatric surgery ward were evaluated by the authors for their frequency and risk factors involved.

PATIENTS AND METHODS:

The records of all patients with congenital gross CNS anomalies admitted in the Paediatric surgery and Neurosurgery departments of Nawabshah Medical College were analyzed over a period of 2 years from January 2006 to December 2007. All patients were assessed from head to toe for any other associated anomalies. Anencephaly patients were observed. Patients with high neural tube defects and associated with other gross associated anomalies were also deferred for operation. The families were given a realistic appraisal of the severity of the condition, its prognosis, and the medical care available, and they actively participated in decisions.

Hydrocephalus patients were advised for ultrasound and/or CT scan brain. Ventricular tap was performed and CSF was sent for D/R and C/S before inserting ventriculo-peritoneal Shunt. Patients with amenable neural tube defect and expected of adequate quality of life were operated after proper counseling.

RESULTS:

Over a period of 2 years, 64 cases with CNS anomalies were seen. Males were in dominance (n 40 – 62.5%) and most of them belonged to rural areas. Male to female ratio was 1.66. The distribution of various cranio-spinal anomalies is shown in table 1.

Anencephaly patients were not compatible with life and both babies died within 12 hours. A case having holoprosencephaly

Table 1: Distribution of Cranio-spinal Anomalies

CNS Anomaly	No. of Cases	Mode of Treatment	
		Conservative	Operative
Anencephaly	02 (3.12%)	02 (3.12%)	NIL
Hydrocephalus	20(31.25%)	03 (4.68%)	17(26.56%)
Teratoma (fronto-orbital)	01 (1.56%)	NIL	01 (1.56%)
Encephalocele	Occipital	06 (9.37%)	06 (9.37%)
	Frontonasal	01(1.56%)	01(1.56%)
Craniosynostosis (coronal)	01 (1.56%)	NIL	01 (1.56%)
Dermoid	02 (3.12%)	NIL	02 (3.12%)
Myelo-meningocele	10 (15.62%)	03 (4.68%)	07(10.93%)
Meningocele	07 (10.93%)	NIL	07 (10.93%)
Sacrococcygeal Teratoma	03 (4.68%)	NIL	03 (4.68%)
Hydrocephalus with myelo-meningocele	07(10.93%)	03 (4.68%)	4(6.25%)
Holoprosencephaly with severe facial abnormalities	01(1.56%)	01(1.56%)	NIL
Lipomyelo-meningocele (transitional)	02 (3.12%)	NIL	02 (3.12%)
Diastematomyelia	01(1.56%)	NIL	01(1.56%)
TOTAL	64	12(18.75%)	52(81.25%)

with severe facial abnormalities was also managed conservatively. Cases with high spina bifida and severe limb deformities and with associated systemic abnormalities were deferred for operation. Fifty two (81.25%) were operated and remaining 12 cases (19.75%) were managed conservatively.

Hydrocephalus cases were the most common (n-27 42.18%). Patients with severe hydrocephalus and fused sutures were not opted for VP shunt. Ventriculo-peritoneal shunts were inserted in 21 cases (32.81%). PS Medical medium pressure shunts were used in most of the patients because of their availability from public hospital store. Seven patients presented with encephaloceles. Six were occipital and one frontonasal encephalocele. Four cases (6.25%) had large encephaloceles and microcephaly which showed severe mental retardation and having problem with feeding.

Spina bifida (myelomeningocele and meningocele, lipomyelomeningocele and diastematomyelia) was seen in 27 cases (42.18%). Two cases were identical twins with limb deformities and both were left to their natural fate. Spina bifida was seen in the family in 5 cases. Meningoceles were seen in cervical (1 case), thoracic (1 case) and lumbar (5 cases). These were excised and repaired without any

neurological deficit. Myelomeningoceles were usually seen with limb weakness and loss of sphincter control. One patient had a diastematomyelia in which bony spur in the thoracic spine was removed and cord freed.

Associated systemic congenital defects were present in 6 cases (9.37%). None of the mothers had folic acid supplementation before conception. Few had folic acid supplementation after pregnancy was confirmed. There was no particular drug history or maternal infection.

DISCUSSION:

Neural tube defects (NTDs) which are one of the commonest anomalies are seen in 1 in 1000 US population. NTDs are more common among women of certain Hispanic subpopulations, and among population groups in Ireland, China, and the United Kingdom. Use of anti-seizure medications, maternal insulin-dependent diabetes mellitus, maternal obesity, maternal hyperthermia and previous NTD-affected pregnancy are the risk factors.

According to Ordinal grading scale:⁴ grade 0 was no anomalies of a given type; grade 1 was an anomaly expected to have no impact on quality of life e.g. choroids plexus cyst; grade 2 was an anomaly expected to have little or no impact on quality of life but which might require surgical or medical treatment e.g. myelomeningocele, hydrocephalus; grade 3 was an anomaly with the potential for serious impact on quality of life, even with optimal medical or surgical treatment e.g. encephalocele and grade 4 was an anomaly incompatible with life e.g. anencephaly.⁵ Bader I and Khan NZ have reported a severe form of frontonasal dysplasia (FND) with bilateral anophthalmia who died within 40 hours.⁶ In our series 2 patients with anencephaly were observed and both died within 12 hours. One patient had holoprosencephaly with severe facial abnormalities.

Treatment options include termination of pregnancy, in utero intervention and postnatal management. Central nervous system anomalies are more likely to lead to abortion. Maternal age and severity of abnormalities directly correlates with termination of pregnancy.⁴ In utero intervention required like in meningocele repair, ventriculoamniotic shunt etc. In our set up there is no screening protocol during pregnancy to diagnose CNS anomalies and facility for intrauterine intervention is not available so we only rely on postnatal measures.

Antenatal ultrasound and MRI help in the determination of congenital anomalies. Khan et al have done ultrasound examination on 60 pregnant women aged 20-35 years and found 8 fetuses with congenital anomalies (13.3%).⁷ In the past 2 decades, the second trimester of pregnancy has been the most common time for prenatal diagnosis of fetal anomalies and chromosomal aneuploidies. Ndumbe FM et al with improvements and technological advances in transabdominal or transvaginal ultrasound scanning has recently detected

upto 80% of CNS anomalies by 13 weeks' gestation.⁸ The fetal MRI is useful in evaluation of ventriculomegaly and associated anomalies.⁹ It is also helpful in differentiating various posterior fossa cystic lesions like Dandy-Walker malformation and variant, posterior fossa arachnoid cyst, and mega cisterna magna.

There is familial tendency, but not Mendelian inheritance. Probably multiple genes involved with variable penetrance. Risk of one child with spina bifida is 0.05%. If one sibling affected, risk increases to 5%. If 2 affected, risk increases to 12-15%. Genetic counseling should be offered to all parents. Authors have observed severe gross limb abnormalities and myelomeningocele in an identical twin in which surgical intervention was deferred and there was familial history of Spina bifida in 5 cases. Fouzia et al found NTD to be the commonest (65.8%) type of anomaly.¹⁰

Hydrocephalus is the commonest CNS anomaly seen.^{11,12} Authors noted hydrocephalus in 27 cases (42.18%). VP Shunts were inserted in 21 cases (32.81%). Kotil K et al have observed large encephaloceles greater than 50mm and associated microcephaly having extremely poor prognosis.¹³ We have also similar observation in large encephaloceles and microcephaly in 4 cases which showed severe mental retardation and having problem with feeding.

CONCLUSIONS:

Patients with high neural tube defects and associated with systemic congenital defects need non-operative measures. Patients with amenable neural tube defect and expected of adequate quality of life require surgical correction after proper counseling. There is a need of public awareness of folic acid supplementation in the antenatal period; and antenatal screening.

REFERENCES:

1. Zaman W, Ziaullah T, Karamat KA. Rubella virus- cause of preventable congenital anomalies. *Pak Armed Forces Med J* 2002; 52: 92-5.
2. Scholl TO, Johnson WE. Folic acid: influence on the outcome of pregnancy. *Am J Clin Nutr* 2000; 71.
3. Mahadevan B, Bhat BV. Neural tube defects in Pondicherry: *Indian J Pediatr.* 2005; 72: 557-9.
4. Schechtman KB, Gray DL, Baty JD, Rothman SM. Decision making for termination of pregnancy with fetal anomalies: Analysis of 53000 pregnancies. *Obstet Gynecol* 2002; 99: 216-22.
5. Tahir S, Alam M, Salam F. Prevalence and management of anencephaly at divisional headquarter Hospital Faisalabad. *Pak J Med Sci* 2002;18: 302-05.
6. Bader I, Khan NZ. Severe form of frontonasal dysplasia with bilateral anophthalmia - a case report with review of literature. *Pak J Med Sci* 2005;21:82-4.

7. Khan SK, Sheikh S, Zarreen A. Ultrasound screening for fetal abnormalities. *Ann King Edward Med Coll* 2004;:138-40
8. Ndumbe FM, Navti O, Chilaka VN, Konje JC. Prenatal diagnosis in the first trimester of pregnancy. *Obstet Gynecol Surv* 2008; 63: 317-28.
9. Wong AM, Bilaniuk LT, Ng KK, Chang YL, Chao AS, Wai YY. Lobar holoprosencephaly: prenatal MR diagnosis with postnatal MR correlation. *Prenat Diagn* 2005; 25: myelomyelo296-9.
10. Perveen F, Tayyab S. Frequency and pattern of distribution of Congenital anomalies in the newborn and associated maternal risk factors *J Coll Physicians Surg Pak*. 2007;17:340-3.
11. Rehman A, Fatima S, Soomro N. Frequency of congenital anomalies and associated maternal risk factors in the lower socio-economic group. *Pak J Surg* 2006; 22: 169-73.
12. Saleem M, Arshad M, Imran A, Latif S, Iqbal S, Cheema MA. Neural tube defects: our experience with 50 patients. *Pak J Pathol Sep* 2000; 11: 85-90.
13. Kotil K, Kilinc B, Bilge T. Diagnosis and management of large occipitocervical encephaloceles: a 10-year experience. *Pediatr Neurosurg*. 2008; 44: 193-8.