Epidemiological Profile and Management of Central Nervous System (CNS) Malformations in Neurosurgery Department of Dera Ghazi Khan Medical College

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Keywords: *anomalies, congenital anomalies, malformations, central nervous system, nuerosurgery*

DOI: 10.37978/tijfs.v7i1.417

Submitted: March 06, 2024
Accepted: March 12, 2024
Published Online: March 13, 2024

How to cite this: Atta et al., 2024. Epidemiological Profile and Management of Central Nervous System (CNS) Malformations in Neurosurgery Department of Dera Ghazi Khan Medical College Int J Front Sci, 7, 1.

Significance: This study sheds light on the epidemiological landscape and current management practices of central nervous system malformations. By delineating the prevalence, demographics, and treatment approaches, the findings contribute to advancing understanding and optimizing strategies for managing CNS malformations within the local healthcare context.

Abstract

Introduction: Malformation of the central nervous system (CNS) typically leaves the family in chaos and medical team helpless. The strict use of preventative measures has significantly decreased the frequency of congenital abnormalities in industrialized nations.
Materials & Methods: At Dera Ghazi Khan Medical College conducted this Cross-sectional analytical study was conducted from January 2023 to December 2023 to evaluate the epidemiological profile and management CNS malformations. All cases of CNS malformations treated in the neurosurgery department were included in the study. The data were analyzed using SPSS version 25.0.
Results: Total 61 cases were included in the study. The patient's age ranged from 1 d to 70 years old. Six (9.8%) patients with CNS abnormalities had prenatal diagnoses. There were 34.8% CT scans, 1.7% MRIs (7 after CT), and 23.7% ultrasound scans, among other tests. In 49 instances (80.32%), there was a surgical rationale: 20 cases (40.81%) could be treated, whereas 29 cases (59.18%) did not. In 11 cases (18%), the surgical rationale was not maintained.
Conclusion: CNS abnormalities were frequent in our study. They cause numerous neurological aftereffects and permanent impairments, and are not usually amenable to surgical treatment. Therefore, it is imperative that the DG Khan region concentrates on enhancing preventative measures.

Introduction:

Congenital CNS malformations are anomalies in the structure or form of the brain and spine present at birth as a result of irregularities in CNS growth. They can arise at any point throughout intrauterine life, irrespective of the cause. There are two types of anatomical defects: post-neurulation (neurax and/or container anomalies) and neurulation (anterior and/or posterior neurospora closure abnormality). Seldom are these CNS abnormalities isolated.[1] A deformed child's birth typically results in chaos for the family as a whole and helplessness in care. Certain conditions might result in severe deformities that pose a threat to life or necessitate intricate medical interventions that provide unsatisfactory outcomes.[2] The frequency of congenital anomalies has significantly decreased in developed nations as a result of advancements in prenatal diagnosis (amniocentesis, ultrasonography, fetal MRI), genetic counseling, and the strict implementation of preventative measures.[3] Low socioeconomic status and mystical-religious beliefs are two factors that enhance the prevalence of CNS congenital abnormalities in Pakistani communities.[4]

This study aimed to assess the epidemiological profile and management of congenital brain abnormalities in the neurosurgery department of the Dera Ghazi Khan Medical College.

Materials and methods

Data for this cross-sectional study were collected retrospectively from January 2023 to December 31, 2023, in the Neurosurgery Department of Dera Ghazi Khan Medical College using a preformed questionnaire. The documentation of patients referred for hospitalization or outpatient examinations over this period comprised the study group. The study covered every patient with congenital CNS abnormalities who was admitted to the neurosurgery department during the study period.

Results Socio-demographic data

A total of 68 cases of CNS abnormalities were admitted during the study period, and 7 of those refused to participate in the study; hence, data from 61 patients were analyzed. The patients' ages ranged from 1 to 70 years. Of the 61 patients, 20 (32.78%) were between 28 days and 2 years of age and 23 (37.70%) were between 2-6 years of age. Table 1 displays the breakdown of the patients by age bracket.

Table- 1: Age distribution of the patients (n=61)

Age	Frequency	Percentage
28 days to 2 years	20	32.78
2 to 6 years	23	37.70
6 to 12 years	9	14.75
12 to 18 years	5	8.196
18 to 65 years	3	4.91
Above 65 years	1	1.63
Total	61	100

The sex distribution of the patients showed that 31 (50.8%) were females and 30 (49.2%) males. Of the 61 patients, 32 (52.5%) lived in urban settings and 29 (47.5%) lived in rural regions. In twenty-three (23) patients, CNS abnormalities were identified throughout the third to eighteenth week of gestation. Fifteen (24.59%) of spine congenital swelling, fourteen (22.95%) of macrocrania, 9 cases (14.75%) of psychomotor retardation, eight cases (13.1%) of cephalic congenital swelling, poly-malformative syndrome (5 cases, 8.19%), comitiality, 4 (6.5%), microcrania (3 case, 4.91%), intracranial hypertension syndrome (2 cases, 3.27%), and torticollis (one case, 1.16%) were the reasons for consultation. In 14 instances (22.95%), the macrocrania were associated with spinal swelling.

Physical examination revealed that of the eight cases with congenital cephalic inflammation, four (50%) had occipital inflammation, two (25%) had nasoethmoidal inflammation, one (12.5%) had frontal swelling, and one (12.5%) had naso-orbital edema. Out of the 15 cases of congenital inflammation in the spine, nine cases (60%) had spina bifida (with five cases localized in the DG Khan region, three cases in the rural region, and one case in the remote region) and six cases (40%) had scoliosis (with three cases in the DG Khan region, two cases in the remote region, and one case in the deltic region). There were 28 instances (80%) of myelomeningocele and seven cases (20%) of meningocele in 35 cases of spina bifida. Paraplegia with sphincter problems was observed in 20 (71.4%) of the 28 patients with myelomeningocele, whereas paraplegia/paraparesis without sphincter issues was observed in 51 patients (29.7%). A total of 61 instances (3.66%) had clubfoot, 2 (0.4%) had cleft lip and palate, and 1 (0.2%) had Crouzon syndrome. Trisomy of chromosome 1, bent knee, and genu valgus are among the deformities or deformations linked to these different CNS anomalies.

There were 200 (34.8%) CT scans, 09 (1.7%) MRIs (7 after CT), and 44 (23.7%) ultrasounds (31 prenatal and 13 postnatal) among other tests. 54 instances (74%) of trans-fontanellar ultrasounds, 11 (15.1%) cardiovascular ultrasounds, 5 (6.8%) abdominopelvic ultrasounds, and three instances (4.1%) swelling ultrasounds were found among the 73 postnatal ultrasounds. Postnatal ultrasonography was insufficient to diagnose CNS abnormalities in 53 (72.6 %) cases; CT was then used to confirm the diagnosis. This led to the diagnosis of CNS abnormalities in the remaining 20 patients. Out of 51 cases, 20 (39.2%) did not have prenatal ultrasonography results indicating a CNS abnormality.

Therapeutic and evaluative data

In 49 instances (80.32%), there was a surgical rationale: 20 cases (40.81%) could be treated, whereas 29 cases (59.18%) did not. In 11 cases (18%), the surgical rationale was not maintained. Following CNS malformation operations during neurosurgery, patients with concomitant extraneurally located anomalies were sent to specialized specialties. Physiotherapy was recommended for each of the 13 individuals with neurological limb disability.

The average period between surgical indications and surgery for the 49 individuals who had the chance to undergo surgery was 31.2 days, with extremes ranging from one day to eight years. A total of 228 patients underwent surgery, comprising 25 cases of hydrocephalus, 12 cases of Spinabifida, 6 cases of encephalocele, 3 cases of compressive arachnoid cysts, and 3 cases of Arnold Chiari malformations. 44 patients (89.79%) were lost to follow-up of the 49 cases with a surgical indication that could not be operated on. Three of the 22 patients of polymalformative syndrome and craniostenosis died prior to surgery. Table 2 displays an overview of the diagnosis and therapy philosophies that had the appearance of a surgical hold because of the technical platform's inadequacy.

Patients who underwent surgery had an average hospitalization of 4.3 days, with a range of 1–25 days. Evolution was favorable in 13 instances (26.53%), stagnant in 32 cases (65.30%), and unfavorable in three cases (6.12%) at the end of the study period. One patient (2.5 %) died. Over an average of 15.8 months following surgery, there was favorable progression with no sequelae in 24 cases (48.95%), mild impairment of sequelae in 20 cases (40.81%), and severe sequelae in 4 cases (4.08%). Two fatalities (1.2%) and nine follow-up losses (5%) were reported.

Discussion

Prevalence and socio-demographic aspects

Our study's yearly prevalence of CNS abnormalities (6.4%) was overlaid on previous authors' findings (5%).[5] Compared to some reported in studies, this frequency in our dataset was substantially lower (13.5%[4]; 31.8%).[6] congenital differences may be attributed, in part, to the quality of the technological platform used to diagnose congenital anomalies and, in part, to the study's selection standards. With endpoints of 0 and 10 years, the mean age at assessment for CNS abnormalities was 17.6 months.[7] This age contrasts sharply with ours (8.21 months). Researches also differ in the sex ratio. For some writers, it was 0.85[4] and 0.86.[8] A sex proportion between 1.54[4] and 1.1[7] was reported by others. The deformity was unaffected by sex.[7] In our research,

51% of the participants were from metropolitan regions. Patients who travel from remote regions to metropolitan areas to receive medical care typically stay here temporarily.

Diagnostic Aspects

In our dataset, the most common reason for hospitalization (41.8%) was congenital spinal edema. With regard to 68.1% of instances, this conclusion was additionally stated by other writers.[1] In our dataset, the combination of spine congenital edoema and macrocrania explained 8.2% of the admission causes. Moreover, 13.88% of other writers[1] made notice of it. This combination implies spina bifida-related hydrocephalus. In our study, 10.5% of patient admissions were due to a polymalformative disorder. It accounted for 29.6% of the admissions grounds.[9]

Antenatal ultrasonography was performed in 80% [10] and 97%[7] of cases. In our study, relatively few patients (n=9.7%) underwent the procedure. The limited geographical and economic availability of this study to our communities may account for this. Furthermore, our nation's current guideline[11] states that ultrasound is not required for pregnancy screening. In 39.2% of the cases in our dataset, the antenatal ultrasound results were insufficient to diagnose CNS abnormalities. In other cases, 25%[7] and 51%[12] did not receive this diagnosis, respectively. This may be because certain but ultrasound imaging quality additionally relies on deformities do not show symptoms until after delivery, the ultra-sonographer and how well they perform.

Table-2: Frequency of congenital anomalies among patients

Diagnosis	Cases with a surgical indication		Cases without surgical indication	Total	
	Operated (n)	Not Operated (n)		(n)	%
Spina Bifida	6	5	0	11	18.03
Spina Bifida + Hydrocephalus	2	4	0	6	9.83
Hydrocephalus	4	5	0	9	14.75
Cephalocele	3	3	0	6	9.83
Dandy Walker malformation	3	2	0	5	8.19
Spina Bifida + Hydrocephalus +Clubfoot	0	0	0	00	00
Hydrocephalus +Dandy Walker malformation	0	0	0	00	00
Craniostenosis	0	0	0	00	00
Intracranial arachnoid cyst	0	0	0	00	00
Spinabifida + Dandy Walker + Arnold	0	0	3	3	4.91
Malformation +Clubfoot	0	1	3	4	6.55
Microcephaly	0	0	0	00	00
Spina bifida +Clubfoot	0	1	0	1	1.63
Hydranencephaly	0	2	2	4	6.55
Arnold Chiari malformation	2	0	0	2	3.27
Corpus callosum agenesis	0	0	0	00	00
Hydrocephalus + Cephalocele	0	0	1	1	1.63
Spina Bifida + Hydranencephaly	0	1	1	2	6.55
Semi lobar holoprosencephaly	0	1	0	1	1.63
Arachnoid cyst + Microcephaly	0	1	2	3	4.91
Lissencephaly + Microcephaly	0	0	0	00	00
Schizencephaly + corpus callosumagenesis	0	0	0	00	00
Hemi vertebra of L1	0	0	0	00	00
Hemi vertebra of L1+club foot	0	0	0	00	00
Caudal regression syndrome + knees flessum + club feet	0	0	0	00	00
Intra-spinal arachnoid cyst	0	2	0	2	6.55
C1anterior archagenes is with subluxation C1-C2 + Vertebral block C6-C7	0	0	0	00	00

Lissencephaly + craniostenosis	0	0	0	00	00
Spinabifida + hydrocephalus + Clubfoot + Horseshoe Kidney	0	1	0	1	1.63
Corpus callosumagenesis + cleft lip	0	0	0	00	00
Crouzon syndrome + Semi-lobar holoprosencephaly	0	0	0	00	00
Microcephaly + Down Syndrome (trisomy21)	0	0	0	00	00
Spinabifida + Genuvalgum	0	0	0	00	00
Microcephaly + nose bone malformation		0	0	00	00
Hydrocephalus + Persistence of the peritoneovaginal canal	0	0	0	00	00
Spina bifida+ Hydrocephalus + Arnold Chiari malformation + Incomplete fusion 8th-9th ribs	0	0	0	00	00
Spina bifida+ Clubfoot+ Persistence of the peritoneopavaginal canal	0	0	0	00	00
Spina bifida + Hydrocephalus + Clubfoot + left renal ptosis	0	0	0	00	00
Schizencephaly + Megalargecistern	0	0	0	00	00
Total	20	29	12	61	100

When it comes to identifying fetal CNS abnormalities in the early stages of pregnancy, ultrasound sensitivity, specificity, positive predictive value, and negative predictive value were 85.7%, 100%, 100%, and 99.9%, accordingly.[13]Foetal magnetic resonance imaging (MRI) is more precise and provides a more thorough assessment of lesions; yet, due to ultrasound's satisfactory outcome and MRI's limited geographic and cost accessibility, MRI is recommended when ultrasound is not available.[10,14] It would be ideal in our situation if ultrasonography was required for pregnancy care and more widely available. In our study, postnatal CT was performed in 79.8% of the cases. In other studies, MRI was conducted in 100% of the instances[5]; however, in our study, this percentage was 1.7%. This may be explained by the fact that the patients in our study had severely restricted access to these healthcare imaging tools, both geographically and financially. The most prevalent problems in the literature were neural tube abnormalities.[1,4,6,7,15] Three diagnostics were made: Dandy Walker's malformation (9.1%), encephalocele (12.5%), and myelomeningocele (79.6%).[1] In 80% of cases, it was spina bifida, and in 19%, encephalocele.[4] We noted the similar thing in our series, which is likely due to the simple reason that, as healthcare series for the vast majority, neural tube abnormalities and hydrocephalus become more symptomatic, creating indications which concern and prompt patients to seek medical attention more frequently.

Depending on the type of malformation, surgery is typically used to treat CNS abnormalities.[7] In 83% of the cases in our analysis, a surgical indication was given.41.1 Of the patients in the study with an operational indication, 41.1% underwent surgery. The lack of a technological platform for the comprehensive management of some patients (polymalformative syndrome) and the high number of instances missed for follow-up (231 cases) could be partly accounted for by patients' inability to pay for the operation and partially by the lengthy waiting periods for surgery. These are some of the contributing factors to why individuals have not been operated upon despite having a surgical indication. In fact, 31.2 days was the average duration of waiting for treatment in our investigation. The inadequate technical infrastructure (equipment and buildings) and the small number of nurses may be the cause of this lengthy typical wait for operation; 18% of cases resulted in mortality.[6] The majority of individuals in our series, for whom there had been a surgery indication, according to us, died before being followed up on. Additional challenges arise when it comes to managing related malformations in this situation, given the difficulty in treating CNS anomalies. As a result, in every instance of surgical care, the related deformity was treated in many teams concurrently with CNS malformation treatment, as is sometimes preferred. Following surgery independently for all CNS abnormalities, the patient was sent to a different specialized institution for therapy of the related abnormality. In our analysis, 91.5% of the cases had a favorable or stable early postoperative evolution. It was in 84% of other places.[16] In our investigation, both medium- and long-

term development were less acceptable than rapid development, which appears to be typically satisfactory. In our analysis, there were 2 fatalities (0.4 %). These two fatalities were individuals who had hydrocephalus, one of which had a myelomeningocele. Therefore, prompt treatment is highly nonlethal. However, even in individuals without a surgical rationale, deficits and significant difficulty integrating into society should be expected when continual monitoring is done correctly.[17,18]

Conclusion

In our study, CNS abnormalities were frequently observed. Most such lesions can be diagnosed and neurosurgical intervention is an option. The three primary CNS abnormalities identified were encephalocele, hydrocephalus, and spina bifida. Nearly all patients had a surgical diagnosis, although less than half could undergo surgery. It is still necessary to enhance the operational prognosis and patient follow-up over time through interdisciplinary therapy and the participation of political administrators in order to strengthen the technological platform. Prevention should be prioritized because there are many CNS abnormalities, and their control can be challenging.

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