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Prognostic Factors in Encephalocele: A Fourteen-Year Survey at King Abdulaziz University Hospital, Jeddah

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ABSTRACT. This study describes a 14-year follow-up of infants with encephalocele seen at King Abdulaziz University Hospital from 1988 to 2002. Out of 13 cases, only ten were born among the 35,851 births recorded in that period. Only occipital subtype was found. Three of them had meningocele, nine cases had meningoencephalocele and only one had meningoencephalocystocele. Eight patients developed hydrocephaly. Only 5 of these patients needed a ventriculoperitoneal shunt insertion, one of them without encephalocele repair. One patient was diagnosed as having Patau's Syndrome (47 XY + 13) with congenital glaucoma and ventriculomegaly. One case had Meckel-Gruber syndrome. Seven patients were contacted, interviewed and/or examined in the outpatient clinic for developmental assessment. Four of them had normal mental and physical development for age. The rest were developmentally delayed. The aim of this study was to investigate the possible prognostic parameters, looking for the most influential risk factors that affected the morbidity and mortality of the study's patients. The authors concluded that the size of the bone defect, the brain content of the sac, the associated hydrocephalus and syndromes are important factors that influence the prognosis of occipital encephalocele from both mortality or morbidity points of view.

Keywords: Prognostic factors, Encephalocele, Saudi Arabia

Introduction

Like spina bifida, *bifid cranium* occurs where a defect in the skull allows the intracranial contents to bulge. The bulging may consist only of meninges (meningocele) or may contain an addition brain tissue (meningoencephalocele). Sometimes a portion of

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the occipital horn of the lateral ventricle protrude in addition: meningoencephaloventriculocele. Broadly, this lesion can be classified based on the location of the mass into occipital, parietal, frontonasal, and nasopharyngeal. The most common lesion is the occipital encephalocele. Cranial meningoceles have good prognoses; whereas, patients with meningoencephaloceles are at increased risk of having visual problems, microcephaly, mental retardation , and seizures. Generally, children with neural tissue within the sac and associated hydrocephalus have the poorest prognoses.

In this article, the authors present their experience of such patients seen over a period of fourteen years at the King Abdulaziz University Hospital (KAUH), Jeddah, Saudi Arabia. KAUH is a 400 bed teaching hospital providing health care services to a multinational population.

Material and Methods

The records of patients with the diagnosis of encephalocele, seen at KAUH during the period from July, 1988 till June, 2002, were studied. Thirteen consecutive cases were found. There were 7 girls and 6 boys. All, but three, were born at KAUH during this study period. During this period, out of 35,851 births, 10 cases of encephalocele were detected.

All were of the occipital type. Two of them had meningocele, ten cases had meningoencephalocele, and one had meningoencephaloventriculocele.

Nine of the study's patients had their encephalocele repaired primarily (six patients during the first four days of life, one at three weeks, one at six weeks, and the last at fifteen months of age). In all the nine cases, it was intended to preserve the brain tissue. In only one case (No. 5 on Table 1) a portion of the herniated neural tissue was excised. Four cases were not operated upon, one with a huge inoperable encephalocele and the other three had small sized encephaloceles covered with normal skin; one of them was Meckel-Gruber syndrome. Three patients had ventriculomegaly only by CT-scan and/or ultrasound examinations. Eight patients developed hydrocephalus. Only five of these patients needed a ventriculo-peritoneal shunt insertion, one of them without encephalocele repair.

One patient was diagnosed has having Patau Syndrome (47 XY +13) with congenital glaucoma and ventriculomegaly (case No. 3 in Table 1); he had meningoencephalocele 4 x 4 cms in diameter, that was repaired on the second day of life. On follow-up, he was found to be developmentally delayed (physical and mental) with repeated attacks of convulsions. He lost follow-up after his third birthday.

The patient with the Meckel-Gruber syndrome (case No. 4 in Table 1) had in addition to the encephalocele, a short webbed neck, short clavicle, short crowded ribs(possible hypoplastic lungs), microphthalmia, dextrocardia, and hydrocephalus. The defect

Table 1. A summary of the data of the 13 cases.

	Type of Encephalocele	Head Circ.	Site & Size Bone Defect	Associated Abnormality	Age of Surgical Repair	Fate	Time of Appearance of Hydrocephalus	Shunted	Herniated Brain Tissue	Blind
1	M.E.V	33.5	Occipital* 10 × 10 cms	Rt. Cap Hgma. & Short neck	Not done	Lost F/U	At birth		Yes	Yes
2	M.E	33.5	Occipital* 2 × 2 cms	Capillary Hemangioma	1 ½ mos.	Alive and well and is 9.5 yrs age	Arrested hydrocephalus Post-operative	No	Yes *minimal*	–
3	M.E	32	Occipital 4 × 4 cms	Patau 47 XY + 13 Polydactyl Undescended testicles	2nd day	Lost F/U after 3 yrs of age with delayed milestones	Ventriculomegaly	No	Yes	Yes
4	M.E	35.5	Occipital 2 × 2 cm	Meckel-Gruber syndrome	None	Died at 3 mos. of age in sepsis	At birth	No	Yes	–
5	M.E	29	Occipital 8 × 10 cms	None	2nd day**	Alive. Devt. Delay at 4.5 yrs	At 2 mos. of age	At 3 mos.	Yes	–
6	M.E	36	Occipital 4.5 × 4.5 cms	None	2nd day	Died at 1.3 yrs in sepsis	At 9 mos. of age	At 10 mos.	Yes	Yes
7	M.E	29.5	Occipital 8 × 8 cms.*	Hypoplastic cerebellum with agenesis of corpus callosum	4th day	Alive with delayed milestones at 2 yrs and 3 mos.	After one year	At 15 mos.	Yes	
8	M.E	36	Occipital 1.5 × 1.5 cm	None	1st day	Alive and well at 4 mos.	Ventriculomegaly	No	Yes *minimal*	–
9	M.E	35	Occipital small 1 × 1 cm	Had very low Apgar Score	None	Alive with delayed milestones at 3 yrs	At 3 mos. of age	4 mos.	Yes	–
10	M.	35	Occipital 2 × 2	Short Tendo Achillis of ✓ the left foot	At 15 mos	Alive and well at 1 year and 3 month	Ventriculomegaly	No	No	–

M = Meningocele; ME = Meningoecephalocele; MEV = Meningoencephaloventriculocele. *Lacking CSF and ulceration. **Partial neural tissue excised.

Table 1. A summary of the data of the 13 cases (continued).

	Type of Encephalocele	Head Circ.	Site & Size Bone Defect	Associated Abnormality	Age of Surgical Repair	Fate	Time of Appearance of Hydrocephalus	Shunted	Herniated Brain Tissue	Blind
11	M	33.5	Occipital* 3 x 3 cms	None	3 wks	Alive and well at 5 years	None	No	0	-
12	M.E	34	Occipital* 2 x 2 cms	Rt. U.D.T.	Not Done	Lost F/U after 1 year	Ventriculomegaly	No	Yes *minimal*	–
13	M.E	35	Occipital 1 x 1 cms	None	4th day	Alive with delayed mile-stones at 6 years	at 6 mos. of age	At 6 mos.	No	-

M = Meningocele; ME = Meningoecephalocele; MEV = Meningoencephaloventriculocele. *Lacking CSF and ulceration. **Partial neural tissue excised.

was 2 x 2 cms, covered with normal skin. He died at three months of age from severe respiratory distress and sepsis.

Results

Table 1 shows a summary of the data of the 13 cases. The incidence of encephalocele over the 14-year study period in KAUH was 0.28/1,000 births. Sex was not a determining factor as almost equal number of patients were seen in both sexes. All thirteen patients were born after full-term pregnancy, two of them were small for date. Only two patients were the products of gestational diabetes pregnancy, otherwise no significant antenatal history was detected. Antenatal ultrasound was done to seven of the patients and only three were diagnosed as having encephalocele. Two of the occipital encephaloceles were ulcerated with leaking CSF.

At birth, weight of the ten patients had normal head circumference and the remaining two had microcephaly. Seven out of the ten patients developed hydrocephalus. Four of them had A V-P shunt inserted, the fifth did not need one because of arrested hydrocephalus and the sixth case continued treatment in another hospital at 3 months of age and lost follow-up. The last case was not shunted because of the poor outcome expected with Meckel-Gruber syndrome. He died at three months of age. Three patients had dilated ventricles associated with their encephalocele, one had Patau syndrome and the other two had associated brain atrophy.

Three patients were blind, two were hydrocephalic, and all had brain tissue herniation within the encephalocele. Two patients died of sepsis (patients No. 4 & 6 in Table 1) while two lost follow-up (patients No. 1 & 3 in Table 1). Eight patients were contacted, interviewed and/or examined in the outpatient clinic for developmental assessment. Four of them had normal mental and physical development for age (none had hydrocephalus or large bony defect but two had minimal protrusion of brain tissue). The rest were developmentally delayed with ranging severity from mild to moderate. All of them had protruding brain tissue and hydrocephalus and all were shunted.

Discussion

Encephalocele is a neural tube defect occurring as a result of failure of fusion of the rostral end of the neural tube, during the fourth week of fetal life. If arrest of neural fusion occurs in the caudal region at the same time, spina bifida may be present in association with encephalocele[1].

The incidence of encephalocele has been estimated as one in every 3,000 - 10,000 births[2], *e.g.*, 0.2/1,000 births in New South Wales[3] and 0.5/1,000 births in Nigeria[4]

To the authors' knowledge, there was no reported incidence for encephalocele in Saudi Arabia. This study found it to be 0.28 in 1,000 births in KAUH as we had 10 cases among 35,851 births.

No sex preponderance was demonstrated in this present study, confirming the same finding in other reports[4, 5]. In contrast to female preponderance in some studies[2, 3]. This lesion can be classified based on location into occipital frontonasal, parietal and nasopharyngeal. In reviewing previous reports, it was found that the most common location was the occipital one, which occurred similarly in all our patients.

Antenatal ultrasound can confirm diagnosis of encephalocele, which has to be differentiated from cystic hygroma, teratoma, and haemangioma[6, 7]. Six of the present study's patients had antenatal ultrasound examinations. Three of them were not diagnosed because of the small size of the lesion. This is supported by the results of other workers[8].

Encephalocele may occur as an isolated lesion or as a component of various syndromes, or associated with other congenital abnormalities. In the investigation series, one case had Meckel-Gruber syndrome which is an autosomal recessive disease. It was described by Meckel in 1822, later by Gruber and, recently, by Optiz and Howe. More than 200 cases have been reported till 1997[9]. The patient died at 3 months of age. The other case was Patau syndrome (trisomy 13) and this is, to the authors' knowledge, the first reported association of encephalocele with Patau syndrome.

All patients of this study had brain ultrasound and/or CT scan done to exclude any other associated intracranial malformation, *e.g.,* hydrocephalus, ventriculomegaly, brain atrophy, *etc.*

The authors found that all patients of this present study had either hydrocephalus or ventriculomegaly on investigation, but no apparent clinical neurological abnormalities were detected at birth. Hydrocephalus was found to develop with encephalocele in 62% of our cases. This is a much higher association than most of the other reports[4, 10]. Only one of them had normal mental development, the child now is 13 years old, but his hydrocephalus had arrested spontaneously at an early age. One of the study's patients who developed hydrocephalus had a lost follow-up at 3 months of age. From the six hydrocephalics patients left, two died at 3 and 15 months of age. The four surviving hydrocephalics were shunted but all of them had delayed milestones. It is clear to the investigators that the development of hydrocephalus in encephalocele patients is one of the poor prognostic factors as it is associated with either delayed milestones or death irrespective of shunt insertion. This result is consistent with the study of Mealy [2].

Development of blindness was found to be associated with hydrocephalus in some cases and associated with large encephalocele in others. Mealy *et al*[2] reported that the timing of operation had no clearly discernible influence on the ultimate outcome.

The result of this study was consistent with his, as no significant difference was observed between those having early versus late surgery when looking into milestones, death, blindness, and development of hydrocephalus. In general, the recommenation by the authors of early surgical interference especially if it is ruptured or discharging CSF.

The mode of delivery or head circumference at birth did not affect the outcome in the present study's cases from mortality or morbidity viewpoints.

It is clear that associated syndromes, the size of the bony defect and neural tissue contents of the encephalocele influenced the prognosis in this present study. The investigators found all patients with small defect in absence of hydrocephalus or associated syndromes had normal development. The four normal cases had small bony defect in comparison with those with delayed milestones or deceased patients. This result confirms other workers[2] who showed poor results with larger lesions.

In conclusion, the large size of the bone defect, the major neural tissue content of the sac, the associated hydrocephalus and syndromes are important factors that influence adversely the prognosis of occipital encephalocele.

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العوامل المُنذرة بقيلة الدماغ ، مسح شامل لمدة أربع عشرة سنة ، أُجري بمستشفى جامعة المللك عبد العزيز- بجدة

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المستخلص. تصف هـذه الدراسة متابعة لمدة أربع عشرة سنة للرضـع المصابـين بالقيـلة الدمـاغي الذين شـوهـدوا في مسـتشـفى جـامـعـة الملك عبـدالـعزيز في الفـتـرة من عـام ١٩٨٨م إلى عـام ٢٠٠٢م . من بين الثـلاثة عـشـر مـريضـاً لم يولد (بين الـ ٣٥٨٥١ ولادة مسـجلة في تلك الفترة) سـوى عشرة فقط . لم يصـادف سوي النميط القذالي . ومن بين الثلاث عـشـرة مـرضى كـان ثلاثة مصـابون بـالقيلة السحـائيـة ، وتسـعـة مصـابون بالقيلة السحـا ئية الدماغيـة وواحد فقط مصـاباً بالقيلة السحـائية الدماغية الكيسية . أصيب ثمانية مـرضى بموة الرأس ولـم يحتج إلى غرز تحويلية بطينية صفاقيـة سوى خمسة من هؤلاء المرضى أحدهم لم يخضع لتصليح القيلة الدماغية، شخصت لدى مريض واحد متلازمة باتاو مع زرق خلقي وضخـامة البطينات ، كـان مريض واحد مـصاباً بمتلازمـة ميكل - غروبر تم الاتصال بسبعة مرضى ومقابلتهم و / أو فحصهم في العيادة من أجل تقـيـيـم نموهم فكان النمـو الجسـدي والنفـسي (العـقلي) لدي أربعـة منهم طبـيعـياً بالنسبـة إلى أعـمارهم وأمـا الثلاثة الآخـرون فكانوا متأخـرين نمواً . كـانت غايتنا دراسـة المتثابتات الإنذارية المحتملة باحثين عن عـوامل الاختطارالأكثر تأثيـراً في معدل وفيات ومرضية مرضـانا . استنتجنا أن حـجم عيب العظم ، والمحتوى الدمـاغي للكيس وموة الرأس والمتلازمات المرافقة عوامل هامة تؤثر في إنـذار القيلة الدماغية القذالية من كلتا ناحيتي معدل الوفيات والمرضية .