

CASE REPORT

Discordant Encephalocele in Monozygotic Twins in Kaduna, Nigeria: A Case Report

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ABSTRACT

Encephaloceles are a rare central nervous system congenital anomaly, and discordant encephalocele in monozygotic twins is very rarely reported in the literature. This is the first report of discordant encephalocele in monozygotic twins from our centre. We present the case of a 2-day old low birth weight male monozygotic first twin with occipital encephalocele, microcephaly and a right sided clonic seizure, which was diagnosed prenatally by ultrasound scan. He had no other associated congenital anomaly and the second twin was phenotypically normal. Computerised tomographic scan showed skull defect with herniation of brain tissue. He was worked up for surgery, had excision and repair under general anaesthesia, made a satisfactory post-operative recovery, and was discharged on an anticonvulsant. However, there was evidence of delayed developmental milestones compared to the other twin by 18 months of age. This case suggests that there may be other non-genetic and non-systemic factors that could be responsible for the development of this congenital anomaly, encephalocele.

Keywords: Multifactorial, neural tissue, sporadic dysraphic malformation

INTRODUCTION

Encephaloceles are an uncommon sporadic dysraphic malformation, with a prevalence of 1 in 2,000–5,000 live births.¹ The aetiology is multifactorial with environmental factors such as folic acid and vitamin-A being implicated.¹⁻³ Syndromic forms and encephalocele associations represent approximately one-third of the cases.^{2,3} Some, such as Meckel syndrome, Knobloch syndrome, and cryptophthalmos syndrome, among others, are monogenic while others are associational.^{2,3} Encephaloceles may be

concordant or discordant in twins.^{1,4} Few cases of discordant encephalocele have been reported in literature.^{1,5} We report a case of monozygotic twins, discordant for occipital encephalocele.

CASE REPORT

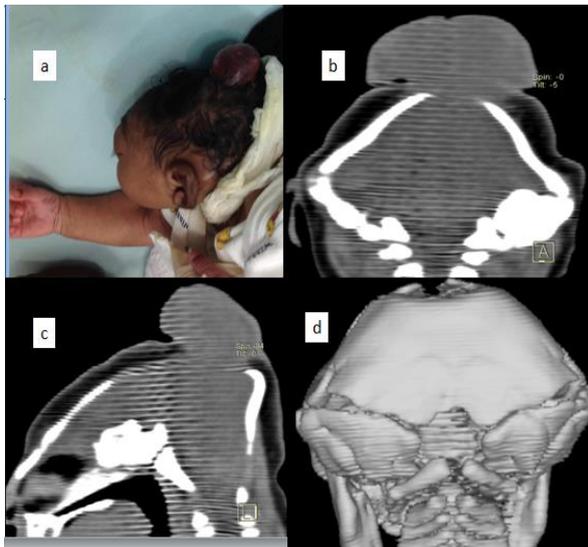
This was the case of a 2-day old male monozygotic twin delivered via a planned caesarean section in a hospital in Kaduna, Nigeria. The elective caesarean section was as a result of an encephalocele diagnosed during a pre-natal ultrasound scanning. He was

noticed to have a swelling at the occipital region at delivery. There was associated right sided clonic convulsion. The review of other systems was essentially normal.

Physical examination revealed a 4cm by 3cm by 2cm swelling at the occipital region. (Figure 1a) The occipito-frontal circumference was 28cm which was small for his age (33-37cm) and he was also found to be small for his gestational age. (Birth weight was 1.8kg). He had no other associated congenital anomalies. His other twin was phenotypically normal.

Computerised tomographic (CT) scan showed an occipital encephalocele with extrusion of intracranial tissue (Figures 1 b-d).

Figures 1a-d. Showing the encephalocele (a) and coronal (b) sagittal (c) and (d) 3D-reconstructed computerised tomographic (CT) scan images of the patient



He was worked up for surgery and had emergency excision and repair under general anaesthesia. Intra-operatively, neural tissue was found in the sac. He made an uneventful recovery with no immediate post-operative complications and was subsequently discharged on carbamazepine. He remained seizure free on the anti-convulsant, 18-months after surgery; however, the patient has delayed developmental milestones compared to the other twin.

DISCUSSION

The diagnosis of occipital encephalocele was made pre-natally in our patient using an ultrasonographic scan, and delivery was done by caesarean section. This was consistent with the current practice of diagnosis with foetal ultrasonography, which is considered as the mainstay of foetal imaging, even though foetal magnetic resonant imaging may provide superior details of central nervous system anomalies.⁶⁻⁸

Our patient presented with a seizure disorder and microcephaly. These findings may be seen in patients with occipital encephalocele although, hydrocephalus rather than microcephaly, may occur in some patients with occipital encephalocele.⁶

Computerised tomographic scan was used in the postnatal evaluation of our patient since it provides an excellent depiction of the bony defect, particularly with coronal, sagittal and 3-dimensional (3D) reconstruction. Nonetheless, postnatal MRI is the diagnostic study of choice for the classification of this disorder and assessment of the extent of neural tissue herniation and hydrocephalus.^{6,7} Magnetic resonance venography and angiography may be useful in delineating vascular relationships to the lesion.⁶ This was not done in our patient due to the non-availability of the facility in our centre.

Our patient still has delayed developmental milestones compared to the other twin, and this may not be unconnected with the microcephaly, seizure disorder and presence of neural tissue within the sac. These factors are known to cause poor outcome in patients with encephalocele.⁶

In our patient the encephalocele was discordant. This suggests that isolated encephaloceles may result from a non-genetic developmental disruption as earlier reported.¹ Some authors have suggested genetic factors, such as consanguinity, differences in ethnic prevalence and higher twin concordance.⁹⁻¹¹ It is, also, unlikely that systemic factors, such as deficiency of folic acid, played a major role in our case because

both twins were exposed to the same intrauterine environment. Also, since our patient is a monozygotic twin, it may be unlikely that genetic factors played a role. This case may suggest that isolated encephaloceles may result from a non-genetic developmental disruption. This mechanism has been suggested in an earlier report.¹

Thus, there is a need for more studies to elucidate those factors responsible for discordant encephalocele in monozygotic twin.

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The main limitation of our study was the inability to carry out a DNA study to confirm the monozygosity of our patient due to the non-availability of such facility in our centre.

CONCLUSION

We conclude that other unreported factors may contribute to the development of encephalocele, and that encephaloceles which are associated with seizure and neural tissue in the sac may lead to delayed developmental milestones.