Case Report

Encephalocele in Resource Poor Setting: The Case of AK

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Abstract

We report a full term infant with features of encephalocele and amniotic band sequence identified at time of birth and presenting with infection. The case highlights a unique presentation of encephalocele, as well as, the significant difficulties of caring for severe congenital malformations in developing resource-poor countries.

INTRODUCTION

Encephaloceles are a rare congenital malformation, which is characterized by herniation of the brain and the meninges through a defect in the skull. Although they have traditionally been classified as a type of neural tube defect and therefore related to anencephaly and spina bifida, recent research has shown that it is distinct in its response to folic acid supplementation as well as associations with other birth defects [1]. Encephaloceles have shown unique patterns of presentation with clear geographic and ethnic patterns. Despite continued advances in the diagnosis and care severe congenital malformations in resource-rich countries, the care of these conditions continues to face significant challenges and poor outcomes in resource-poor countries, as highlighted by this case.

CASE PRESENTATION

AK is a 2 day old male infant that presented to a hospital in Gulu, Uganda with a 1 day history of fever and pustular drainage from anencephalocele. At presentation he was febrile with a temperature of 39.9°C, heart rate 162 beats per minute and pulse oximetry of 99% in both upper and lower extremities on room air. He was noted to have multiple congenital malformations – most notably a large frontal encephalocele measuring 5.5x5cm with yellow discoloration, absence of overlying dermal tissue and large underlying calvarial defect. The presence of neural elements was identified with transillumination. Other abnormalities included bilateral exophthalmos, syndactyly of his right fourth and fifth digits with an amniotic band of the right proximal third digit and bilateral calcaneovalgus deformity. There was no cleft lip or palate. His lungs were clear to auscultation, with no respiratory distress. The cardiovascular exam was normal, with no murmur and 2+ symmetric pulses. The abdomen was soft, nontender, with no appreciated organomegaly or masses. His testes were bilaterally descended with normal male genitalia. There was no spinal abnormality, including absence of any midline hair tufts or sacral dimples. AK had normal tone in all extremities, with normal Moro, grasp and suck reflexes (Figure 1).

A complete blood count obtained at the time of presentation demonstrated a white blood count of 5.92, hemoglobin of 15.8, hematocrit of 42.4 and platelet count of 261. A rapid diagnostic test for malaria at that time was negative.

AK’s was immediately started on IV ceftriaxone. A sterile dressing of gauze and normal saline was applied. He was noted to breastfeed without difficulty. The fever improved within 24 hours of antibiotics. Both a head ultrasound and echocardiogram were requested, but were unable to be completed as the machine was broken at the time. AK died on the third day of hospitalization.

Figure 1 Post-mortem image of AK.
AK was born at an estimated 38 weeks of gestation by normal spontaneous vaginal delivery in cephalic presentation to a 22 year-old gravida 3 now para 3. The labor course was uncomplicated; however it was noted by the provider that an ill-defined mass was palpated on cervical examination at 6cm of dilation. There was no further documentation of this finding. He weighed 4kg at birth with spontaneous immediate cry and Apgar scores of 9 at one minute and 10 at five minutes. He was noted at this time to have multiple abnormalities, which were documented as “a big round mass on the head”, conjoined fingers, and nasal abnormalities. He was discharged on postpartum day 1 with a referral for further evaluation and management.

The pregnancy was noted to have minimal complications. The mother attended the recommended 4 antenatal visits and received the scheduled malaria prophylaxis with fansidar and mebendazole as per guidelines. She however, did not begin Folic Acid supplementation until 36 weeks of gestation. The pregnancy was complicated by the use of hormonal contraception at the time of conception – she received one dose of intramuscular depotmedroxyprogesterone acetate 1 month prior to conception. Her pregnancy was also complicated by malaria in the first trimester and she was successfully treated with quinine.

This is the third born child to the 22 year-old mother and 26 year-old father, who is peasant farmers. Both parents are known to have sickle cell trait, with the oldest child, a 6 year-old female, having sickle cell disease. The second child is a 3 year-old male with no known medical problems. There is no family history of congenital malformations or specifically neural tube defects.

DISCUSSION

This case highlights several variations on previously published data regarding the epidemiology of encephalocele. Unlike in the case of AK, both frontal encephaloceles and encephaloceles associated with amniotic bands are not typically located along the midline and have a very high association with facial clefts [2]. Additionally the calcaneovalgus deformity seen in AK is traditionally associated with intrauterine growth restriction, whereas the talipes equinovarus deformity is more commonly associated with the presence of encephalocele.

This inconsistent presentation of anencephalocele makes AK unique. It may suggest that the abnormality is most consistent with disorganization (Ds) and amniotic band sequence, similar to the cases described by Purandare et al. [3], rather than either encephalocele or pure amniotic band sequence. However, unlike AK, all of the cases described by Purandare were diagnosed prenatally, 2 were delivered by c-section, 1 was delivered by induction of labor and 1 was a spontaneous intruterine fetal demise. All 3 viable infantswere described to have low Apgar scores at the time of delivery and an associated cleft lip or palate.

Previous research on the epidemiology of encephaloceles in Africa has suggested distinct anatomical patterns, and ethnic variation in lesion location, with occipital lesions more prevalent among individuals of Bantu origin and occipital and sincipital lesions roughly equal in distribution among the Nilote [4]. AK was identified as being of the Acholi tribe, which is of Nilote origin. This study failed to identify any relationship between seasons of birth and the number or type of lesions, although clear relationships exist between significant peri-conception maternal stress and the prevalence of neural tube defects. This may have been a factor in the case of AK, as he was likely conceived in early December 2014, which is traditionally among the driest months of the year.

The mode of delivery for encephaloceles is controversial without clear evidence supporting or rejecting a vaginal delivery. Outcomes can only be extrapolated from a study by Luthy et al, which identified a decrease in motor function in neonates with meningomyelocele were delivered via caesarean section compared to vaginal delivery [5]. AK was not diagnosed prenatally, as routine ultrasound is not performed in standard antenatal care in Uganda. The lack of prenatal diagnosis prevented the ability to establish a delivery plan. AK, weighing 4 kg, surprisingly survived a vaginal delivery without complications.

Additionally the case of AK, while unfortunate in its outcome, also brings attention to many of the challenges of dealing with complex congenital malformations in resource-limited settings. Lack of prenatal ultrasound did not allow for early diagnosis of AK’s multiple congenital anomalies or early referral. There was more than a 24 hour delay between when AK was discharged from the health center in which he was born and his arrival at the hospital, which delayed care and increased his risk of infection. Limited availability of investigations, including additional laboratory tests, cultures and imaging hinder a complete evaluation of AK’s condition.

REFERENCES