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CASE REPORT

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Frontonasal encephalocele with bilateral congenital microblepharon: A case report

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Abstract: : A six-day old term neonate was born with a frontonasal ulcerated discharging mass with purulent fluid but no cerebrospinal fluid. The left eye socket was empty and there was bilateral microblepharon. Computerized tomography scan showed irregular shaped soft tissue mass, the same density as the brain tissue, continuous with the frontal lobe and associated defect of frontal bone/nasium. The mass displaced the left globe inferiorly but there was a demarcation between the globe and the mass. Ventricular systems were grossly dilated and distorted (lateral and 3rd ventricles). However, the 4th ventricle was normal.We present a patient with an unusual constellation of clinical and radiologic findings that have not been, hitherto described.

Key words: Frontonasal, encephalocele, neural tube defect, midline defects, microblepharon.

Introduction

The development of the frontonasal region is complex. Aberrant embryogenesis leads to three main types of anomalies: nasal dermal sinus, anterior encephalocele and nasal glioma. Knowledge of the developmental anatomy of the anterior neuropore and postnatal maturation will assist the radiologist well when it comes to imaging frontonasal masses.[1,2] Nasofrontal midline masses are uncommon, with an incidence of one in 20,000 to 40,000 live births in America.[3] These masses originate from the nervous system as a result of embryologic maldevelopment and frequently have intracranial connections. Although they may be adjacent to the midline, they are still referred to as midline naso frontal masses. Correct and prompt diagnosis is important in order to prevent complications and provide accurate long-term prognosis. To the best of our knowledge the association of fronto nasal encephalocele, holoprosencephaly and microblepharon have not been reported in the literatures.

We report one case of frontonasal encephalocele with background holoprosencephaly, bilateral microblepharon and anophthalmia which presented with radiological diagnostic dilemma.

Case Report

A six- day old male term neonate with a weight of 2100g delivered by a 20-year-old healthy Nigerian Para 2^{+0} 2 alive. She received no antenatal care and no history suggestive of abnormal liquor volume was established. However, the mother denied using herbal medication. There was no history of vaginal discharge. The mother did not have any febrile illness or rash during pregnancy. Delivery at home was spontaneous vaginal. Membrane ruptured one hour prior to delivery. The baby was on breast milk only. The baby was the second of two children in the family. Mother was a 20-year old junior secondary school certificate of education holder and father a 23-year old farmer with senior secondary school certificatewith no history suggestive of consanguineous marriage. There was no history of illicit drug use by the mother or the father.

There was no history of miscarriages, congenital anomalies or neurologic defects in the family. Physical examination revealed a mass at the base of the nasal bridge measuring 5cm by 5cm, non-tender, freely mobile with excoriation on the surface, areas of necrosis and ulceration, discharging purulent and serous fluid. The left eye orbit was empty, with bilateral microblepharon. (Fig 1).

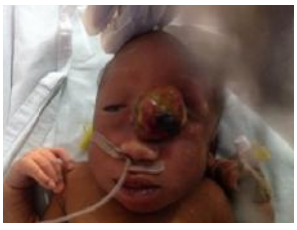

Fig 1: Showing the frontonasal protrusion on admission

Fig 2: Showing the Patient after surgery

There was no exophthalmos or hypertelorism. The right eye reacts to light and cornea was clear. Anterior chamber had normal depth and pupil was reactive. There were no other malformations or abnormal findings. Systemic examination was normal.

The cranial computerized tomography (CT Scan) (Fig 3 and 4) showed a frontonasal protrusion with grossly dilated and distorted lateral and 3rd ventricles. Normal cerebral architectural differentiation into lobes was not distinctly discernable. Patient had craniotomy, excision, duroplasty and repair on the 11th day on admission (figure 2). Our patient had no intraoperative haemorrhage and recovered appreciably from anaesthesia. However, the child died twenty-five hours after surgery. No autopsy was obtained and cause of death could not be determined.

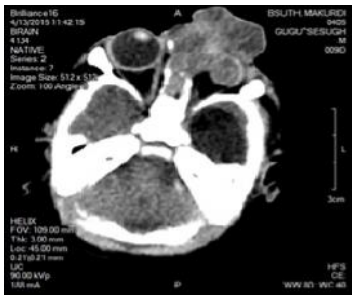

Fig 3: Cranial CT scan showing frontonasal protrusion with associated defect of frontal bone

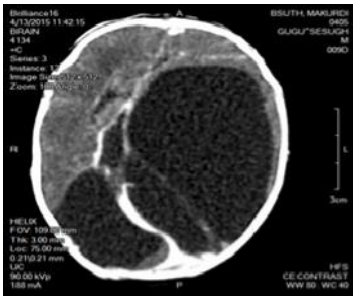

Fig 4: Cranial CT scan showing dilated ventricular systems and distorted lateral and 3rd ventricles), the 4th ventricle is normal

Discussion

The occurrence of a frontonasal encephalocele, bilateral microblepharon, anophthalmia and holoprosencephaly is rare and have not been described in the literature, to our knowledge. Various syndromes have been described, including the Barber Say syndrome, Ablepharon-Macrostomia,but none involving intracranial anomalies, suggesting that the spectrum presented by our patient may represent an association or syndrome yet to be elucidated. Genetic studies would have been very helpful in this regard,but our capacity in this regard is limited.

A frontonasal mass in a newborn infant may present a difficult diagnostic problem.[2,3] Clinical diagnosis of presentation of a frontonasal mass does not constitute a dilemma but when it occurs in the setting of other multiple anomalies, then, it becomes a problem, as in our index case which did not fit into any known syndrome or association. Further more describing the Computerized Tomographic images and making a distinct diagnosis was equally difficult radiologically (compare figures 3 and 4). A correct diagnosis is important in order to formulate an accurate prognosis and consider appropriate surgery. Nasofrontal encephaloceles are more common in Thailand, Malaysia, and Indonesia than in Europe or the United states.[2,3] In series reported from Africa, occipital encephaloceles are more common than frontal encephaloceles, except in South Africa where they enjoy equal distribution.[4] Congenital midline masses, particularly encephaloceles, may lead to cosmetic or infectious complications because of intracranial connections. Most complications can be avoided if lesions are treated successfully by early surgery.[2,5] In cases of encephaloceles, early excision is imperative to promote normal facial growth. The neurodevelopmental outcome after resection is generally good. Hypertelorism was not present in our patient; this is at variance with the finding by Obande et al where their patient had pancraniosynostosis and phenotypic syndromic facies.[6]

There are known genetic and environmental factors associated with their geographic occurrence of encephalocele, such as maternal diabetes mellitus, alcohol use as well as use of teratogenic retinoic acid.[3] Genetic studies were not conducted in our patient due to lack of facilities. There are reports of association of encephalocele with trisomies.[7,8] Sometimes there may be associated cerebrospinal fluid (CSF) rhinorrhea, though this was not present in our patient.

Surgery was performed where the patient had craniotomy, excision, duroplasty and repair.[9]These procedure corrected the frontonasal encephalocele. Our patient had extensive procedure, good anaesthesia, and adequate surgical haemostasis but the procedure was not well tolerated by the neonate. The patient had three apneic attacks after surgery and never really recovered from the effect of surgery and anaesthesia. He eventually died 25 hours after surgery.

The mass was already infected (fig 1) when the patient presented at the hospital. The outcome of the patient may have been adversely affected by delayed presentation and poverty. The outcome of cases like this can be improved with universal access to the National Health Insurance schemes which makes health care affordable. MRI and genetic studies would have assisted with further evaluation of the patient.[10]

Conclusion

A frontonasal mass in a new born may present a difficult diagnostic problem because these are rare anomalies. Our patient presented with an unusual constellation of clinical and radiologic findings that have not been, hitherto described in association. The relative significance of these impressive compromise of neuro-ectodermal features is unknown to us at the moment.

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