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Case Study

Magnetic resonance imaging of bilateral congenital anophthalmia: A case report

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The spectrum of ocular developmental disorders includes a wide range of malformations, depending on the embryonic age of commencement of the disease. One example of these malformations is Anophthalmia. It is an uncommon congenital anomaly associated with problems of social integration especially in developing climes. Magnetic resonance imaging (MRI) plays important role in the diagnosis and differentiation of these malformations. We report a case of a child born with congenital anophthalmia to describe MRI features of this rare ocular anomaly.

Key words: Anophthalmia, congenital, magnetic resonance imaging.

INTRODUCTION

The spectrum of ocular developmental disorders includes a wide range of malformations, depending on the embryonic age of commencement of the disease (Llorente-González et al., 2011). One example of these malformations is Anophthalmia, which in medical practice, is an all-encompassing term used to describe the clinical and radiologic absence of a globe in the presence of ocular adnexa (eyelids, conjunctiva, and lachrymal apparatus) (Duke-Elder, 1964; O'Keefe et al., 1987; Dantas et al., 2002). Anophthalmia is closely related to microphthalmia and may be difficult to differentiate them. Microphthalmia is described as a globe with a total axial length that is at least two standard deviations below the mean for age (Verma and Fitzpatrick, 2007).

Anophthalmia is an uncommon malformation; hence there is paucity of reported cases globally. Nevertheless, there is a documented incidence of 3 per 1000 live births (Guthoff et al., 2004; Morrison et al., 2002; Campbell et al., 2002). Chuka-Okosa et al. (2005) in a study, conducted in Enugu Southeastern Nigeria, reported anophthalmia to represent 9.3% of congenital eye anomalies seen over an 8 year period. Ukponmwan (1999) in Benin City, Midwestern Nigeria, reported only two cases seen over a 20 year period. Congenital anophthalmia can be isolated or in one third of cases (Verma and Fitzpatrick, 2007; Speeg-Schatz et al., 1997) may be associated with other syndromes such as trisomy 13, trisomy 18 (Nagarajan et al., 2018), Lenz syndrome (Nyberg et al., 2003), Goldenhar-Gorlin syndrome (Nyberg et al., 2003), Warrensburg syndrome (Sener, 1998).

Once the diagnosis has been established, systematic examination with both ocular and systemic imaging tests (ultrasonography, computed tomography, MRI) should be performed to rule out additional neurological, renal, cardiac

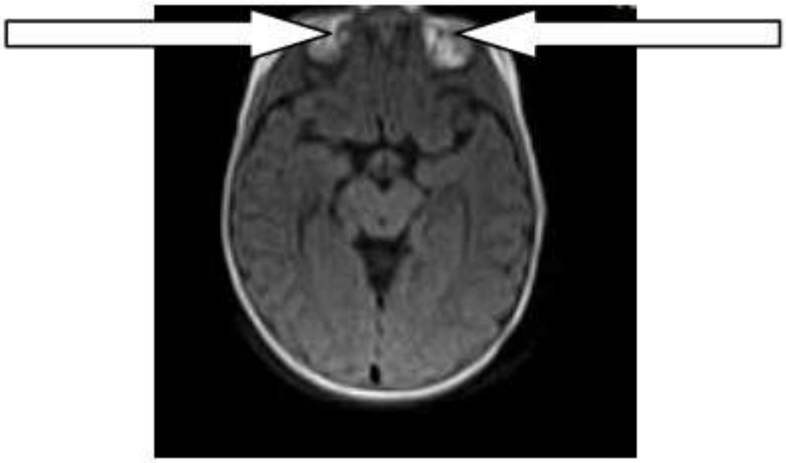

Figure 1. Axial T1-W MR image shows absent globes bilaterally with small amount of amorphous intra-orbital soft tissue and reduced orbital size(arrows). Brain is normal.

or other associations (Mafee, 2005). MRI is the imaging modality of choice for the diagnosis and characterization of anophthalmia. It provides excellent soft tissue contrast of the orbit and brain at the same time to exclude associated intracranial abnormalities. Apart from MRI, Computed tomography (CT) scan and ultrasound scan can also be used for the diagnosis of congenital anophthalmia.

Abnormal development of the eyes has numerous implications including aesthetics, social and psychosocial challenges for the child especially in developing climes where interventions that promote social integration and wellbeing may be lacking. To the best of our knowledge, this is the first bilateral anophthalmia case reported in Port Harcourt City, South-South, Nigeria.

CASE REPORT

Brain MRI scans were performed using 0.35 Tesla General Electric machine with standard protocols and parameters (slice thickness: 3mm, interval: 3mm, flip angle: 90^{o}, FOV: 18cm, TR: 424.3ms, TE: 15.7ms, Matrix size: 256 x 256 and NEX: 2) on a one month old female child who was referred for investigation on account of missing eyes. T1-weighted images (WI), T2-WI and post-contrast T1-WI were obtained at axial, sagittal and coronal planes. MRI scans show absent globes bilaterally with disorganized residual intra-orbital soft tissue and reduced orbital size. The cerebrum and cerebellum are morphologically normal. These findings are in keeping with Anophthalmia.

DISCUSSION

Diagnosis is mainly clinical and usually confirmed by neuroradiological imaging. In this patient under review, neuroimaging revealed bilateral absence of the globe with hypoplastic orbits and poorly developed orbital tissues. These findings are in keeping with findings of similar studies conducted by Albernaz et al. (1997); Kouassi et al. (2006); Aliyu et al. (2015); Okeigbemen and Dawodu (2014); Diomande et al. (2017); Trivedi and Venkatesh (2009) and Celebi and Sasani (2014). In (Albernaz et al., 1997) study, reported imaging findings of a clinical anophthalmia case series with bilateral anophthalmia, in which there were a variety of intraorbital, intracranial and craniofacial anomalies. (Kouassi et al. (2006) study, reported a case of bilateral congenital anophthalmia in association with the syndrome of Patau. In (Aliyu et al., 2015), study conducted in Kano Northern Nigeria, also reported bilateral congenital anophthalmia. In Benin City Midwestern Nigeria, Okeigbemen and Davodu (2014) study, equally reported bilateral anophthalmia case. Diomande et al. (2017), reported three cases of isolated bilateral hereditary congenital anophthalmia in the same family with no abnormality on general physical examination. Similarly, Trivedi and Venkatesh (2009) also reported a case of anophthalmia with bilateral microphthalmia without any systemic abnormality. Celebi and Sasani (2014) also reported two cases of isolated bilateral congenital anophthalmia in the same family.

In this patient under review, anophthalmia manifested as absence of globes with presence of amorphous tissues contained within small bony orbits. This finding is similar to the finding of a related study conducted by Albernaz et al. (1997). In their study, they reported that in one case, orbits contained amorphous tissues of intermediate T1 signal intensity and low T2 signal intensity on MR images. All the involved orbits were considered to be small in both longitudinal and lateral dimensions.

In anophthalmia vision cannot be restored. Surgery is

mainly cosmetic. In developing climes like ours, this type of heavy and sometimes disappointing treatment is difficult to achieve by sometimes untrained teams. Moreover, the level of poverty of the parents limits their accessibility to the treatment, with aesthetic and social consequences.

CONCLUSION AND RECOMMENDATIONS

Anophthalmia is an uncommon form of congenitalocular malformation. The consequence of this anomaly in the developing world is devastating. The discovery of a congenital anophthalmia calls for a comprehensive review in order to seek the etiology to prevent recurrence. Proper patient's care and their families by a multidisciplinary team will contribute to the total wellbeing and social integration of these patients.

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