This report presents a rare example of a bilateral congenital anophthalmos and an agenesis of the optic pathways. The MR imaging studies revealed that the eyeballs, optic nerves, optic chiasm, optic tracts and optic radiation were absent. The chromosomal examination was normal. Mild mental retardation was also observed. Apart from the rarity of the anophthalmos and the total absence of the optic pathways, no etiologic reason for this pathology could be detected, which makes this case more significant.

Key Words: Bilateral anophthalmos, optic pathways, agenesis, MRI

INTRODUCTION

A maldevelopment of forebrain structures is known to be a result of insults occurring during the first few weeks of gestation. Anophthalmos, which is an absence of the eye, is an example of this maldevelopment that can also be found with associated intracranial anomalies, namely the absences of the optic nerves, optic chiasm, optic tracts, optic radiations and anomalous corpus callosum. The insults that cause this maldevelopment can be summarized as trauma, congenital infections, heredity and several syndromes. Here we present a case of congenital anophthalmos and agenesis of the optic pathways with no convincing etiologic reason.

CASE REPORT

The patient is a 24-year old female who had bilateral congenital anophthalmos. Her family has three girls and the patient is the oldest. She was born after an uneventful pregnancy. Her parents were healthy and showed no consanguinity.

The ophthalmologic examination after birth showed an absence of the eyeballs. She was accepted as an isolated congenital anophthalmos case. Since then, she has had no radiological examinations, especially a CT or an MRI procedure, which would have allowed the pathologies to be observed within the brain.

The family history was unremarkable. The chromosomal examination was normal. The other two sisters of the family were completely normal.

In the current ophthalmologic examination after 24 years, the patient had hypoplastic orbits and well formed but rudimentary eyelids and brows. She had a bilateral anophthalmos.

MR imaging studies revealed that the eye bulbs were absent and replaced by residual soft tissue (5 mm in diameter) which was isointense with muscle in the T1-weighted sequences (Fig. 1 and 2). The extraocular muscles were inserted to the residual bulb and were medially displaced (Fig. 1). The optic nerves, the optic chiasm, the optic tracts and the optic radiations were all absent (Fig. 1-4). The lateral ventricles were enlarged on both sides (Fig. 2). The corpus callosum was present and was slightly diminished in size (Fig. 3).

The patient also had mild mental retardation.
Eye development is first evident at the beginning of the fourth week. The optic grooves appear in the neural folds at the cranial end of the embryo. As the neural folds fuse to form the forebrain, the optic grooves evaginate to form the optic vesicles, which project from the wall of the forebrain into the adjacent mesenchyme. As the optic vesicles grow, their distal ends expand and their connections with the forebrain constrict to form optic stalks, which are the precursors of the optic nerves. The optic nerve is formed by more than a million nerve fibers that grow into the brain proceeding centrally to decussate through the primitive chiasmal commissura. A deficiency in the proper formation of the connections between the globes and the optic pathways results in hypoplasia or an absence of related white matter tracts.

Fig. 1. Axial T1-weighted MR image shows bilateral fibrotic eye bulbs in both orbits (arrow). The rudimental medial and lateral recti are medially displaced (arrow head). Minimal enlargement of the lateral ventricles but no hypotense signal changes of the optic radiation can be observed.

Fig. 2. Inversion recovery sequence shows the enlarged ventricles and an absence of the optic radiations (arrow).

Fig. 3. Midsagittal T1-weighted MR image shows an absence of the optic chiasm (arrow) and a normal corpus callosum.

Fig. 4. Coronal T1-weighted MR image shows an absence of the optic chiasm (arrow).
An interruption of embryogenesis within the first few weeks of gestation is the main cause of the primary form of congenital anophthalmos, which encompasses the aplasia of the eyeballs and the absence of the optic pathway. Various factors have been reported to be the cause of this extremely rare anomaly. Among those trauma, congenital infections, heredity, and several syndromes have been reported. Anomalous corpus callosum, neurocutaneous disorders (incontinence pigmenti) and, particularly in unilateral ones, some craniofacial syndromes may accompany the congenital anophthalmos. However, a case of a total absence of the optic pathways was also reported without any accompanying pathology.

Diagnostic amniocentesis has been reported to be a factor responsible for the congenital anophthalmos without the absence of optic pathways. In our case, there is no possibility that the anophthalmos resulted from a diagnostic amniocentesis.

Heredity has been showed to be a cause of anophthalmos. Most anophthalmos cases are sporadic and probably have a multifactorial etiology. Nevertheless, some authors have reported an autosomal or X-linked recessive inheritance. Brunquell, et al. reported a case of an X-linked anophthalmos. The patient was a 27-year-old man with severe developmental defects and accompanying pathologies such as seizures, small head, clubbed feet, and locomotor deficiencies. He also had a maternal male cousin with unilateral anophthalmos and contralateral microphthalmia. On the other hand, Sensi et al. reported a family with congenital anophthalmos with a dominant pattern of inheritance. Two children of the family were affected with anophthalmos, of whom only one was bilateral. That case differed from the others by an absence of accompanying anomalies as mental retardation, congenital malformations and other ocular anomalies. Our case did not have any congenital malformations except for mild mental retardation but had a total absence of the optic pathway. In addition, the family history showed that there was no other member with anophthalmos.

REFERENCES

1. BenEzra D, Sela M, Peer J. Bilateral anophthalmia and...
unilateral microphthalmia in two siblings. Ophthalmologica 1989;198:140-4.
2. Albernaz SA, Castillo M, Hudgins PA, Mukherji SK. Imaging findings in patients with clinical anophthalmos. AJNR Am J Neuroradiol 1997;18:112-3.
3. Brunquell PJ, Papale JH, Horton JC, Williams RS, Zgrabik MJ, Albert DM, et al. Sex-linked hereditary bilateral anophthalmos. Arch Ophthalmol 1984;102:108-13.
4. Marcus DM, Shore JW, Albert DM. Anophthalmia in the focal dermal hypoplasia syndrome. Arch Ophthalmol 1990;108:96-100.
5. Sener RN. Cranial MR imaging findings in Waardenburg syndrome: anophthalmia, and hypothalamic hamartoma. Comput Med Imaging Graph 1998;22:409-11.
6. Moore KL, Persaud TVN. The developing human. 6th ed. Philadelphia: W.B. Saunders Company; 1998.
7. Sensi A, Incorvaia C, Sebastiani A, Calzolari E. Clinical anophthalmos in a family. Clin Genet 1987;32:156-9.
8. Daxecker F, Felber S. Magnetic resonance imaging features of congenital anophthalmia. Ophthalmologica 1993;206:139-42.
9. Scott IU, Warman R, Altman N. Bilateral aplasia of the optic nerves, chiasm, and tracts in an otherwise healthy infant. Am J Ophthalmol 1997;124:409-10.
10. Waheed K, Jan W, Calver DM. Aplasia of the optic chiasm and tracts with unifocal polymicrogyria in an otherwise healthy infant. J Pediatr Ophthalmol Strabismus 2002;39:187-9.
11. Pearce WG, Nigam S, Rootman J. Primary anophthalmos. Histological and genetic features. Can J Ophthalmol 1974;9:141-5.