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BASAL ENCEPHALOCELE ASSOCIATED WITH MORNING GLORY SYNDROME

Case report

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ABSTRACT - The basal encephaloceles refer to rare entities and they correspond to herniation of brain tissue through defects of skull along the cribiform plate or the sphenoid bone. A rare morning glory syndrome, with characteristic retinal defect has been reported in association with basal encephaloceles. Hypophysis hormonal deficiencies may occur. We accounted for a pituitary dwarfism with delayed diagnosed transsphenoidal encephalocele associated with morning glory syndrome, showing the alterations found in retinography, computed tomography and magnetic resonance imaging.

KEY WORDS: basal encephalocele, morning glory syndrome, computed tomography, magnetic resonance imaging, pituitary dwarfism.

Encefalocele basal associada a síndrome "morning glory": relato de caso

RESUMO - As encefaloceles basais são entidades raras e correspondem a herniações do tecido cerebral através de um defeito do crânio, ao longo da lâmina crivosa etmoidal ou do osso esfenoidal. A rara síndrome morning glory, com alterações de fundo de olho características pode apresentar-se associada à encefalocele basal. Deficiências hormonais hipofisárias podem ocorrer. Relatamos caso de nanismo hipofisário com encefalocele transesfenoidal de diagnóstico tardio associada à síndrome de morning glory, mostrando as alterações na retinografia, tomografia computadorizada e ressonância magnética.

PALAVRAS-CHAVE: encefalocele basal, síndrome de "morning glory", tomografia computadorizada, ressonância magnética, nanismo hipofisário.

The basal cephaloceles refer to rare entities of difficult diagnosis and correspond to herniation of the brain tissue through a birth or acquired defect in the skull along the cribiform plate or through the sphenoid bone. It may be associated to hormonal disturbances or ocular malformation and, amongst, the rare morning glory syndrome[1-3], which name is given due to the retinal aspect similar to the tropical flower of same name (Fig 1A). It is believed that such anomalies result from a succession of events for the medium line conclusion during the gestation period[3]. The computed tomography (CT) and magnetic resonance image (MRI) exams play a very important role for these anomalies since they evaluate the whole skull and present structures in the hernial content[1,2].

We present a case of defect on the medium line in which the TC and MRI images are fundamental to clarify the diagnosis.

CASE

A 8 years-old boy, was taken to have his retarded neurospychomotor evaluated. Born at term and with no intercurrences, he has evolved with a low weight and height gain since his first year of life, disproportional to the family growth potential. He presented converging squint, nystagmus and visual loss of the left eye. The isolated deficiency of the growth hormone diagnosis was confirmed. He received hormonal reposition presenting unsatisfactory response. The retinography and ophthalmological exam showed

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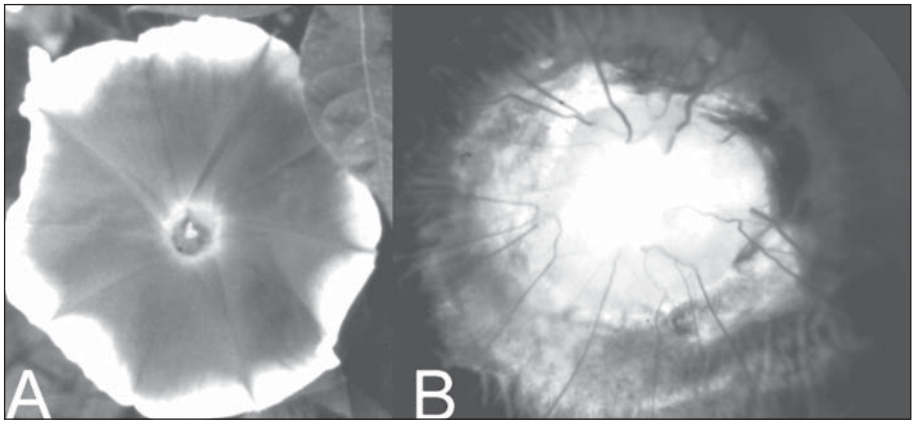

Fig 1. (A) The flower morning glory. (B) Left eye retinography with a morning glory aspect: optical disk enlarged choanoid and cupped aspect, pink coloration with a central white mass which hides the vessels ways at the back of the disk. It's is surrounded by a grey ring, a little lifted, with irregular borders and mixed with some colored areas. The vessels are multiple, thin and radiated. The yellow membrane of the remaining vitreous over the superior temporal area of the disk.

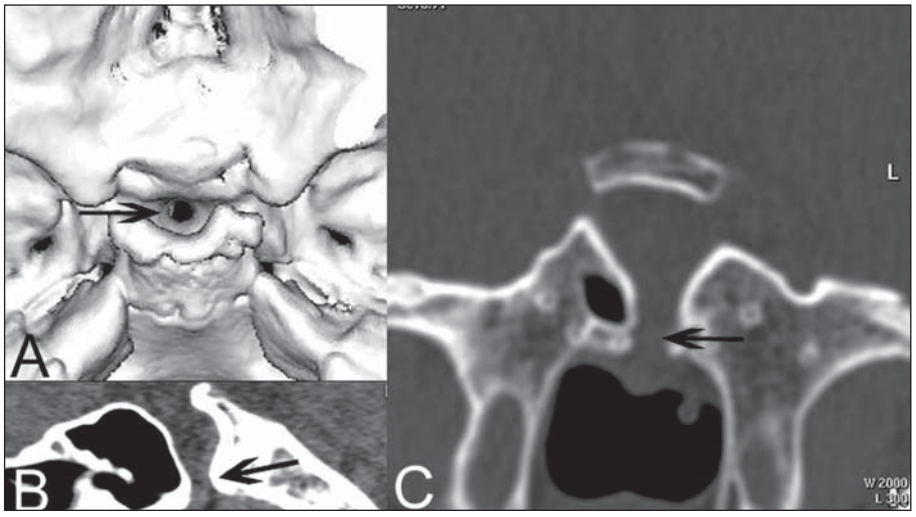

Fig 2. (A) A 3D reconstructed sella turcica CT image showing the main damage of the floor (arrow). (B) CT image reconstruction in the sagittal plane, in soft window showing the damage on the sellar floor and the encephalocele (arrow). (C) Coronal CT image in bone window showing the main flaw on the sellar floor (arrow).

characteristic alterations of the morning glory syndrome in the left eye, observing an optical disk with an enlarged choanoid and cupped aspect, with a pink pigmentation and a central white mass which hid the way of the vessels at the bottom of the disk. The disk was surrounded by a little elevated grey ring, with irregular borders and mixed with colored areas. The vessels were multiple, thin and radiated (Fig 1B).

At the age of 20, he was submitted to imaging exams of the sellar region. On the CT of the sella, with volumetric acquisition and three-dimensional reconstruction, a defect at the main area of the sellar floor was observed (Fig 2). On the MRI (Phillips Gyroscan 1,5T), sagittal and coronal images from the sellar region were obtained on the T1 weighted spin echo (T1WSE) sequence before and after the paramagnetic contrast medium intravenous administered, and in T2 weighted spin echo sequence. A sellar content constituted by the pituitary stalk, optical chiasm, adenohypophysis and neurohypophysis occurring on the right side was observed. It was observed the extension of the anterior portion of the third ventricle into the interior of the sella, and the hernial content with tissue characteristics not defined on the inner side of the sellar floor and of the sphenoidal sinus (Fig 3).

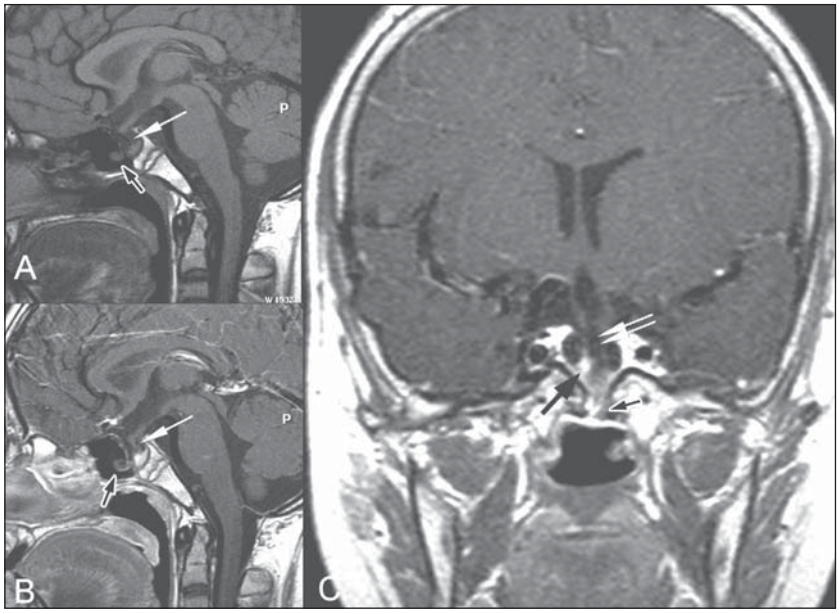

Fig 3. MRI images of the sagittal plane on T1 without iodine contrast (A), sagittal with iodine contrast (B) and coronal with iodine contrast (C) showing the hypophysis in the inner sella (black arrow). Inferior retraction of the pituitary stalk (white arrow), the optic chiasm and anterior portion of the third ventricle to the inner sella (double white cross). The bone flaw on the sellar floor with the hernial content of undistinguished anatomic aspect, characterized as gliocele (black arrow with white border), is located into the sphenoidal sinus.

DISCUSSION

Encephalocele is a congenital defect of the skull bone and of the dura-mater with extracranial herniation of any intracranial structure. It is found with a geographic variation and with different occurrences when related to sex and race, and in different association to the neural tube malformations, suggesting that some of them may present genetic origin, and that the several types of encephalocele may correspond to distinct genetic defects[4]. It can be divided into four groups. The cephalic meningoceles, which are constituted by the leptomeninges and the cerebrospinal fluid (CSF), and the glioceles, which are glial cells cysts containing CSF. The meningoencephaloceles, which consist of leptomeninges, CSF and brain parenchyma, recognizes the meningoencephaloventriculocele when parts of the ventricles and of portions of the choroid plexus participate on the herniation. And the atresic changes on the encephalocele, characterized by tuberous lesions situated on the medium line of the scalp, either in the vertex (parietal form) or in the occipital protuberance (occipital form)[4].

The basal encephalocele is the rarest and of the most difficult diagnostic and it corresponds to 1% to 10% of all of them[1,2]. The basal encephalocele frequently escapes the diagnosis and may be detected at adult age[2,4,5]. On the transsphenoidal encephalocele, the bone defect occur as a result of the chondrification of the intersphenoidal synchondrosis defect on the sphenoid body, causing the persistence of the craniopharyngeal canal, which normally closes itself by the fiftieth gestation day. Its persistence allows the passage of several portions of the intracranial structure such as the hypophysis, the anterior portion of the third ventricle floor, optic chiasm and optic nerves[1,2,4]. The symptoms may be developed at the neonatal period or at the first childhood, and they consist of expansive processes of the epipharynx and pituitary dwarfism[3,6,7], and the hypertelorism is almost constant. When these symptoms do not manifest themselves, visual disturbances and hypothalamohypophysial dysfunction[1] may lead to the diagnostic, as occurred in our case.

The basal encephaloceles associate themselves to optic malformation and retinal defect[1], observing an increase on the prevailing (67.7%) of the morning glory syndrome[3,7-10], which is an unusual congenital anomaly on the optic nerve. It was described by Reis and by Handman[3]. Kindler called it morning glory[3,11]. Its frequency on the population is unknown[5] and it is transmitted hereditarily with an autossomal dominant pattern. Most cases are unilateral[3,4] but there are rare cases of bilaterality[6,7] with twice as much frequency on females[3]. It is featured by the increase and copping of the optic nerve on the optic disk region with the persistence of a glial tissue with a yellow color in the middle constituted by hyaloid remains. The vessels follow a radial pattern to the periphery. The coloboma is surrounded by a lifted ring of retinal pigmentation, which resembles the morning glory flower. Clinically, there is a decrease of the unilateral visual accuracy frequently associated to the displacement of the retina, which occurs in 30% to 38% of the cases[12-14].

Some theories have been proposed to explain the malformation. Itakura and coworkers[8] described a succession of events which culminated in the bottom of the eye anomaly. According to these authors, the scirrhous palate would have embryological origin on the first brachial arch, the palatine process, which would originate itself from the maxillary process, melting completely with the septonasal, which in turn derives from a frontal saliency around the sixtieth gestational day. In this stage, no internal layers of the retina and of the optic nerve are well differentiated. During the seventh gestational week, the axons of the ganglionic cells of the retina start to form the optic nerve reaching its full development around the twenty-seventh gestational week. If a transsphenoidal encephalocele blocks the palate fusion, and as this phenomenon precedes the optic nerve formation, there could be an abnormal development of the nerve with a white glial tissue formation in its centre[8,13,15]. These abnormalities may present themselves in many combinations and in different degrees of severity and, therefore, the possibility of association cannot be discard even on the absence of exuberating clinical evidence, but being always indicated to a detailed evaluation through imaging exams.

The hypothalamic structures involvement may be associated to endochrinal alterations, mainly the growth hormone deficiency, though it must be considered the occurrence of the deficiency of multiple adenohypophysial hormones, which will appear later on the evolution process[1]. In this case, it was only observed a growth hormone deficiency and it has not yet developed any other hormonal deficiency.

The imaging exams have an important role on the basal encephaloceles diagnosis. Machado Jr and coworkers described the case of a patient with no previous neurological and/or endochrinal symptoms subjected to a CT exam because of a mild cranial trauma. A right parasellar lesion was observed, requiring a MRI exam to identify the incidental sphenoidal meningoencephaloventriculocele[16]. The encephalocele was into the sphenoidal sinus through the side wall with a discrete sellar deformation, although without hypothalamic or hypophyseal compromising.

MRI is the imaging diagnostic procedure of choice since it allows to precisely identifying the presence of meninges, brain parenchyma and blood vessels inside the bone defect[1]. Besides, it provides broad encephalic anatomic evaluation which facilitates the identification of other anomalies. The considered T1WSE imaging presents an anatomic resolution and it must be used to trying to identify cerebral structures which are normally deformed inside the herniation. The usage of intravenous paramagnetic contrast media helps to identify vascular structures and it might be important to evaluate surgical risks[17]. The images must also be obtained in larger areas for a complete evaluation of the brain parenchyma to disclose malformations. In our case, the structures in the interior of the sphenoidal sinus were not characterized as brain tissue and so being classified as gliocele[5]. The CT exam can better show the bone defect and it must be considered as a complementary method of imaging diagnostic, and the 3-D reconstruction facilitates this process.

The skull image study is fundamental in the diagnostic process of the ocular congenital alteration related to the retarded growth due to a possible association to the basal encephalocele. The chosen procedure is MRI using CT as a complementary method. The association of basal encephaloceles to endochrinal disorders and visual alteration suggests that a brain imaging study must be performed and completed with an exam directed to the hypothalamo-hypophysial region.

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