IMAGE

Case 14

Caso 14

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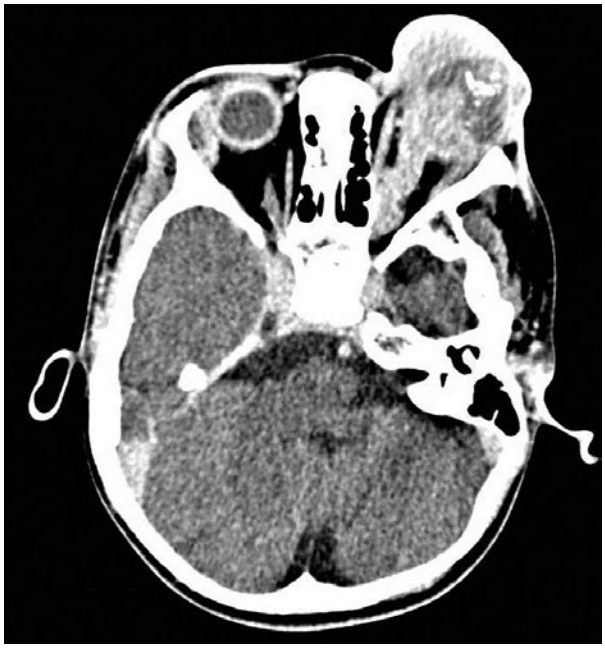

Figure 1 - Contrasted CT scan of the skull.

CASE

Male child, two years old, native of Parauapebas (PA), presented a white reflex in the left eye in pictures since the first months of life. Evolved with a progressive decrease in visual acuity, headache, and eyeball alterations highlighted in pictures and CT scan.

Based on the clinical history and images, the more likely diagnosis is:

a. congenital cataracts;
b. preorbital cellulite;
c. congenital glaucoma;
d. extraocular retinoblastoma.

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IMAGE ANALYSIS

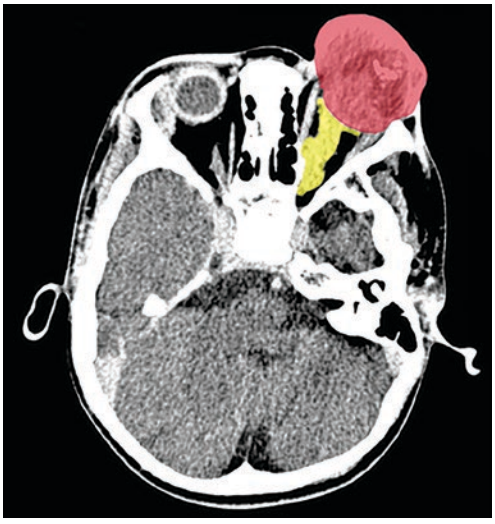

Figure 2 - Cross-sectional contrasted CT scan of the skull at the level of the sella turcica. Highlighted is the mass in the left ocular orbit and proptosis (in red) associated with ipsilateral optic nerve thickening (in yellow).

DIAGNOSIS

The framework of leukocoria (white pupil) from birth, growth of a mass in the orbital cavity, and proptosis of the eyeball is epidemiologically and clinically very suggestive of retinoblastoma. The headache, in this context, likely denotes involvement of the central nervous system by the tumor.

The congenital cataract is the opacification of the crystalline lens observed at birth and just as in the retinoblastoma, it is an early sign of leukocoria. It is one of the leading causes of blindness in children and is commonly caused by intrauterine infections, metabolic disorders, and genetic factors. If left untreated in the early months of life it can evolve with amblyopia and nystagmus or strabismus.

The congenital glaucoma, increased intraocular pressure in children with ocular malformations, has manifestations that are distinct from those presented in this case. It is characterized by tearing, photophobia, and disproportionately large eyes with bluish and opaque corneas. It is also among the leading causes of infant blindness, which can be prevented by early treatment.

The periorbital cellulitis is an infection of the soft tissues adjacent to the eyeball with acute evolution, potentially serious, to which the framework of infection of the upper airways generally succeeds. It can present diplopia, eye pain, eyelid edema, and proptosis.

DISCUSSION OF THE CASE

Retinoblastoma (RB) is the most common intraocular malignant tumor in childhood corresponding to about 3% of Pediatric neoplasms. It involves one in every 20,000 live births annually, with incidence peaking in the first three to four years of life. RB is originally from the embryonic retinal neuroectodermal membrane and occurs due to mutations in a tumor suppressor gene, the RB1.

The RB signs and symptoms depend on its size and location, the most common being leukocoria, present in 79% of the cases, also called “cat’s eye” in the red reflex test or in pictures. Other manifestations include strabismus (10.7% of cases), conjunctival hyperemia, decreased visual acuity, and secondary glaucoma. When the tumor becomes extraocular, as in this case, it presents itself as an orbital mass (3.4% of cases) with proptosis or invasion of the optic nerve. In cases with metastasis to the central nervous system, there may be headache, vomiting, anorexia, and irritability. If there is bone metastasis, it can follow with local pain.

RB is responsible for over 50% of cases of leukocoria, however, this signal can also be found in cataract, retinopathy of prematurity, primitive vitreous hyperplasia persistence, Coats disease, toxocariasis, and astrocytic hamartoma.

Parents and pediatricians play a critical role in detecting ocular alterations; an early ophthalmologic evaluation of all children with leukocoria and strabismus is crucial. Direct funduscopic examination can have up to 100% diagnostic sensitivity if performed with pupillary dilation.

Imaging exams such as CT scan or cranial MRI are indicated for the evaluation of the tumor extension. The RB treatment aims at preservation of life and vision, which depends on the tumor staging. The available therapeutic modalities are enucleation, transpupilar thermotherapy, cryotherapy, laser, brachytherapy, external radiotherapy, and chemotherapy.

The child in this case presents an extraocular retinoblastoma with extension to optical pathways (Figure 3) and metastases to frontal and occipital lobes, bone (Figure 4), and bone marrow. There was a long delay for its diagnosis and referral, which limited the

prognosis and, therefore, justifies the purpose of this discussion to warn about the early signs of the disease.

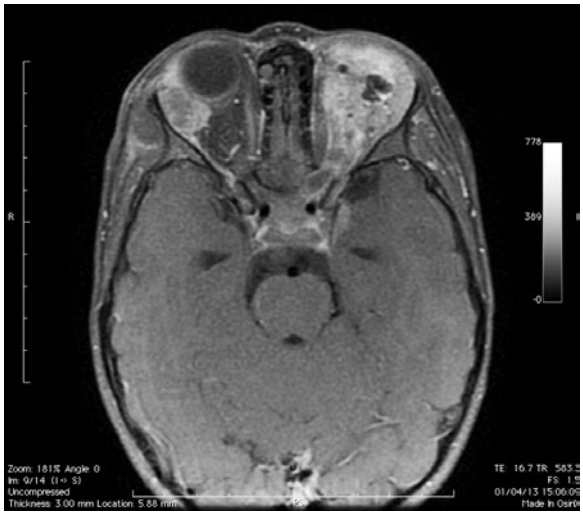

Figure 3 - MRI after enucleation of the left eye, T1-weighted, in a cross-sectional view, at the level of the optic chiasm and optic nerves, showing the thickening of these structures that had been infiltrated by the tumor.

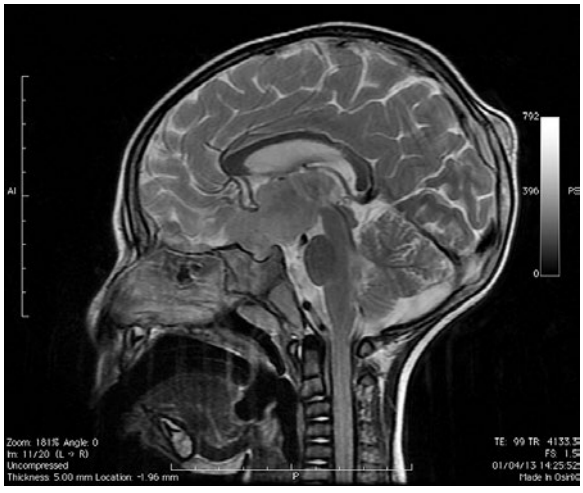

Figure 4 - MRI after enucleation of the left eye, T2--weighted, in a sagittal view, approximately at the middle line level. Metastases in the brain parenchyma and occipital bone metastasis are observed.

RELEVANT ASPECTS

Retinoblastoma (RB) is the most common malignant intraocular tumor in childhood and afflicts annually one every 20,000 live births, with 80% of the diagnoses in children younger than three or four years old.

The RB signs and symptoms depend on its size and location, the most common being leukocoria (79% of cases) and strabismus (10.7% of cases).

Parents and pediatricians play a critical role in early detection of eye alterations; the evaluation by an ophthalmologist and direct funduscopic examination are essential.

The RB treatment aims at the preservation of life and patient's vision, which depends on the tumor staging. The earlier the diagnosis, the better the prognosis tends to be.

The red reflex test or "little eye test" is used in Brazil in newborn screenings for various diseases including retinoblastoma and congenital cataracts and glaucoma.

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