Fetal diagnosis of congenital ocular toxoplasmosis: a case report
Diagnóstico fetal de toxoplasmosis ocular congénita: a propósito de un caso

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ABSTRACT
Congenital toxoplasmosis has serious implications for the prognosis of the newborn, as it can affect the brain and the eye with severe sequelae. The characteristic active lesion of congenital ocular toxoplasmosis is chorioretinitis which is diagnosed at birth by fundus examination. There are no fetal reports of detection of this lesion by ultrasound, but there are reports of severe lesions such as microphthalmia and cataract. We present a case of congenital toxoplasmosis in the fetus of a pregnant woman with untreated HIV who showed abnormal images in the eye on ultrasound that correlated with lesions in the vitreous and retina in the fundus images at birth.

Key words: Congenital toxoplasmosis, ocular, Toxoplasma gondii, Chorioretinopathy

RESUMEN
La toxoplasmosis congénita tiene graves implicancias para el pronóstico del recién nacido, pues puede afectar al cerebro y al ojo con secuelas severas. La toxoplasmosis ocular congénita tiene como lesión activa característica a la retinochoroiditis, que se diagnostica al nacer por medio del fondo de ojo. No existen reportes fetales de la detección de esta lesión por ultrasonido, pero sí de lesiones severas como microftalmia y catarata. Presentamos un caso de toxoplasmosis congénita en el feto de una gestante con VIH sin tratamiento quien al ultrasonido mostró imágenes anormales en el ojo que se correlacionaron con lesiones en vitreous y retina en las imágenes del fondo de ojo al nacer.

Palabras clave. Congenital toxoplasmosis, ocular, Toxoplasma gondii, Chorioretinopatía

INTRODUCTION
The worldwide prevalence of toxoplasmosis seroconversion is variable (USA 20%, France 60%). In South America prevalence rates are high, with the main reference being found in Brazil in 60-80% of the population(1,2). Primary maternal infection by toxoplasmosis is usually asymptomatic, but in gestation it has been related to abortion, fetal death, growth restriction, ventriculomegaly, ocular involvement, hearing impairment and neurodevelopmental deficits(3). Congenital toxoplasmosis infection (CT) varies in severity and degree of fetal involvement depending on the trimester in which it occurs, being more severe if occurring in the first trimester. CT is frequent in our environment, although its exact prevalence is unknown. In its severe forms, it can cause severe neurological and ocular sequelae in the newborn.

Regarding prenatal study, the diagnosis of suspicion is made by maternal serology, to later confirm fetal infection with PCR in amniotic fluid. Ultrasound can show characteristic lesions such as fetal brain lesions, and alterations in the brain parenchyma can be identified at a very early stage. However, with regard to ocular involvement, only severe and late lesions (cataract, microphthalmia, strabismus) have been described on ultrasound(4). Although the characteristic lesion of ocular toxoplasmosis is retinochoroiditis(3,5), no case of prenatal diagnosis by ultrasound has been published in the world literature.

In the present communication we publish the first images of ocular involvement in a fetus affected by congenital toxoplasmosis presumably attributed to retinochoroiditis.
CASE REPORT

A 35-year-old multigestation woman (G3P2002) at 37 weeks of gestation was referred with a diagnosis of human immunodeficiency virus (HIV) infection. A significant epidemiological history was ruled out. With only two prenatal controls, she was diagnosed with HIV at 32 weeks and antiretroviral treatment was started at 34 weeks. In the routine ultrasound, ventriculomegaly was found (ventricular atrium of 13.8 mm), so neurosonography was requested, which showed hyperechogenic lesions of diffuse border of cottony appearance distributed in various areas of the brain parenchyma and symmetrical dilatation of the ventricular system, findings suggestive of congenital infection by toxoplasma (Figure 1). In the evaluation of the eyes, an irregularity in the retina near the optic nerve was observed in the left eye. A possible toxoplasmosis lesion was suggested. No alterations of the vitreous, crystalline lens or cornea were observed, nor strabismus (Figures 2 and 3). The head circumference was in the 2nd percentile and the fetal weighted head circumference was in the 11th percentile for gestational age.

Delivery was by cesarean section on the seventh day after exploration. The newborn had Apgar 9-9 and was classified as having probable congenital HIV infection and TORCH, hepatosplenomegaly, mild anemia, and thrombocytopenia. Peripheral blood PCR was positive for HIV at 7 days after birth and antiretroviral treatment was started.

A fundoscopic examination was performed at 24 days of life and the right eye was found to have vitritis and the left eye had a whitish lesion tractioning between the macula and the optic nerve. Serology for toxoplasmosis at 30 days of life was IgM negative and IgG positive. Treatment for congenital toxoplasmosis was started with cotrimoxazole. Fundus examination at 6 months of life showed: right eye with normal fundus, left eye with fibrotic vitreoretinal scar lesion tractioning towards the optic nerve. At one-year follow-up, the infant was on antiretroviral treatment and was diagnosed with pulmonary tuberculosis.

DISCUSSION

Congenital ocular toxoplasmosis infection produces primary lesions by direct invasion of the endothelium through dendritic cells or by tachyzoite entry into retinal endothelial cells, glial cells and pigment epithelium. Necrotizing retinitis is generated and the underlying choroid may also be affected by the accompanying inflammation (3,5).

![Figure 1. A) Ventricular atrium in ranges of ventriculomegaly. B) Coronal view at the level of the anterior horns showing symmetrical dilatation of both horns. C) Sagittal view of the brain showing a hyperechogenic lesion with diffuse borders of cotton wool-like appearance in the vicinity of the corpus callosum.](image-url)
The standard diagnostic tool for ocular involvement of toxoplasmosis is the fundus examination. Ocular lesions described by fundus in acute ocular toxoplasmosis usually manifest as a well-defined focus of vitritis due to retinal necrosis and if there is involvement of the underlying choroid it is called retinochoroiditis. Active lesions are described as whitish, dark-rimmed foci. These active lesions tend to heal in 2-4 months, leaving an atrophic area with a white center due to exposure of the sclera and a hyperpigmented scar border (which resolves from the periphery to the center). Inflammation of the vitreous (vitritis) characterized by a darkening of the vitreous tends to be more prominent near active retinochoroiditis lesions. Severe vitritis may lead to epi-retinal membrane formation with subsequent vitreoretinal traction near the area of retinochoroiditis\(^1\).
The high association of ocular involvement in cases of CT with brain involvement in infants is not correlated in fetal reports. Severe eye involvement is sporadically reported\(^3\). This is possibly determined by the primarily retinal involvement, which is difficult to objectify, and we could only potentially evidence it as an irregularity at the moment of maximum retinal thickening. If there is vitreous involvement it could also be evidenced, but these usually occur after the active stage, with the exception of vitritis.

The absence of information on fetal retinochoroidal involvement forces us to review and extrapolate the findings in infants and adults to understand the findings in the fetus.

Vitreous and retinochoroidal involvement of active toxoplasmosis has been shown to be evidenced by ultrasound in adults, finding images in vitritis characterized by the presence of multiple punctate echoes and retinochoroidal thickening produced by inflammatory exudate with perilesional edema, which could potentially be replicable in the fetus if assessed at appropriate stages of ocular involvement\(^6\). This could explain the ultrasound image of a thickening in the retina of the fetus (Figures 2 and 3).

By reviewing the main CT series in newborns and infants we can get an idea of the behavior in the fetal stage. Bosch-Driessen in a large series of cases of ocular toxoplasmosis (154 cases from the Netherlands) identified 13 cases (8%) of congenital infection, evaluated in the first two years of life. All of them presented inactive scars in the retina, predominantly central and with involvement of the macula\(^7\).

Kodjikian presents a series of 430 cases of children with CT, of whom 130 (30%) had ocular toxoplasmosis with retinochoroiditis at birth. Seventy-seven percent were cicatrical foci and 23% were active foci, characteristically mostly of central distribution (64%). Other ocular lesions such as strabismus, nystagmus and cataract were also described\(^8\).

Melamed\(^9\) examined 44 children under one year of age diagnosed with CT in Porto Alegre, Brazil. Maternal infection was demonstrated mostly in the third trimester (47.7%), although the timing was unknown in a high percentage (43%). Ocular involvement was found in 31 cases (70%), the predominant lesion was retinochoroiditis in 21 children with a classic ‘cartwheel’ appearance and variable degrees of pigmentation (65%); it was of central location in 48%, active retinal lesions were visible in 15% of cases and vitreous opacity (vitritis) did not allow fundoscopy in 5 cases. Other ocular lesions were strabismus, nystagmus, cataract and microphthalmia (12, 7, 6 and 5 cases, respectively).

Conceicao\(^10\) presents a series of children with CT with ocular manifestations during an epidemic in a locality in Brazil, presumably associated with water contamination. Of the 184 pregnant women who were treated for toxoplasmosis infection, 29 newborns were diagnosed with CT, 45% with cerebral calcifications and 65% with ocular manifestations. Of these ocular manifestations, 76% of the lesions corresponded to retinochoroidal lesions in the posterior segment and in the vicinity of the optic nerve-macula and 13% showed optic disc abnormalities. Retinochoroidal lesions with signs of activity were found in 6 infants in association with vitritis.

In conclusion, what we can consolidate so far from the series in infants is that the initial active lesion of retinochoroiditis is associated with vitreous alterations and may present as a thickening. This lesion is centrally and posteriorly distributed. In the present case, the findings were compatible with these characteristics, both fetal and neonatal, which suggests that the findings in the fetus correspond to the period of activity with retinochoroidal edema of a retinochoroiditis lesion (Figures 2 and 3).

**References**


