MACULAR RETINOCHOROIDAL SCARS IN TWO SIBLINGS: A PRESUMED CASE OF CONGENITAL OCULAR TOXOPLASMOSIS

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ABSTRACT

Background*: Congenital infection results from vertical transmission of Toxoplasma gondii during pregnancy and may cause permanent visual impairment due to retinochoroiditis involving the macula. Although congenital ocular toxoplasmosis is well recognized, reports involving siblings remain uncommon.* ***Case Presentation****: We report two sisters presenting with longstanding visual impairment caused by presumed congenital ocular toxoplasmosis. The first patient, a 22-year-old woman, presented with bilateral blurred vision since childhood. Best-corrected visual acuity (BCVA) was 0.16 in both eyes. Fundus examination demonstrated bilateral hyperpigmented macular retinochoroidal scars consistent with inactive toxoplasmic retinochoroiditis. The second patient, a 25-year-old woman, complained of blurred vision in the left eye since childhood. BCVA was 1.0 in the right eye and 0.20 in the left eye. Funduscopic examination revealed a solitary hyperpigmented retinochoroidal scar involving the left macula. Neither patient showed signs of active intraocular inflammation. The mother had no history of ocular toxoplasmosis or visible retinal scars.* ***Conclusion****: The occurrence of characteristic macular toxoplasmic scars in siblings strongly suggests presumed congenital ocular toxoplasmosis despite the absence of maternal ocular lesions. Recognition of congenital infection is important because macular involvement results in irreversible visual loss. Prevention of maternal infection during pregnancy and appropriate prenatal management remain essential to reduce fetal morbidity.*

Keywords: *congenital ocular toxoplasmosis; retinochoroidal scar; macular scar; siblings; Toxoplasma gondii*

INTRODUCTION

Toxoplasma gondii is an obligate intracellular protozoan parasite that infects approximately one-third of the world's population. It can replicates in intestine of cats and other felines, as a definitive hosts and Warm-blooded animals and humans are intermediate hosts.[1] Human infection is commonly acquired through ingestion of tissue cysts in undercooked meat, consumption of food or water contaminated with oocysts shed by cats, or transplacental transmission during pregnancy.[2] Ocular toxoplasmosis is the leading cause of infectious posterior uveitis worldwide and represents an important cause of permanent visual impairment among young adults.

Congenital toxoplasmosis occurs when maternal infection is transmitted to the fetus during pregnancy.[1] T. gondii may cross the placenta and enters the fetal circulation during the phase of parasitemia resulting congenital toxoplasmosis infection.[1] Although the likelihood of fetal transmission increases with advancing gestational age, infections acquired during the first trimester generally result in more severe ocular and neurological manifestations. The classic ocular manifestation is necrotizing retinochoroiditis, which subsequently heals with a characteristic hyperpigmented retinochoroidal scar. Lesions involving the macula frequently cause irreversible central visual loss because retinal architecture cannot be restored after healing.

Most congenital infections occur following primary maternal infection. However, rare cases have been reported following reactivation of latent infection or reinfection with genetically different strains in immunocompetent mothers. The diagnosis of congenital ocular toxoplasmosis in adults may be challenging because systemic manifestations are often absent, maternal infection may have gone unrecognized, and retinal lesions are discovered only after visual symptoms become apparent.

Toxoplasma gondii has multiple transmission pathways involving both definitive and intermediate hosts.[3] Cats is the definitive hosts, it unsporulated oocysts in their feces, which become infective after undergoing sporulation in the environment. These sporulated oocysts can contaminate water, soil, and food sources such as raw vegetables, and may subsequently be ingested by intermediate hosts, including cattle, sheep, poultry, and pigs, as well as humans. In intermediate hosts, the parasite forms tissue cysts, which can infect humans through the consumption of raw or undercooked meat. In addition to oral transmission, infection can occur vertically when tachyzoites are transmitted transplacentally from a pregnant woman with a primary infection to the fetus, resulting in congenital toxoplasmosis. Transmission may also occur through blood

transfusion or organ transplantation from infected donors (Figure 1).

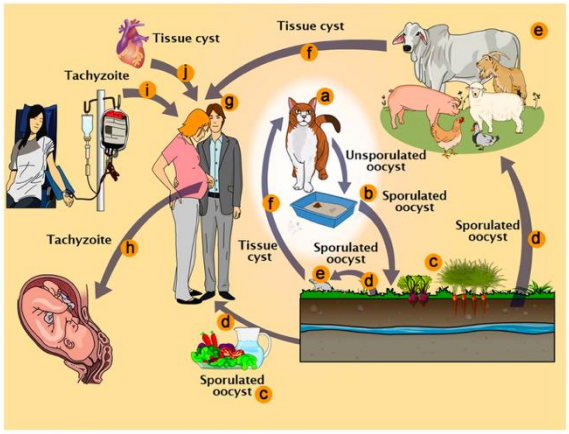

Figure 1. Life cycle of toxoplasmosis (Courtesy: Attias et al.)[3]

This Case reports describing presumed congenital ocular toxoplasmosis occurring in siblings remain uncommon. We report two sisters presenting with bilateral and unilateral macular toxoplasmic scars, respectively, highlighting the clinical characteristics and diagnostic considerations of presumed congenital infection.

CASE PRESENTATION

This Case Report describing two cases of congenital toxoplasmosis. First case is 22-year-old woman presented with bilateral blurred vision that had been present since childhood without episodes of ocular pain, redness, or photophobia. There was no previous history of ocular trauma, intraocular surgery, or systemic inflammatory disease.

Best-corrected visual acuity (BCVA) was 0.16 in both eyes. Anterior segment examination was unremarkable, with no evidence of anterior chamber inflammation. Intraocular pressure was within normal limits. Dilated fundus examination revealed well-defined hyperpigmented retinochoroidal scars involving the macular region of both eyes (Figure 2). The lesions were sharply demarcated with surrounding retinal pigment epithelium atrophy and fibrosis, compatible with inactive toxoplasmic retinochoroiditis. No vitreous inflammation or active retinal lesions were observed.

The patient was diagnosed with presumed congenital ocular toxoplasmosis with bilateral macular involvement. Because no active inflammation was present, antiparasitic therapy was not indicated. Visual rehabilitation and regular follow-up were recommended.

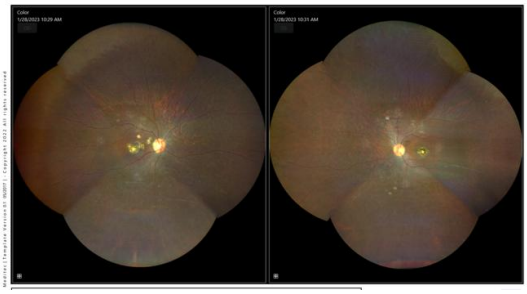

Figure 2. Toxoplasmic retinochoroidal scar in the macula on both eyes (Courtesy of the author)

Second case is 25-year-old woman, the older sister of case 1, presented with longstanding blurred vision in her left eye since childhood. She denied previous episodes of ocular inflammation or trauma.

Best-corrected visual acuity was 1.0 in the right eye and 0.20 in the left eye. Slit-lamp examination in the anterior revealed no abnormalities. Funduscopic examination demonstrated a solitary hyperpigmented retinochoroidal scar involving the left macula, while the right fundus appeared normal (Figure 3). Similar to the first case, no active retinitis or vitreous inflammation was identified.

Based on the characteristic appearance of the lesion and the presence of similar retinal scars in her sibling, the patient was diagnosed with presumed congenital ocular toxoplasmosis involving the left eye. Observation was advised because the lesion represented inactive disease.

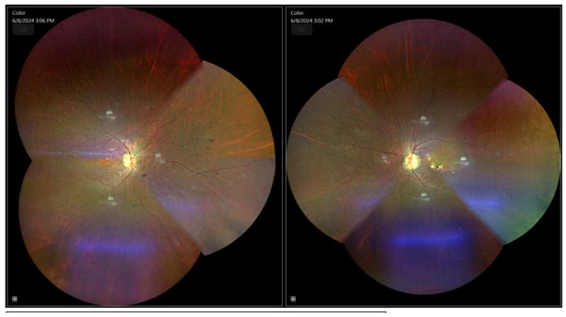

Figure 3. Toxoplasmic retinochoroidal scar in the macula on left eye (Courtesy of the author).

DISCUSSION

Congenital ocular toxoplasmosis represents an important cause of irreversible visual impairment among children and young adults. The necrotizing retinochoroiditis associated with Toxoplasma gondii infection may result in permanent retinal destruction and subsequent scar formation, leading to irreversible alterations in retinal structure.[4] Involvement of the macular region, as demonstrated in both patients in this report, is particularly associated with an unfavorable visual outcome because damage to photoreceptors and retinal pigment epithelial cells cannot be reversed after the inflammatory process has resolved

The diagnosis in these cases was established based on the characteristic ocular findings. Both siblings demontrated typical inactive hyperpigmented

retinochorioretinal scars involving the macula, without signs of active inflammation. Other potential causes of chorioretinal scarring, including congenital infections such as cytomegalovirus, rubella, toxocariasis, tuberculosis, and previous ocular trauma, were considered less probable due to the morphology of the lesions and the absence of supporting clinical history.

Ophthalmologic examination of the mother revealed no evidence of previous ocular toxoplasmosis or chorioretinal scarring. However, the absence of maternal ocular manifestations does not exclude congenital transmission, as maternal Toxoplasma gondii infection is frequently asymptomatic. Congenital infection may occur following primary maternal infection during pregnancy and, in rare circumstances, may also result from reactivation of latent infection or reinfection with different parasite strains.

The occurrence of congenital toxoplasmosis in siblings has been described only rarely.[5] Previous reports suggest that delayed clearance of circulating parasites after maternal infection or reactivation of latent infection during subsequent pregnancies may explain repeated congenital transmission. Pregnancy induces physiological immunomodulation, which may facilitate transient parasite reactivation even in immunocompetent women. The relatively short interval between pregnancies has also been proposed as a potential contributing factor in recurrent congenital infection.

Neither patient demonstrated active retinochoroiditis at presentation; therefore, antiparasitic treatment was not indicated.[6] Current management guidelines recommend treatment only during active ocular inflammation or when lesions threaten vision. Macular scars represent permanent retinal damage, and treatment cannot restore lost visual function. Consequently, low-vision rehabilitation and periodic monitoring for recurrence remain the principal management strategies.

Prevention remains the most effective strategy for reducing congenital toxoplasmosis. Pregnant women should avoid consumption of undercooked meat, thoroughly wash fruits and vegetables, practice careful hand hygiene after handling raw meat or soil, and minimize exposure to cat feces.[7] Serological screening during pregnancy, where available, facilitates early diagnosis and timely treatment, thereby reducing the risk of fetal transmission and severe ocular complications.

This report has several limitations, including the lack of serological confirmation and molecular identification of the parasite strain. Nevertheless, the characteristic funduscopic findings, early onset of visual symptoms, bilateral familial occurrence, and lack of alternative etiologies strongly support this diagnosis

CONCLUSION

This case report describes siblings with characteristic macular retinochoroidal scars consistent with presumed congenital ocular toxoplasmosis. Although definitive confirmation was unavailable, the clinical findings strongly suggest congenital infection. These cases emphasize that congenital ocular toxoplasmosis should be considered in young patients presenting with longstanding unexplained visual impairment and characteristic macular scars, particularly when similar findings are observed among siblings. Preventive strategies during pregnancy remain the cornerstone for reducing congenital transmission and preventing lifelong visual disability.

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