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Case Report - Veterinary Medicine

Ophthalmoscopic and ultrasonographic findings of collie eye anomaly in a female dog – case report

[*Achados oftalmoscópicos e ultrassonográficos da anomalia do olho do Collie em uma cadela – relato de caso*]

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ABSTRACT

Collie Eye Anomaly (CEA) is a congenital genetic defect primarily found in Collie dogs but occasionally in other breeds. Its significance lies in its genetic implications for breeding in predisposed breeds. Diagnosing CEA can be challenging and relies on clinical history, epidemiological data, clinical and ophthalmological examinations, and ocular ultrasonography to assess the extent of damage and prognosis. This article presents a case of CEA in a 6-month-old Border Collie dog who exhibited reduced vision and acute hyphema. The condition was diagnosed through direct ophthalmoscopy and ocular ultrasonography

Keywords: malformation, canine, ophthalmology, ultrasonography

RESUMO

A anomalia do olho do Collie (AOC) é caracterizada por um defeito congênito de origem genética, relatada predominantemente em cães de raças de pastoreio e descrita com menor frequência em outras linhagens. A importância da AOC está atribuída à relevância genética na seleção de reprodutores de animais das raças predisponentes. O diagnóstico da AOC é desafiador e deve ser baseado em histórico clínico, dados epidemiológicos, exames clínicos e oftalmológicos, associados à ultrassonografia ocular, a fim de determinar o grau de lesão e o prognóstico. Este artigo relata um caso de AOC em cadela da raça Border Collie, de seis meses de idade, a qual apresentou redução da visão e hifema agudo, diagnosticadas por meio da oftalmoscopia direta e da ultrassonografia ocular.

Palavras-chave: malformação, canina, oftalmologia, ultrassonografia

INTRODUCTION

Collie Eye Anomaly (CEA), initially described in 1953 by Magrane, is characterized by a hereditary and congenital defect in the back of the eye, and it affects both sexes equally (Mason and Cox, 1971; Rampazzo *et al*., 2005). This condition is more commonly observed in Collie lineage, such as the Border Collie and Shetland Sheepdog, and occasionally in other breeds like Beagle, Dachshund, and German Shepherd. The anomaly is associated with a mutation in the *NHEJ1* gene, leading to individuals carrying a modified genome, whether or not accompanied by visible phenotypic characteristics. (Marelli *et al*., 2022).

While the disease's occurrence is considered rare, studies have revealed a prevalence of 9.71% among animals evaluated as carriers of the mutant allele in the European dog population and approximately 1.98% in the North American dog population. These findings emphasize the importance of genetic assessment in animals selected for breeding (Majchrakova *et al*., 2023).

It is a bilateral condition, but the extent of involvement of ocular structures can differ between the two eyes (Palanova, 2015). The

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pathogenesis of the disease involves a defect in the mesodermal differentiation of the fibrous and vascular layers in the posterior region of the eye. This leads to conditions such as chorioretinal dysplasia, choroidal hypoplasia, optic disc coloboma, retinal detachment, and other vascular abnormalities. Depending on the severity of these alterations, individuals may experience visual deficits ranging from partial visual impairment to complete blindness (Barnett, 1979; Palanova, 2015).

Choroidal hypoplasia is consistently bilateral and can be detected through a fundus examination. It presents as an underdeveloped region with a noticeable absence of pigment in the retina and/or choroid, often appearing as a pale area in the temporal (lateral) vicinity of the optic disc. In these affected areas, blood vessels are more conspicuous but less numerous, often displaying a tortuous appearance. On a histological level, tapetal cells are typically absent, the pigment epithelium is reduced, and the choroid is thinner than what's considered typical (Barnett, 1979).

Coloboma is a less frequently observed anomaly in CEA and manifests as small, shallow, or occasionally deeper cavities that extend into a portion of the optic nerve. This can result in an enlargement of the optic disc area, and the presence or absence of blood vessels over this region can vary. It's important to note that while choroidal hypoplasia typically doesn't impact vision, coloboma can potentially lead to a reduction in visual acuity (Rampazzo *et al*., 2005).

Additional abnormalities, including retinal detachment, hyphema, and tortuosity of retinal blood vessels, can also be observed in CEA. The most accurate diagnosis of CEA typically occurs in puppies at 6 to 7 weeks of age. It's advisable for these dogs to undergo annual reevaluation to monitor their condition and any changes (Palanova, 2015).

Changes such as retinal detachment and intraocular hemorrhages can be detected through ultrasonographic examination. Coloboma, on the other hand, can be observed as a defect or excavation in the curvature of the posterior eye wall with varying depth and dimensions, typically appearing as a hypoechoic to anechoic area (Pinto, 2013). However, it is worth noting that specific descriptions in the literature regarding the ultrasonographic features of dogs affected by CEA are limited.

The primary aim of this report is to provide a description of CEA in a 6-month-old Border Collie female dog who presented with reduced vision and acute hyphema in the left eye. This diagnosis was confirmed through ophthalmoscopy and ocular ultrasonography.

CASE REPORT

A 6-month-old, non-spayed Border Collie female dog weighing 14.45kg was presented at the Veterinary Hospital for examination. During the ocular clinical assessment, the dog displayed acute hyphema in the left eye (Fig. 1 A), with no reported history of trauma or previous injury. The left eye also exhibited an absent threat response, while the right eye demonstrated a preserved response. Intraocular pressure measurements revealed elevated pressure in the left eye (27mmHg) and normal pressure in the right eye (15mmHg). The ophthalmoscopic examination of the fundus of the right eye revealed choroidal hypoplasia with the presence of larger and tortuous blood vessels, along with indications of a coloboma in the optic nerve region. (Fig. 1 B and C). However, due to the presence of hyphema in the left eye, direct ophthalmoscopic examination of the posterior segment was not possible. Therefore, ocular ultrasonography was recommended for the evaluation of intraocular and retrobulbar structures.

An ultrasound device, specifically the MyLab40 model from Esaote, was employed along with a linear probe operating at a frequency range of 10 to 12 MHz. The transcorneal positioning of the probe was facilitated using acoustic gel and a plastic shield. In the examination of the right eye, only the presence of echogenic foci within the lens and a small depression in the region of the optic disc were observed. Other ocular structures appeared to conform to normal ultrasonographic standards. In the examination of the left eye, several notable observations were made: the anterior chamber displayed an enlargement in its diameter, accompanied by the presence of echogenic points, indicative of intralenticular contentes. Within the vitreous chamber, membranous echoes formed a "V" shape in the

central region and were observed adhering to the optic disc. Additionally, a significant number of pinpoint echoes were detected in the subretinal area. In the region of the optic nerve, a rounded and hypoechoic excavation, measuring approximately 0.39 cm in thickness, was identified (Fig. 1 D and E).

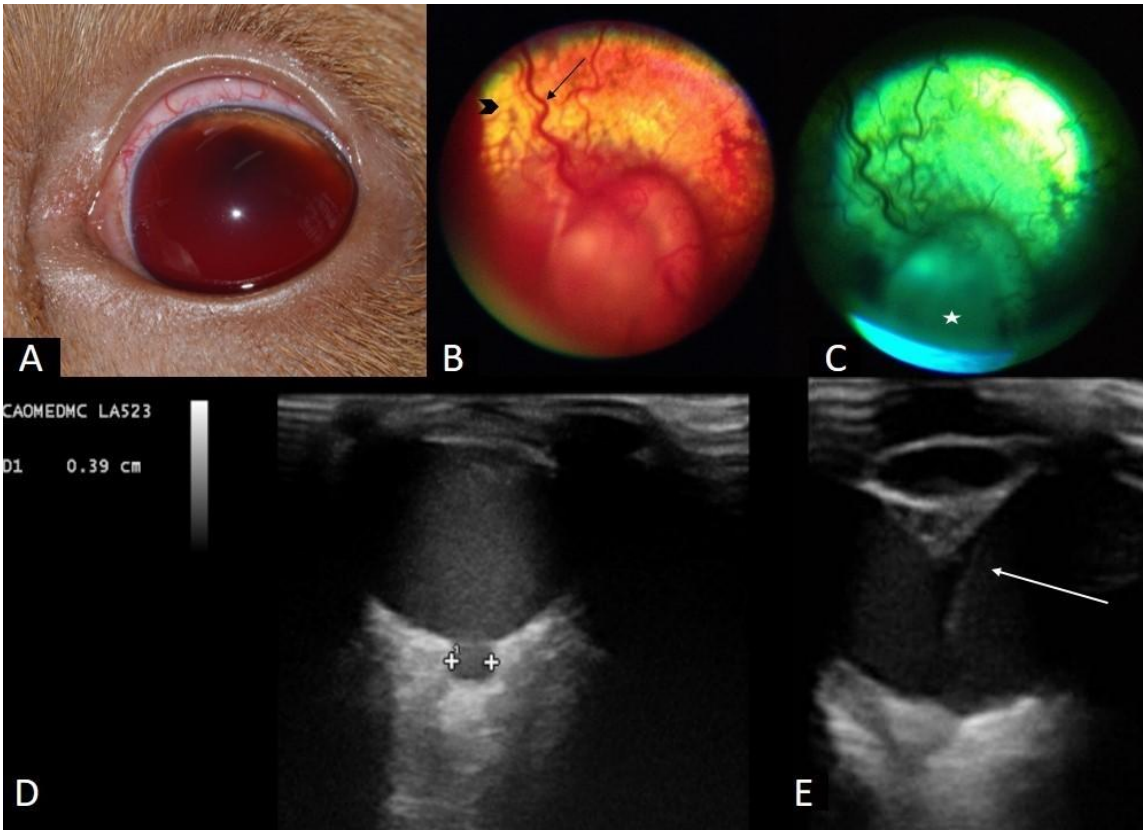

Figure 1: A. Photograph of the left eye of a 6-month-old Border Collie dog, showing the presence of hyphema in the anterior chamber. **B.** Ophthalmoscopic image of the right eye with dilated and tortuous blood vessels in the retina (arrowhead) and a visible area of the sclera, in line with choroidal hypoplasia (arrow). **C.** Ophthalmoscopic image of the right eye utilizing a green filter. A coloboma defect is clearly visible in the region of the optic disc (marked with a star). **D.** Ultrasonographic image of the left eye showing a hypoechoic excavation area in the optic disc region, with a thickness of approximately 0.39 cm (between cursors), suggesting the presence of a coloboma. **E.** Formation of a membranous echo in the central vitreous chamber (arrow), which is indicative of retinal detachment.

These findings were consistent with retinal detachment, intraocular hemorrhage, and coloboma in the left eye.

Treatment with steroidal anti-inflammatories was initiated; however, there was no improvement in the clinical condition. Consequently, the decision was made to proceed with enucleation surgery on the left eye, and the excised material was sent to the Veterinary Pathology department for further examination.

During the macroscopic evaluation, a significant amount of blood was observed in the anterior, posterior, and vitreous chambers of the eye. During the enucleation process and optic nerve section, an eye perforation was discovered. The tissues were subsequently fixed with formalin, and a histopathological examination was carried out.

The histopathological examination revealed the following findings: diffuse neutrophilic and eosinophilic infiltrates in the anterior, posterior, and vitreous chambers. Multifocal moderate

lymphohistioplasmocytic inflammatory infiltrates in the sclera and moderate multifocal uveitis with lymphoplasmacytic and neutrophilic inflammatory infiltrates. Total retinal detachment, which was associated with hypertrophy of the retinal pigment epithelium and the presence of subretinal inflammatory infiltrates.

DISCUSSION

This case report presents the ophthalmoscopic and ultrasonographic findings associated with Collie Eye Anomaly (CEA) in a 6-month-old Border Collie dog, a breed known for the congenital occurrence of CEA. The patient's age aligns with the typical onset of this condition, and it is important to note that CEA does not exhibit a sexual predilection, as supported by previous research. (Mason and Cox, 1971; Rampazzo *et al*., 2005). Nevertheless, it is worth acknowledging that this anomaly has been reported in other breeds, which may raise the possibility of reconsidering the nomenclature of the condition in the future (Abranches *et al*., 2011).

In accordance with Barnett (1979), this report underscores the bilateral nature of the condition, but it also highlights the differences in the extent of involvement of ocular structures between the two eyes.

Direct ophthalmoscopic examination of the fundus is crucial for the identification of lesions associated with CEA. In the examination of the right eye fundus, choroidal hypoplasia was evident. This was indicated by the presence of a whitish area near the optic disc and the visualization of dilated and tortuous vessels in the sclera. These findings are consistent with CEA, in line with the description provided by Barnett (1979). Additionally, in the region of the optic disc of the right eye, a coloboma defect was observed in the lower temporal portion. It is important to note that while choroidal hypoplasia is more common, coloboma is a less frequent alteration, present in approximately 35% of cases, as mentioned by Palanova (2015).

Nevertheless, it is worth noting that the evaluation of the posterior segment can become challenging when the anterior chamber is clouded by opacities. In the case of the left eye, the hyphema or hemorrhage observed during direct ophthalmoscopic examination may result from retinal detachment and the abnormal formation of retinal blood vessels, providing a possible explanation for the patient's condition, as suggested by Gelatt (2014).

As mentioned by Palanova (2015), the presence of hemorrhage in the posterior and vitreous chambers, as identified in the histopathological examination in this case, is not a common occurrence in CEA cases. This occurrence is relatively rare, affecting approximately 1% of dogs affected by the disease.

The diminished threat response and loss of vision in the left eye observed in the patient were linked to the total retinal detachment and widespread intraocular hemorrhage, in accordance with the description provided by Cunha *et al*. (2008).

Ocular ultrasonography is a readily available and cost-effective examination commonly performed in veterinary practice. When transparent media of the eye become opaque, ocular ultrasonography is the recommended diagnostic tool for assessing intraocular and retrobulbar lesions, as emphasized by Veiga *et al*. (2012). However, despite its recommendation, the ultrasonographic examination in this case did not reveal the echogenic points in the anterior chamber, which were later observed in the histopathological examination. This discrepancy may be attributed to the 12MHz probe frequency, which may result in reduced detail in very superficial regions.

Ideally, in such cases, a high-resolution ultrasound biomicroscopy (UBM) device would be preferred. Unfortunately, as noted, this option was not available for the specific clinical case presented (Allemann, 1995).

Retinal detachment can be congenital or occur prior to the age of two years, as noted by Bedford (1982). It commonly presents unilaterally, aligning with the patient condition as they had total retinal detachment in the left eye, possibly congenital considering the patient’s age.

During the ultrasonographic examination, distinctive findings included membranous echoes forming a V shape in the central vitreous

chamber, which is indicative of retinal detachment. Additionally, a substantial number of echogenic points in the subretinal area were observed. These ultrasonographic characteristics are in line with total retinal detachment and subretinal hemorrhage, consistent with the description provided by Veiga *et al*. (2012). These findings were subsequently confirmed by the histopathological examination.

The ultrasonographic examination played a critical role in assessing the more severely affected eye, the left eye, given the opacification of the anterior chamber, which hindered fundus evaluation by ophthalmoscopy.
In the optic disc region, apart from retinal detachment and subretinal hemorrhage, a hypoechoic, oval-shaped area lacking evident vascularization on color Doppler was associated with coloboma. This finding aligns with the descriptions provided by Pinto (2013) and Abreu *et al*. (2020), both of whom noted a defect or excavation in the curvature of the posterior eye wall with varying depth and dimensions in two-dimensional mode (mode B).

Similar lesions were also observed in the right eye, albeit less pronounced, characterized by a small area of depression in the optic disc region.

The histopathological examination encountered certain limitations because of the sectioning of the optic disc during enucleation, which made it inconclusive for the proper evaluation of the posterior segment and the choroidal region. However, other histopathological findings, such as the widespread infiltrate of inflammatory cells in the chambers, sclera, and uvea, were consistent with panophthalmitis, in accordance with the descriptions provided by Slatter (2013) and Jubb *et al*. (2016).

Panophthalmitis is a condition that can result from injuries related to inflammation in ocular tissues, total retinal detachment, intraocular hemorrhage, and a history of glaucoma. Unfortunately, due to the excision of the optic disc, an assessment of glaucoma was also not possible, as discussed by Gelatt (2014) and Jubb *et al*. (2016).

The present report addresses the less common alteration in CEA, which is coloboma. According to Pinto (2013) and Abreu *et al*. (2020), this anomaly manifests as a defect or excavation in the usual curvature of the posterior eye wall, with variations in depth and dimensions. It is important to note that specific descriptions in the literature concerning the ultrasonographic characteristics of dogs affected by CEA are notably scarce, as emphasized in the current report.

CONCLUSION

Ultrasonography serves as a crucial adjunctive tool in the diagnosis of Collie Eye Anomaly. It enables the identification of various ocular abnormalities, particularly coloboma, which may remain unseen in direct ophthalmoscopic examination due to hindrances posed by opacities within the transparent ocular tissues. The integration of ophthalmoscopic and ultrasonographic findings proved indispensable for both therapeutic decision-making and the establishment of the patient's ocular condition prognosis.

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