*Veterinary Ophthalmology* (2016) **19**, 6, 525–530 DOI:10.1111/vop.12328

CASE REPORT

# Atypical chorioretinal coloboma in a Golden Retriever: a retinographic, fluoroangiographic, and optical coherence tomography study

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**Abstract**

*Purpose* To report a case of canine atypical chorioretinal coloboma where ophthalmoscopic, fluoroangiographic and optical coherence tomography characteristics are described.

*Animal Studied* A 2-year-old Golden Retriever dog in which routine ophthalmoscopic examination allowed diagnosis of a posterior coloboma on the left eye.

*Procedures* Retinography, fluorescein angiography, and optical coherence tomography were performed to characterize the lesion of the left fundus.

*Results* Ophthalmoscopy revealed a pigmented circumscribed lesion dorsal to the optic nerve of the left fundus, surrounded by small areas of hyper-reflectivity and retinal vessels with an abnormal pattern. Fluoroangiography revealed a constant hypofluorescence of the defect and a complete altered vascular pattern associated with the coloboma. Optical coherence study demonstrated an important craterlike depression showing an atrophic neurosensory retina, and a lack of retinal pigment epithelium and choroidal tissue.

*Conclusions* Fluoroangiographic and optical coherence tomographic studies of an atypical chorioretinal coloboma in a dog are reported for the first time. Both imaging techniques are of great importance to characterize this type of congenital defects.

**Key Words:** atypical coloboma, choroid, dog, fluorescein angiography, optical coherence tomography, retina

## INTRODUCTION

Ocular colobomas result from defective embryogenesis.[1] Almost any ocular structure can be involved, but the vascular tunic of the eye (iris and choroid) is mostly affected.[2] Colobomas can be unilateral or bilateral. According to the site of genesis, they can be typical or atypical. Typical colobomas are located in the inferonasal quadrant and are caused by an incomplete closure of the embryonic fissure of the optical vesicle. They can affect any part of the globe from the iris to the optic nerve. Less frequently, colobomas can also be located in any other position and are termed atypical.[1,3,4] The pathogenesis of this atypical malformation is still unclear but seems to be associated with primary defects of the uveal epithelium (anterior segment colobomas) and/or retinal pigment epithelium (posterior segment colobomas).[5–7] Colobomas can be caused by a genetic mutation or by toxic environmental agents.[8] They are frequently associated with other developmental defects, mainly microphthalmia.[1,8]

Typical colobomas have been described in rats, mice, snow leopard, horses, donkeys, cattle, cats, and dogs.[5,9–16] Atypical colobomas have been described mainly in horses.[17] There are few cases described of atypical posterior colobomas in dogs, affecting the optic disk and the choroid.[18,19] To the authors' knowledge, there are no reports of fluoroangiographic and optical coherence tomographic studies of posterior atypical colobomas in dogs.

In this report, we describe an atypical unilateral chorioretinal coloboma in a Golden Retriever. Clinical,

electrophysiological, angiographic, and optical coherence tomographic findings are described.

## CASE REPORT

A 2-year-old Golden Retriever was presented at the Ophthalmology Service of the Veterinary Teaching Hospital of Madrid with a history of palpebral masses for the last month. Ophthalmic examination revealed two papillomatous lesions affecting the nasal palpebral skin of the right eye, 2 cm distant from the upper eyelid margin. Normal menace response, dazzle reflex, pupillary light reflexes, and slit-lamp biomicroscope examination were normal in both eyes. Intraocular pressures (Tono Vet®; Icare, Helsinky, Finland) were within normal range, 17 mmHg and 19 mmHg for the right and left eyes, respectively. Direct (Panoptic ™, Welch Allyn, AB Médica, Barcelona, Spain) and indirect ophthalmoscopy (Vantage Plus Led Digital®; Keeler, AJL Ophthalmic, Madrid, Spain) demonstrated a normal fundus in the right eye but revealed a focal lesion in the dorsal periphery of the tapetum of the left fundus. The lesion was round, nearly twice the size of the optic nerve head, pigmented and inconsistently circumscribed by a thin hyper-reflective rim. The surrounding retinal vessels showed an abnormal pattern (Fig. 1). The rest of the fundus was considered normal.

Imaging tests (fluorescein angiography and optical coherence tomography) and electroretinography were performed to characterize the lesion.

Full-field flash electroretinography (ERG) was recorded under general anesthesia using the Visio-System electrodiagnostic unit (Siem Bio Médicale, Nimes Cedex, France). Following premedication with a combination of medetomidine (Domtor®; Pfizer, Alcobendas, Spain) 0.01 mg/kg, and butorphanol (Butomidor®; Richter Pharma AG, Wells, Austria) 0.3 mg/kg intramuscularly, anesthesia was induced with intravenous propofol (Propofol®-Lipuro 1%; Braun Medical, Barcelona, Spain) and maintained with isoflurane 1.0% (IsoVet®; Piramal Healthcare, Northumberland, UK). The pupils were fully dilated with three drops of 1% tropicamide ophthalmic solution (Tropicamida; Alcon Cusí, Barcelona, Spain) within a 10-min interval. An ERG was performed according to the guidelines provided by the European College of Veterinary Ophthalmologists (ECVO). Electroretinogram responses in both eyes were within normal limits.

**Figure 1.** Atypical coloboma of the left eye. A focal, pigmented lesion dorsal to the optic nerve was appreciated. Small hyper-reflective areas circumscribed the defect and surrounding retinal vessels showed an abnormal pattern.

Following ERG recording, an optical coherence tomography (OCT) study was performed (3D 1000 Topcon Fourier Domain; Topcon España, Barcelona, Spain). All horizontal line scans revealed an important craterlike depression showing an atrophic neurosensory retina, and a lack of normal retinal pigment epithelium and choroidal tissue (Fig. 2a). The margin of the coloboma presented a humplike appearance due to the thickening of the retinal layers at this point (Fig. 2a). The defect was more severe in the dorso-temporal sector of the lesion where an abrupt disruption of the retina was appreciated with total absence of chorioretinal tissue (Fig. 2b). These images were consistent with a dorsal chorioretinal coloboma.

Finally, fluoroangiographic examination was performed after intravenous bolus injection of 10 mg/kg of fluorescein sodium solution (Fluoresceina Oculos 10%; Thea, Barcelona, Spain) through an 18-gauge catheter in the cephalic vein. Serial fundic photographs were taken with a fundus vision system (Visucam Lite; Carl Zeiss, Jena, Germany) from 4 s to 7 min at intervals of 1 s throughout the angiogram. Fluorescein angiography revealed a focal colobomatous lesion dorsal to the optic nerve and perfectly delimitated from the surrounding ocular fundus. During the choroidal flush, a defect in vascular filling was observed in this area and a constant hypofluorescence was maintained throughout all phases of the fluoroangiography (Fig. 3a). Choroidal impregnation was normal on the remaining ocular fundus completing itself in a homogeneous way at the end of the arterial phase. The arterial phase evidenced a linear hyperfluorescence surrounding the lesion which grew in intensity throughout the angiogram reaching its maximum during the late phase (Fig. 3a, 3d). Thin tortuous arteries were appreciated crossing the defect and deviating markedly from typical vascular patterns (Fig. 3b). During the arterial–venous phase, while some vessels converged into the coloboma, others appeared to emerge from the floor of the coloboma (Fig. 3c). Arterial-venous phase had an asynchronous delay during the filling of the veins that converged in the affected area as fluorescence was only observed during the late phase.

On re-examinations, 6 and 12 months later, direct and indirect ophthalmoscopy, and fluorescein angiography were performed not appreciating any significant changes in the lesion.

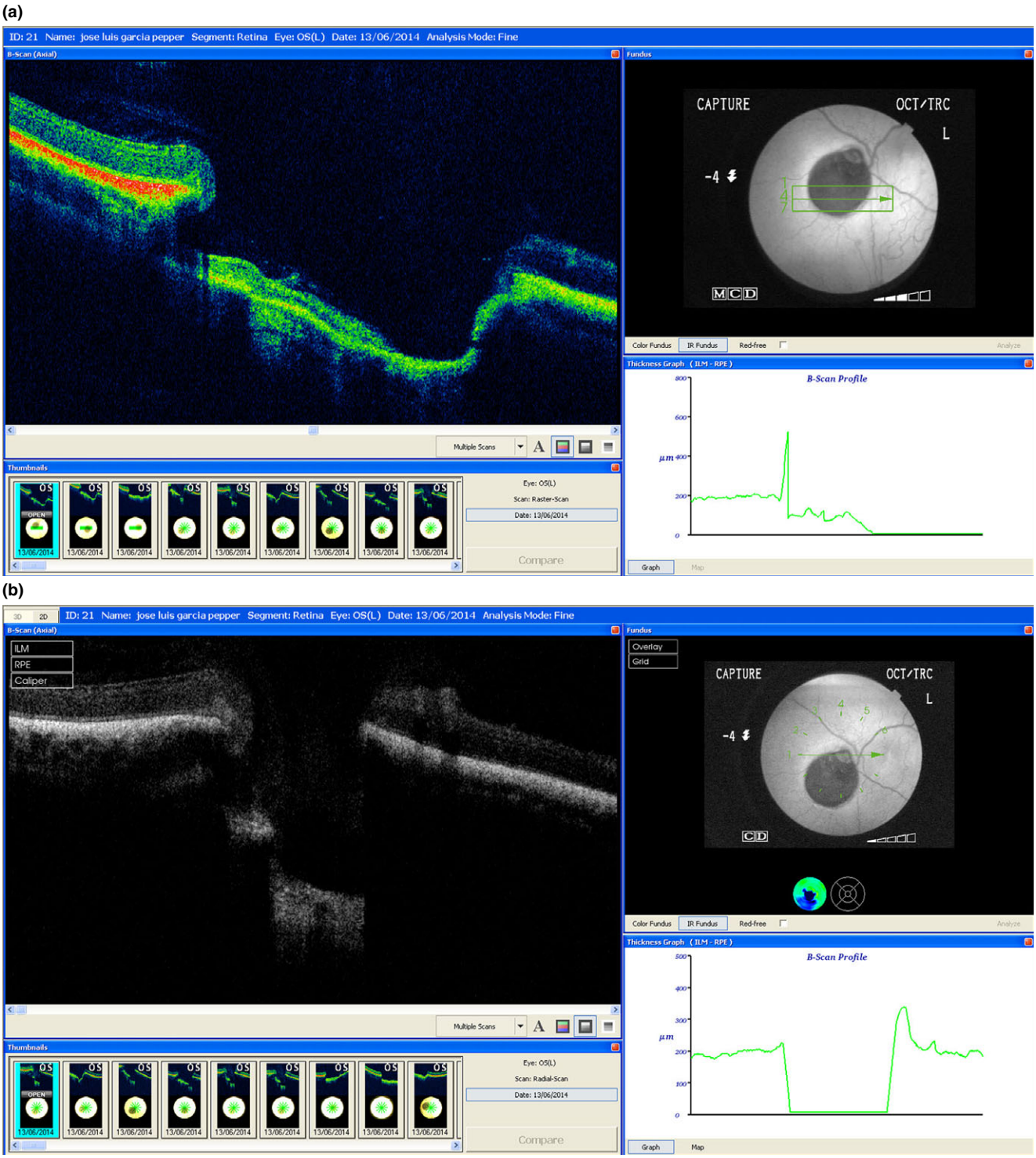


**Figure 2.** OCT image of the lesion revealing a craterlike depression with an atrophic neurosensory retina, and lack of normal retinal pigment epithelium and choroidal tissue; the margin of the defect showed a humplike appearance (a). An abrupt disruption of the retina with total absence of chorioretinal tissue was appreciated in the dorso-temporal sector (b).

## DISCUSSION

Atypical colobomas are infrequent. While typical colobomas, located in the inferonasal quadrant, are due to an incomplete closure of the embryonic fissure of the optical vesicle, atypical colobomas origin is not certain. They seem to be related to primary defects of the uveal epithelium (anterior segment colobomas) and/or retinal pigment epithelium (posterior segment colobomas).[1,5–7] Genetic mutations and environmental toxic factors have been

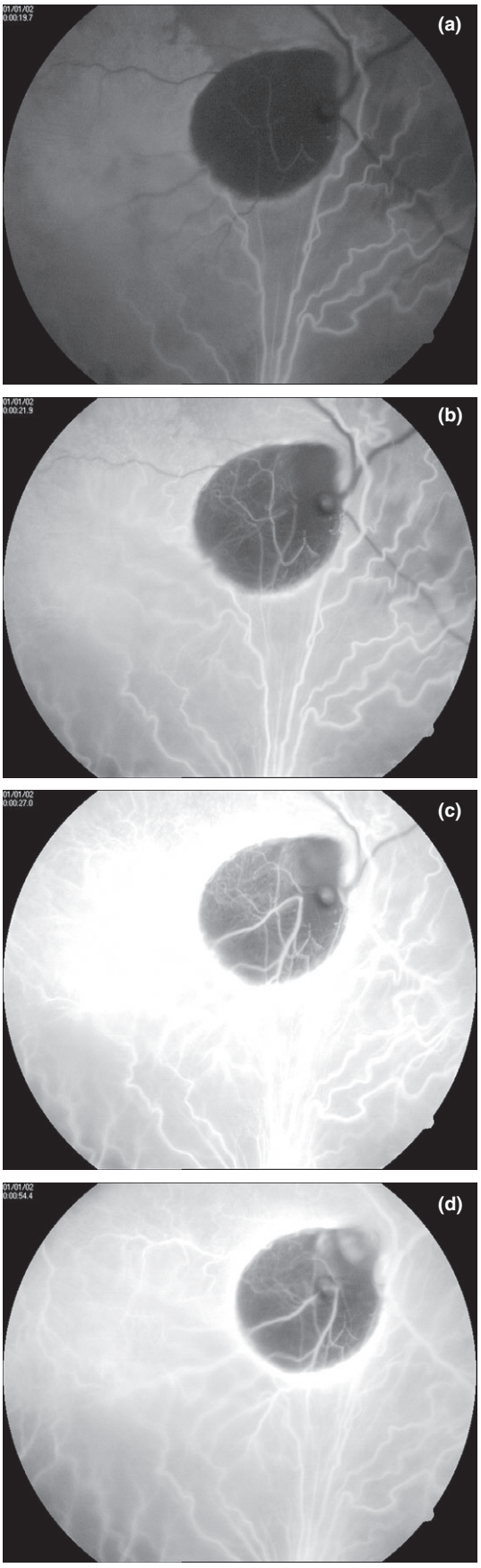


**Figure 3.** Fluoroangiography images showing the initial arterial phase (a) where a hypofluorescence of the defect was evidenced and maintained throughout all phases of the fluorescein angiography. A linear hyperfluorescence surrounding the lesion was appreciated at this phase. The arterial phase evidenced thin tortuous arteries crossing the defect and deviating markedly from typical vascular patterns (b). The arterial–venous phase (c) revealed complete vasculature hyperfluorescence. While some vessels converged into the defect, others appeared to emerge through the floor of the coloboma. During the late phase (d), the hyperfluorescence around the lesion reached its maximum intensity.

pointed out as possible causes of colobomas.[8] Histopathologically, they are circumscribed areas of nearly bare sclera that can lack normal retinal pigment epithelium, neurosensory retina, and choroid.[1,20] The tissue overlying the coloboma is an extension of the retina called the intercalary membrane.[21] The most common complications associated with these defects are retinal detachments and choroidal neovascularization.[22–24]

Normally, chorioretinal colobomas appear clinically as white lesions because the sclera is directly visualized. Less frequently, they can appear pigmented.[1] The sclera can be covered by a hypoplastic or dysplastic retina, and retinal vessels can be appreciated crossing the defect with an abnormal pattern.[9,17,20] Borders of the lesion are normally definable, and irregular clumps of pigment can be present along the rim.[1] In this dog, the lesion was located dorsal to the optic disk corresponding to an atypical location.[1,3,4] The coloboma was dark in appearance due to the presence of pigment, which probably implied the presence of some residual suprachoroid. A small rim of hyper-reflectivity was observed surrounding the lesion probably due to the thinning of the retinal layers in this area. Ophthalmoscopy examination did not reveal choroidal or retinal vessels crossing the coloboma, but the surrounding vessels showed a complete abnormal pattern.

Clinically, posterior segment colobomas can be totally asymptomatic or may be associated to vision loss depending on the size and location of the defect.[1] In this case, vision tests were normal for the affected eye even though the lesion is bigger in size than the optic nerve head. Probably, the peripheral dorsal location of the lesion, away from the optic disk, plays an important role in this fact.[25] Also no significant differences were appreciated in the electroretinographic responses of both eyes which demonstrated that the defect was not large enough to affect the global electrical activity of the retina. This lack of significant vision loss explains why in this case the chorioretinal coloboma was an incidental finding during a routine ophthalmoscopy examination.

OCT has proven a beneficial tool to confirm the diagnosis of colobomas.[21,26,27] OCT studies of posterior atypical colobomas in dogs have not been previously reported. OCT images obtained in this case agree with tomographic findings described in chorioretinal colobomas in humans.[21,25,26] The absence of tissue revealed as a

craterlike depression where an atrophic neurosensory retina was appreciated together with the absence of retinal pigment epithelium and choroidal tissue. The margin of the coloboma showed a humplike appearance due to inward turning of the retinochoroidal tissue together with thickening of the layers at this point.[21,26]

Fluorescein angiography has been performed in other species to evaluate the pattern of blood vessels in colobomas, but no previous studies have been reported in dogs.[9,14,20] The angiogram showed a complete altered vascular pattern associated with the coloboma. A constant hypofluorescence was present on the defect due to the lack of choroidal vasculature in the affected area. The absence of hyperfluorescence in the late phases of the angiogram indicates the lesion is unlikely a scar.[30] Thin, twisted arteries of the hypoplastic retina were appreciated crossing the lesion and deviating markedly from the normal anatomical patterns. Retinal veins appeared dilated and also showed an abnormal pattern, some emerging from the floor of the coloboma. The emergence of blood vessels into the fundus of colobomatous eyes and their abnormal pattern have also been described in humans and are presumably extraocular vessels.[20] In our opinion, this fact may explain the asynchrony in the filling of these vessels during the venous phase of the angiogram. Fluorescein angiographs in rats also revealed abnormalities in retinal vessels, with twisted arteries and congested and enlarged veins.[9] The arterial phase evidenced a linear hyperfluorescence surrounding the lesion which grew in intensity throughout the angiogram reaching its maximum intensity during the late phase. This image has also been described in colobomas in other species.[9,14,20] This area of abnormal choroidal vessels with altered permeability probably corresponds to the development of choroidal neovascularization.[9,14,24,28] It has been postulated that the discontinuity of Bruch's membrane at the margin of the coloboma allows aberrant growth and penetration of contiguous choroidal vessels into the subretinal space at its junction.[24,28,29,30]

## CONCLUSION

Atypical posterior colobomas are infrequent in dogs. This report describes for the first time OCT and fluorescein angiography studies in an atypical chorioretinal coloboma in a Golden Retriever demonstrating the importance of these two complementary diagnostic tools in the characterization of this type of congenital lesion. In this case, the coloboma was clinically asymptomatic; thus, we must point out the importance of routine ophthalmoscopic examination to diagnose this defect.

## REFERENCES


1. Onchowei BC, Simon JW, Bateman JB *et al.* Ocular colobomata. *Survey of Ophthalmology* 2000; **45**: 175–194.
2. Cook CS. Ocular embryology and congenital malformations. In: *Veterinary Ophthalmology*, Vol **I**., 5th edn. (ed. Gelatt KN) Wiley-Blackwell, Ames, 2013; 3–38.
3. Gelatt K, Huston K, Leipold H. Ocular anomalies of incomplete albino cattle: ophthalmoscopic examination. *American Journal of Veterinary Research* 1969; **30**: 1313–1316.
4. Gwin R, Wyman M, Lim D *et al.* Multiple ocular defects associated with partial albinism and deafness. *Journal of the American Animal Hospital Association* 1989; **17**: 401.
5. Cook C, Burling K, Nelson E. Embryogenesis of posterior segment colobomas in the Australian Shepherd dog. *Progress in Veterinary and Comparative Ophthalmology* 1991; **1**: 163–170.
6. Cook CS, Generoso WM, Hester D *et al.* RPE dysplasia with retinal duplication in a mutant mouse strain. *Experimental Eye Research* 1991; **52**: 409–415.
7. Zhao S, Overbeek PA. Regulation of choroid development by the retinal pigment epithelium. *Molecular Vision* 2001; **7**: 277–282.
8. Gregory-Evans CY, Wiliams MJ, Halford S *et al.* Ocular coloboma: a reassessment in the age of molecular neuroscience. *Journal of Medical Genetics* 2004; **41**: 881–891.
9. Ninomiya H, Kuno H, Inagaki S. Vascular changes associated with chorioretinal and optic nerve colobomas in rats (Crj: CD (SD), IGS). *Veterinary Ophthalmology* 2005; **8**: 319–323.
10. Matsuura T, Kodoma Y, Ozaki K *et al.* A newly established mutant strain with mild-type ocular coloboma (retinochoroidal coloboma without microphthalmia) in albino mice. *Birth Defects Research. Part A. Clinical and Molecular Teratology* 2006; **4**: 266–271.
11. Barnett KC, Lewis JCM. Multiple ocular colobomas in the snow leopard (*Uncia uncia*). *Veterinary Ophthalmology* 2002; **5**: 197–199.
12. Bildfell R, Watrous B, Maxwell S *et al.* Bilateral optic disc colobomas in a Quarter Horse filly. *Equine Veterinary Journal Supplement* 1990; **10**: 325–327.
13. Schuh JC. Bilateral colobomas involving the optics discs in a Quarterhorse. *Journal of Compendium Pathology* 1989; **10**: 50–54.
14. Martín-Suarez EM, Galan A, Gallardo JM *et al.* Bilateral typical complete colobomas in a donkey: retinographic and fluorangiographic description. *Veterinary Ophthalmology* 2009; **12**: 338–342.
15. Mc Comarck J. Typical colobomas in Charolais bulls. *Veterinary Medicine Small Animal Clinician* 1977; **72**: 1626–1628.
16. Martin CL, Stiles J, Willis M. Feline colobomatous syndrome. *Veterinary & Comparative Ophthalmology* 1997; **7**: 39–43.
17. Mathews AG, Crispin SM, Parker J. The equine fundus. II: normal anatomical variants and colobomata. *Equine Veterinary Journal Supplement* 1990; **10**: 50–54.
18. Briziarelli G, Abrutyn D. Atypical coloboma in the optic disc of a Beagle. *Journal of Comparative Pathology* 1975; **85**: 237–240.
19. Rubin LF. Atypical coloboma of the choroid in a dog. *Journal of the American Veterinary Medical Association* 1963; **143**: 841–842.
20. Gopal L. Pattern of blood vessels in eyes with coloboma. *Indian Journal of Ophthalmology* 2013; **61**: 743–748.
21. Gopal L, Khan B, Jain S *et al.* A clinical and optical coherence tomography study of the margins of choroidal colobomas. *Ophthalmology* 2007; **114**: 571–580.
22. Schubert HD. Structural organization of choroidal colobomas of young and adult patients and mechanism of retinal detachment. *Transactions of the American Ophthalmological Society* 2005; **103**: 457–472.
23. Gelisken F. Choroidal neovascularization in retinochoroidal coloboma: thermal laser treatment achieves long-term stabilization of visual acuity. *Eye* 2006; **20**: 969–972.
24. Guirgis MF, Lueder GT. Choroidal neovascular membrane associated with optic nerve coloboma in a patient with CHARGE association. *American Journal of Ophthalmology* 2003; **6**: 919–920.
25. Barnard S, Shneor E, Brauner J *et al.* Bilateral chorioretinal coloboma discovered with ultra-wide field retinal imaging. *Journal of Optometry* 2012; **5**: 150–154.

26. Gopal L. A clinical and optical coherence tomography study of choroidal colobomas. *Current Opinion in Ophthalmology* 2008; **19**: 248–254.
27. Oh JY, Yu YS, Hwang JM *et al.* Optical coherence tomography finding in a case of macular coloboma. *Korean Journal of Ophthalmology* 2007; **21**: 174–177.
28. Naithani P, Vashisht N, Mandal S *et al.* Intravitreal bevacizumab in choroidal neovascularization associated with congenital choroidal and optic nerve coloboma in children: long-term improvement in visual acuity. *Journal of American Association for Pediatric Ophthalmology and Strabismus* 2010; **14**: 288–290.
29. Goodwin P, Shields CL, Ramasubramanian A *et al.* Ranibizumab for coloboma-related choroidal neovascular membrane in a child. *Journal of American Association for Pediatric Ophthalmology and Strabismus* 2009; **13**: 616–617.
30. Lescure F. Choroid. In: *Fluorescein Angiographic Atlas of the Small Animal Ocular Fundus*. (ed. Lescure, F) Pratique Médicale et Chirurgicale de l'Animal de Compagnie, Paris, 1998; 189–208.