

SHORT COMMUNICATION

Multiple ocular colobomas in the snow leopard (*Uncia uncia*)

K. C. Barnett* and J. C. M. Lewis†

*Animal Health Trust, Center for Small Animal Studies, Lanwades Park, Kentford, Newmarket, Suffolk CB8 7UU, UK, †International Zoo Veterinary Group, Keighley Business Center, South Street, Keighley, West Yorkshire BD21 1AG, UK

Address communications to:

K. C. Barnett

Tel.: +44 (0)1638 552700

Fax.: +44 (0)1638 555600

e-mail:

smallanimal.centre@aht.org.uk

Abstract

Two singleton female snow leopard cubs are reported with bilateral central upper lid colobomas. In addition, one cub had a coloboma of the fundus in one eye extending from the lower optic disc region. Surgical treatment by wedge resection was successful in both cases. Details of ocular colobomas in other snow leopards reported in the literature are described and it is suggested that the exact etiology of the condition in this species may be discovered by further study of similar colobomas in the domestic cat.

Key Words: coloboma, eyelid, ocular fundus, snow leopard

INTRODUCTION

A coloboma is a congenital malformation in which a portion of the structure of the eye is lacking. These lesions occur early in gestation and result from failure of the embryonic fissure to close. Colobomas are common congenital anomalies and are usually bilateral, but like many congenital anomalies show wide variation in their degree of severity. Typical colobomas are located infero-nasally in the region of the fissure; atypical colobomas occur elsewhere in the eye. The iris and uveal tract are usually involved, but often more than one ocular structure is affected. Coloboma and microphthalmos often occur together.

Colobomas occur in the eyes of humans and several animal species. They may be inherited, for example, as part of the Collie Eye Anomaly in such breeds as the Rough Collie and Shetland Sheepdog and as posterior segment colobomas in Charolais cattle. They may also be due to teratogenic agents including environmental factors, deficiencies, etc.

Colobomas also occur as part of several syndromes. Multiple ocular colobomas (MOC) in snow leopards, including microphthalmos in one eye and eyelid deformity in both eyes, were first described in a captive-bred male cub born in 1976 in Helsinki.¹ The dam was also mildly affected; the cub was blind in spite of surgical treatment and was destroyed. Further cases have been recorded in Helsinki and also from zoos in Amsterdam and Zurich,² from the Henry Doorly zoo in Omaha,³ and from zoos in Jersey⁴ and Munich.⁵ No cases have yet been described in wild-born snow leopards. This short communication records two further cases in the UK and their successful treatment.

Case histories

The first case was a 6-month-old female singleton with a central coloboma of both upper eyelids, but with no other ocular abnormalities apparent on detailed ophthalmoscopic examination under anesthesia. The sire of this animal was approximately 8.5 years old at the time of the cub's conception, and the dam approximately 6.5 years old. The affected animal and both parents had inbreeding coefficients of 0 (the inbreeding coefficient is the probability of an animal receiving the same allele from both parents, i.e. a measure of the degree to which an animal is inbred), and the parents had produced two normal litters previously, each of a single male. Case two was another singleton female cub born at a different collection, aged 8 months when treated, again with central upper lid colobomas. The sire of this animal was approximately 10.5 years old at the time of the cub's conception, and the dam approximately 11.5 years old. The sire had an inbreeding coefficient of 12.5%, the dam 0, and the affected cub 0.87%. This pair had produced two normal previous litters of two and three cubs, respectively. In case two, the right eye (Fig. 1) was more severely affected than the left eye (Fig. 2) with approximately one third of the eyelid edge completely absent and the defect continuing upwards into the periorbital region. In this area the palpebral conjunctiva was reflected directly onto the globe eliminating any fornix and allowing the adjacent hairs of the lid to irritate the cornea and conjunctiva (trichiasis) producing a localized vascular and pigmented keratitis. On detailed ophthalmoscopic examination, there was a coloboma affecting the lower optic disc region, extending several disc diameters below the disc at the 6 o'clock position, in which choroidal vessels were



Figure 1. Central upper lid coloboma. Note absence of involvement with either canthus.



Figure 2. Central upper lid coloboma. Less extensive area than other eye.

visible. The left eye had a similar central upper lid coloboma but less extensive than in the right eye. Case two presented with severe blepharospasm. Ophthalmoscopic examination of the four parent leopards under general anesthesia failed to reveal any evidence of MOC.

Both cubs were treated under general anesthesia by bilateral simple wedge resection removing the whole coloboma and extending upward well into the upper eyelid. In the most severely affected case (right eye, case two) at least one third of the upper eyelid was removed. Repair was effected with simple, interrupted 5/0 and 6/0 vicryl sutures in two layers through palpebral conjunctiva and eyelid edge and skin. Following surgery one application of an antibiotic and lubricant eye ointment was applied. Healing was by first intention and on recovery from anesthesia blepharospasm was noticeably improved. Neither cub showed any sign of postsurgical self-inflicted injury despite the removal of considerable amounts of eyelid edge and consequent shortening of the lid which showed no propensity to entropion. No doubt the successful long-term result of the surgery was aided by the young age of the cubs at the time of operation and postoperative stretching of the remainder of the upper lids.

DISCUSSION

The common features in all the cases of coloboma in the snow leopard is involvement of the upper lid in one or both eyes, central or lateral, accompanied by colobomas affecting various parts of the globe, e.g. iris or fundus. Colobomatous defects include persistent pupillary membranes and, in the severest form, microphthalmos, and both have been recorded in both snow leopard and domestic cat, as has retinal dysplasia (retinal folds), often seen in conjunction with degrees of microphthalmos. This paper would seem to be the first report of coloboma affecting the central part of the lid with normal medial and lateral canthi, the lateral (temporal) lid being more commonly affected. The lower lid is never involved. The combination of colobomas affecting both globe and lid has only been described in the domestic cat and

the snow leopard and the relationship between the two is not understood. The globe develops early in gestation and from neuro-ectoderm but the lid develops from mesoderm and much later in gestation.

The discovery of the exact etiology of MOC in captive snow leopards may well result from further study of the domestic cat where absence of part of the upper lid, sometimes referred to as agenesis, is not a rare condition. Martin *et al.* recently described MOC in four out of five Domestic Short-haired tabby kittens, the remaining kitten and both parents being unavailable for examination.⁶ Lesions varied from microphthalmos and blindness to minor abnormality of the lateral lid margin. This was the first report in a litter although previously Bellhorn *et al.* described seven isolated cases of ocular coloboma in the domestic cat including pupillary membranes and posterior segment coloboma but not all of which showed eyelid agenesis.⁷ In MOC the presenting sign is usually agenesis and is therefore obvious, but it is important that all members of a litter, and wherever possible their parents, undergo detailed ophthalmoscopic examination to reveal posterior segment colobomata, which are not always accompanied by eyelid involvement.

It is crucial to find the etiology of MOC in the snow leopard, as endangered species with limited gene pools are always theoretically prone to genetic abnormalities. Colobomas may or may not be inherited. The first case described was in a male cub severely affected and blind, whose dam was mildly affected and had been mated to her sire; other cubs from this mating were normal.¹ In the USA, three of four cubs from two litters, born to the same sire and dam were affected, although the same dam mated to an unrelated sire produced two normal cubs.³ These facts suggest possible inheritance. However, eyelid colobomas, particularly those affecting the upper lid, are rarely inherited in humans and a detailed analysis of 16 cases of MOC in snow leopards of the Helsinki line failed to reveal evidence of a single deleterious gene transmitted according to simple genetic rules.⁸ It seems therefore that a genetic etiology is unlikely. The teratogenic effect of a feline virus has been suggested as an alternative

cause, as have both hyper- and hypovitaminosis A and a deficiency of folic acid.⁵ However, no convincing evidence supporting these suggestions has been forthcoming.

The two cases reported here do not provide any new theory as to the cause of the condition, but they do raise again the possibility of inheritance. Although the two sires were unrelated either to each other or to the dams, the latter were litter sisters. Housed at different UK collections and provided with different diets supplemented with different levels of vitamin A, none of the affected cubs' parents were known to be affected by any viral infection either before or during the pregnancies, and only inactivated vaccines had been used in both collections. It is extremely unlikely that the two dams were exposed to the same teratogenic toxin during pregnancy. No other cases of ocular coloboma have been reported from this line, but many animals are lost to follow-up and very few have undergone detailed ophthalmoscopic examination. The global captive snow leopard population has been skilfully managed resulting in a relatively low mean inbreeding coefficient (2.4% in 1996). However, in light of the cases reported in this paper, perhaps the possibility of a genetic explanation for MOC in snow leopards should be re-examined, and caution exercised in breeding from affected animals.⁹ It is likely, however, that attempts to demonstrate or eliminate a genetic basis for the condition will be severely hampered until all captive-bred snow leopards are subjected to detailed ophthalmoscopic examination as a matter of course, whether or not they are exhibiting overt clinical signs.

The importance of studies in the domestic cat, including the detailed ophthalmoscopic examination of whole litters and both parents, wherever possible, may assist in discovering the etiology of this similar but unusual syndrome in the snow leopard. The combination of peri-ocular (eyelid), anterior segment (pupillary membrane) and posterior segment (optic disc and fundus) colobomas would seem to be unique to the

cat family and in particular to the Domestic Short-hair and snow leopard. There must be an explanation as to how these various colobomas may be connected and due to the same cause. The anomalies in the domestic cat, although not rare, are of little significance, but they are important in the snow leopard, particularly if a genetic component is involved.

ACKNOWLEDGMENTS

We would like to acknowledge Clive Barwick and Dave Fowler for their skilled animal husbandry.

REFERENCES

1. Wahlberg C. A case of multiple ocular coloboma in a snow leopard. *International Pedigree Book of Snow Leopards*, Vol. 1, 1978.
2. Wahlberg C, Tarkkanen A. On the multiple ocular coloboma with retinal dysplasia (MOC) in snow leopards, *Panthera uncia*. *International Pedigree Book of Snow Leopards*, Vol. 2, 1980.
3. Phillips LG. Palpebral coloboma in three snow leopards. *Annual Proceedings of the American Association of Zoo Veterinarians*. Seattle, 1981.
4. Barnett KC. Ocular colobomata in the snow leopard *Panthera uncia*. *Dodo, Journal of Jersey Wildlife Preservation Trust* 1981; **18**: 83–85.
5. Schäffer E, Wiesner H, von Hegel G. Multiple okuläre Kolobome (MOC) mit persistierender Pupillarmembran beim Schneeleopard (*Panthera uncia*) *Tierärztliche Praxis* 1988; **16**: 87–91.
6. Martin CL, Stiles J, Willis M. Feline colobomatous syndrome. *Veterinary and Comparative Ophthalmology* 1997; **7** (1): 39–43.
7. Bellhorn RW, Barnett KC, Henkind P. Ocular colobomas in domestic cats. *Journal of the American Veterinary Medical Association* 1971; **159** (8): 1015–1021.
8. Gripenberg U, Blomqvist L, Pamilo P *et al*. Multiple ocular coloboma (MOC) in snow leopards (*Panthera uncia*) *Hereditas* 1985; **103**: 221–229.
9. Blomqvist L. Analysis of the global captive snow leopard, *Uncia uncia*, population in 1996. In: *International Pedigree Book of Snow Leopards, Uncia uncia*, Vol. 7. Helsinki Zoo, Helsinki, 1998; 6–20.