CASE REPORT

A Rare Ocular Anomaly of Congenital Bilateral Aphakia in a Goat Kid - A Case Report

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Ocular defects in food-producing livestock have a significant impact on the economy of the rearing community. Every year there is an increase in the number of food-producing animals and reports of congenital anomalies become evident in the total number of animals affected per year (Williams, 2010). Reports on ocular anomalies in ruminants that too in the eyes of sheep and goats are very limited (Sangamwar *et al.,* 1997). Aphakia means absence of lens. Congenital aphakia is an exceptionally rare anomaly that is reported only in very few livestock. This anomaly is either due to the failed induction of lens placode or due to the resorption or expulsion of the lens *in utero* after normal development (Gelatt and Plummer, 2022). Aphakia is commonly associated with anterior segment dysgenesis. The diagnosis can be done with slit lamp biomicroscopy, retinoscopy and ultrasonography. Enucleation is one of the choices of treatment in livestock (Sato *et al.,* 2017).

Case History and Observations

A three-month-old, non-descript female goat kid was presented to the Veterinary Clinical Complex, Veterinary College and Research Institute, Orathanadu (India) with a history of impaired vision from birth onwards. The animal had normal voiding habits with a good appetite, but it could feed and suckle milk only when guided towards the teats.

On clinical examination, the body temperature was normal, and the conjunctival mucous membrane was pink and moist. Macroscopically, there seemed to be bilateral corneal opacity (Fig. 1). On ophthalmic examination, palpebral reflex could be appreciated but not the menace and pupillary light reflexes in both the eyes. The lens was absent in both eyes (Fig. 2), which was confirmed with slit lamp biomicroscopy. The animal also had an elevated response to normal auditory stimuli and navigated its surroundings only by utilizing the auditory information. Based on these ophthalmic findings, this condition was diagnosed as blindness due to 'Bilateral Congenital Aphakia'.

Treatment and Discussion

This kid was born with a female co-twin to a first kidder goat, and it was normal except congenital bilateral aphakia. The animal was supplemented with Inj. Vitamin AD_3E 0.3 mL i/m,Inj. Thiamine hydrochloride 0.3 mL i/m, and oral minerals. The prognosis was explained to the owner and advised for enucleation. The owner was unwilling to enucleation and instead chose to raise the animal without any treatment.

The lens is a dynamic organ that can adjust the refractive power of the eye to facilitate accommodation, enabling the eye to focus the light on the retina. The lens is an avascular structure, so that it looks clear macroscopically (Sakti, 2021). Congenital aphakia or the absence of crystalline lenses in the eyes is an infrequent condition in animals, which may be primary or secondary. Primary aphakia is associated with the failure of lens development due to the failure of induction of lens placode which happens after the optic vesicle makes contact with the surface ectoderm and generally occurs in combination with the anterior segment dysgenesis or microphthalmos (Gelatt and Plummer, 2022), whereas in secondary aphakia, normal development of lens takes place, but it is resorbed, destructed or extruded earlier *in utero* (Dubielzig *et al.,* 2010). Sangamwar *et al.* (1997) noticed the unilateral absence of lens in a sheep and a goat, in a study. In both cases, the lens got dissolved following inflammation in the eye which was indicated by mononuclear cell infiltration in the choroid and optic nerve fiber layer which made it thickened. Further in the same study, the authors reported that cases of aphakia in both sheep and goats were not

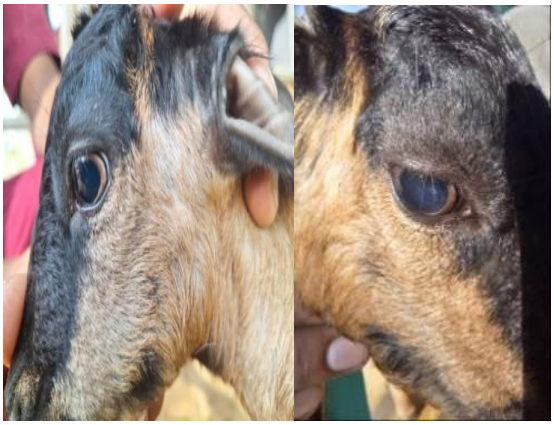

Fig. 1: Macroscopic appearance of aphakic eyes

Fig. 2: Opaque appearance of cornea

traceable in the available literature. In this present case, in both eyes, the lens was absent.

In experimental animals like mice, the arrest in the development of lens is found to be due to the double deletion mutation of the *Pitx3* gene (Rieger *et al.,* 2001) or the altered gene expressions involving *Pax6* and *Six3* genes (Grimm *et al.,* 1998). Lens development starts with the thickening of the surface ectoderm to form a lens placode, which invaginates to form a lens vesicle. The lens vesicle contains a proliferative lens epithelium anteriorly and a layer posteriorly which differentiates from primary fiber cells. Later, the anterior epithelial cells migrate towards the equator and differentiate into secondary lens fibers. This differentiation is associated with the expression of the *Foxe3* gene which is directly regulated by the *Pitx3* gene (Ahmad *et al.,* 2013). Hence, a lack of expression or mutation in these genes can affect lens development.

Since the development of the optic cup is influenced by the embryonic lens, aphakia is also associated with multiple ocular abnormalities like microphthalmia, anterior segment dysgenesis, Peters' anomaly, retinal dysplasia, retinal detachment and congenital staphylomas (Sato *et al.,* 2017; Gelatt and Plummer, 2022). In the present case, aphakia was not associated with any other ocular abnormalities.

A complete ophthalmic examination is needed to diagnose aphakia. The absence of lens results in a severe hypermetropic eye, where the image is formed behind the retina. This refractive status of the aphakic eye can be assessed using retinoscopy (Davidson *et al.,* 1993). A slit lamp biomicroscope can be used for examining the defects in the anterior segment of the eye. The anterior chamber is found to be deeper than normal in the aphakic eyes (Sakti, 2021). In this case, the anterior chamber was deep, and no lens could be visualized in the optical pathway of eyes through the slit lamp biomicroscope. Aphakia and other associated ocular abnormalities can also be diagnosed with ultrasonography with a 10 MHz micro-convex probe, where it is characterized by the absence of the hyper-echoic posterior lens capsule (Ragab and Fathy, 2018).

Enucleation is the treatment of choice as there may be complications of secondary glaucoma (Kugelberg *et al.,* 2000) and retinal detachment without a good visual prognosis (Sato *et al.,* 2017). An experimental study in neonatal monkeys suggested both intraocular lenses (IOLs) and contact lenses are viable options for the management of aphakia (Boothe *et al.,* 2000). Refractive keratoplastic techniques like epikeratophakia, radial keratotomy and implantation of intralamellar lenticules in the corneal stroma (keratophakia) have been demonstrated in laboratory animals such as primates, cats and dogs. Among these procedures, epikeratophakia is a minimally invasive and refractive surgical procedure where the lamella of the donor cornea is transplanted to the anterior surface of the recipient cornea (Munger *et al.*, 2013). However, they are developed for human use and have not been widely adopted in veterinary practices.

In summary, bilateral congenital aphakia, a very rare ocular anomaly in caprine species is reported for the first time. In human beings, sophisticated techniques are being used to diagnose this condition, but those procedures are used very limitedly in livestock. Further, molecular studies are warranted to identify the genes responsible for congenital aphakia in livestock.

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