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Multiple congenital ocular abnormalities (MCOA) in Rocky Mountain Horses and Kentucky Mountain Saddle Horses in Europe

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Summary

The study describes the prevalence of multiple congenital ocular abnormalities (MCOA) in Rocky Mountain Horses and Kentucky Moutain Saddle Horses in Europe. Materials and methods: 35 RMH und KMSH were examined between 1999 and 2010. Their coat color were chocolate (24), seal brown (7), and one each of bay, black, chestnut and palomino. Ciliary body cysts (CBC) were found in 17/35 horses. Two (2/35) horses had multiple congenital ocular abnormalities consistent with anterior segment dysgenesis (ASD). None of the seal brown, black or bay horses had ocular abnormalities, while 18/24 chocolate horses had lesions. One chestnut mare had also bilateral CBC. Fourteen horses (40%) were unaffected. While the prevalence of multiple ocular abnormalities appears to be lower in Europe than in the US, the prevalence of ciliary body cysts is almost the same. Prior to breeding, RMH und KMSH should be carefully examined for the presence of CBC and MCOA.

Keywords: Mountain horse, eye, congenital anomaly, anterior segment dysgenesis

Multiple kongenitale okuläre Anomalien beim Rocky Mountain Horse und beim Kentucky Mountain Saddle Horse in Europa

Die Studie beschreibt das Auftreten multipler kongenitaler okulärer Anomalien und ihre Prävalenz bei Rocky Mountain Horses (RMH) und Kentucky Mountain Saddle Horses (KMSH) in Europa. 35 RMH und KMSH wurde zwischen 1999 und 2010 ophthalmologisch untersucht. Die Verteilung der Fellfarben war Chocolate (24) seal brown (schwarzbraun) (7), sowie je ein Brauner, Rappe, Fuchs und Palomino. Ziliarkörperzysten wurden bei 17/35 Pferden gefunden. Zwei Pferde hatten multiple kongenitale okuläre Anomalien. Keiner der Braunen, Schwarzbraunen oder Rappen zeigte Augenveränderungen. Hingegen hatten 18/24 der Chocolate, sowie eine Fuchsstute Veränderungen. Vierzehn (40%) der untersuchten Pferde zeigten keine Veränderungen. Während die Prävalenz multipler kongenitaler okulärer Anomalien geringer zu sein scheint, verglichen mit den Zahlen aus den USA, ist die Prävalenz der Ziliarkörperzysten vergleichbar. Vor einer Zuchtverwendung sollten RMH und KMSH auf Ziliarkörperzysten und multiple kongenitale okuläre Anomalien untersucht werden.

Schlüsselwörter: Mountain horse, Auge, kongenitale Anomalie, Dysplasie Vordersegment

Introduction

The term Mountain Horse is a collective name for three closely related gait horse breeds, the Rocky Mountain Horse (RMH), the Kentucky Mountain Saddle Horse (KMSH), and the Mountain Pleasure Horse (MPH). Basic characteristics are medium-sized (14 to 16 hands) horses with gentle temperament, an easy ambling four beat gait and a solid body color (Fig. 1). In the Mountain horse breeds the coat color "chocolate" with white or flaxen mane and tail is very popular and has become strongly associated with this horse although any solid body color is accepted with no white marks above the knee or hock except for facial markings. Mountain Horses derived originally from Eastern Kentucky, US and are used today as pleasure horses, as well as for trail, competitive or endurance riding. In 1986 the American Rocky Mountain Horse Association was founded and a registry was established which has shown a steady and well-regulated growth in the number of horses registered. Today, around 40 animals are estimated to live in Europe, mainly in France, Germany and England.

Congenital ocular abnormalities in 514 Rocky Mountain Horses (RMH) in the US were described in detail. The Rocky Mountain Horse breed association, which was proactive in determining whether these abnormalities were a severe defect, supported this study. As a consequence mandatory eye exams for breeding animals of those breeds mentioned above were installed. As some of the ophthalmic lesions are of small size and of subtle nature these examinations have to be performed by a board certified ophthalmologist (ACVO) and filled out on eye examination forms provided by the Equine Eye Registration Foundation (EERF) located at Purdue University.

As their popularity increased, the first Mountain Horses were imported to Europe from the US. Because of the low number of breeding animals in Europe, ocular health has received great attention. The purpose of this article is the description and evaluation of the prevalence of congenital ocular anomalies in RMH and KMSH in Europe.

Materials and methods

A retrospective study of the EERF sheets of all examined Mountain Horses (RMH and KMSH) between November 1999 and March 2010 was performed. Included were horses that were certified by the breed registry and their progeny that

were deemed certifiable by the breed registry based on gait, temperament, conformation and parentage. The animals were examined ophthalmologically by slit lamp biomicroscopy (Kowa SL-14, Eickemeyer, Tuttlingen, Germany) and indirect ophthalmoscopy (Heine Omega 100, Eickemeyer, Tuttlingen, Germany) with a 20 diopters condensing lens in a darkened stable before and after dilation of the pupil with 1%tropicamide (Mydriaticum Novartis Ophthalmics, Switzerland). All examinations were performed by the same ophthalmologist (BMS). Eye examinations in Mountain Horses in Europe have to be performed by an ACVO diplomate to be accepted in the US.

Fig. 1 Typical representative of a Rocky Mountain horse with dark chocolate coat color and white mane and tail.

Fig. 2 A large translucent cystic structure (ciliary body cyst) arising from the temporal part of the ciliary body is extending into the vitreous cavity.

The horses included 23 RMH and 5 KMSH and 7 KMSH/RMH (registered with both breed associations). There were 13 males and 22 females. Their age at the time of examination ranged from 2 months to 10 years. Among these 35 animals 24 had a chocolate coat color, 7 horses were seal brown, 1 was a bay horse, 1 horse was palomino-colored, one horse was a chestnut with flaxen mane and tail, and 1 horse was black (Table 1).

Results

Ciliary body cysts (CBC) were the most frequently detected lesions (17/35 horses) (Fig. 2). They were always located temporally and were bilateral in 11/17 horses, involved the left eye in 4 and the right eye in 2 horses.

Retinal dysplasia was found in 1/35 animal in combination with CBC.

Multiple ocular anomalies were detected in 2/35 horses. In these horses a telecanthus and exophthalmos were obvious. On close examination cornea globosa, deep anterior chamber (Fig.3), miotic and dyscoric pupil with flattened, circumferentially oriented granula iridica at the pupillary ruff, iris hypoplasia, and goniosynechiae were evident (Fig. 4). These two

Fig. 3 Close-up of a horse with multiple ocular abnormalities: a cornea globosa, a deep anterior chamber (© Dr. D. T. Ramsey)

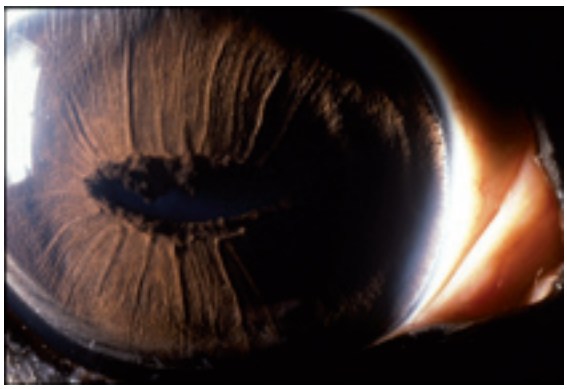

Fig. 4 Miotic and dyscoric pupil with flattened, circumferentially oriented granula iridica at the pupillary ruff and iris hypoplasia(© Dr. D. T. Ramsey).

horses did not show CBC. Two horses showed unilateral focal cortical cataracts but no signs of ASD (Table 1).

In 14/35 horses no ocular abnormalities were found. None of the seal brown, black or bay horses had ocular lesions, while 18 of the 24 chocolate colored horses (75%) had lesions.

Discussion

In horses congenital ocular defects are rare and constitute 0,5 – 5,3% of the collective congenital defects . Very often the exact cause is not determined. Only a few studies have reported results of ophthalmic examinations of large numbers of horses of a specific breed . In 1999 Ramsey et al. described congenital ocular abnormalities in 514 Rocky Mountain horses after the detection of a high prevalence of ophthalmic lesions in that breed.

Table 1 Relevant data of the 35 RMH and KMSH

	Breed	Birthdate	Coat color	Gender	Diagnosis
1	RMH	01.01.08	Chocolate	f	CBC OS
2	RMH	13.06.01	Chocolate	m	normal
3	RMH	26.06.01	Chocolate	m	normal
4	KMSH/RMH	07.06.01	Chocolate	f	CBC OS
5	KMSH/RMH	12.06.93	Chocolate	f	CBC OU, RD
6	KMSH	01.08.91	Chestnut	f	CBC OU
7	KMSH	01.06.95	Dark Chocolate	f	normal
8	KMSH	15.03.00	Bay	m	normal
9	KMSH/RMH	23.04.00	Palomino	m	MC,M,EGI,G, C, OU
10	KMSH	22.05.00	Chocolate	m	CBC OU
11	RMH	17.05.07	Chocolate	f	CBC OD
12	RMH	20.05.07	Black	f	normal
13	RMH	04.06.07	Chocolate	f	CBC OS
14	RMH	25.05.07	Chocolate	m	CBC OU
15	RMH	10.05.05	Seal brown	f	normal
16	RMH	17.05.05	Chocolate	f	CBC OU
17	RMH	05.05.05	Chocolate	m	CBC OD
18	RMH	30.04.05	Seal brown	f	normal
19	RMH	14.05.03	Seal brown	m	normal
20	RMH	17.05.03	Seal brown	f	normal
21	RMH	12.05.03	Chocloate	f	CBC OU
22	RMH	23.05.03	Chocolate	f	normal
23	RMH	17.05.02	Seal brown	m	C OD
24	KMSH/RMH	24.04.99	Dark chocolate	f	CBC OU
25	KMSH/RMH	22.05.01	Dark chocolate	m	normal
26	RMH	18.03.99	Chocolate	m	MC, M, EGI, G, OU
27	KMSH/RMH	29.05.01	Seal brown	f	normal
28	RMH	29.04.95	Chocolate	f	CBC OU
29	RMH	14.04.03	Chocolate	f	CBC OS
30	RMH	28.05.99	Chocolate	f	CBC OU
31	RMH	04.04.02	Dark chocolate	f	C OS
32	RMH	11.04.99	Chocolate	f	CBC OS
33	RMH	02.03.03	Seal brown	m	normal
34	KMSH	04.05.03	Chocolate	m	CBC OU
35	KMSH/RMH	28.05.01	Dark chocolate	f	normal

OD right eye, OS left eye, OU both eyes, CBC ciliary body cyst, MC megalocornea, M miosis, EGI encrircling granulae iridicae, G goniosynechiae, C cataract, RD retinal dysplasia

Large, translucent cystic structures arising from the posterior surface of the iris, ciliary body or peripheral retina were the most frequently detected abnormality (48%), which is similar to our findings. These cysts were only present in the temporal part of the eye and extended into the vitreous. The size of the cysts varies between 2-20 mm in diameter. In most cases the cysts occur bilaterally.

The second most common abnormality (37%) reported by Ramsey et al. were single to multiple well-delineated, darkly pigmented curvilinear streaks of the retinal pigmented epithelium in the peripheral tapetal fundus. These curvilinear streaks had their origin and ending near the temporal part of the retina and always extended towards the optic papilla. The streaks were mainly bilateral but not symmetrical and were only detected in eyes of horses with cysts of the iris, ciliary body or peripheral parts of the temporal retina. These findings are in contrast to ours, as we did not observe curvilinear streaks in any of the examined Mountain horses.

Retinal dysplasia was the third most common (24%) ocular abnormality detected by Ramsey et al. Most frequently a dysplasia of the temporal part of the peripheral retina was detected, which was characterized clinically as linear folds or vermiform streaks. Retinal dysplasia was most frequently bilateral and was detected only in eyes of horses with ciliary body or peripheral temporal retinal cysts. In our study retinal dysplasia was found only in one horse presenting clinically with unilateral linear folds, which were also associated with CBC.

In 14% of the horse population studied by Ramsey et al. a bilateral syndrome of multiple congenital ocular abnormalities involving the cornea, nasal and temporal iridocorneal angle, iris, lens, ciliary body and peripheral retina was observed. All horses with multiple ocular abnormalities also had cysts of the iris, ciliary body, or peripheral retina, retinal dysplasia, retinal detachment, and curvilinear streaks of the retinal pigmented epithelium. Abnormalities of the iris inclu-

ded miosis, dyscoria, stromal hypoplasia of the anterior and peripheral iris with occasional transillumination defects, absence of a discernible iris collarette, and a visible sphincter pupillae muscle. Radially oriented deep stromal strands of iris tissue extended from the papillary ruff towards the ciliary zone of the iris. The granula iridica were flattened and circumferentially oriented at the pupillary ruff when compared with the iris of a normal RMH. Typically, pupillary light responses were decreased or absent in horses with iris abnormalities and the pupils failed to dilate with 1% tropicamide. According to Ramsey et al. repeated topical administration of 1% tropicamide, 10% phenylephrine or 1% atropine did not induce mydriasis or had minimal to no effect on the diameter of the pupils. Megalocornea was found in 60% of horses with MCOA and was always bilateral. The diagnosis of megalocornea was made when a clear cornea with unremarkable central corneal topography, grossly observable excessively large optical diameter of the cornea with a short radius of curvature and notably globular contour of the cornea with atypical protrusion was seen in comparison to eyes of horses with normal-appearing corneas. Generally, horses with megalocornea had excessively deep anterior chambers with a normal appearing lens-iris diaphragm. Concurrently, most horses with megalocornea had macropalpebral fissures. Besides, in 91% of horses with MCOA different degrees of anatomical abnormalities of the iridocorneal angle were detected. These abnormalities were always bilateral and characterized by excessive mesenchymal tissue in the iridocorneal angle. According to the study of *Ramsey* et al. (1999) cataracts were always present in horses with MCOA, and were characterized by an immature, spherical, nuclear opacity that was most dense at the anterior nuclear-cortical junction of the temporal part of the lens. A posterior ventral subluxation of the temporal part of the lens with concurring iridodonesis and phacodonesis occurring always bilaterally was found in 25% of the horses with megalocornea. Finally an abnormal prominence of the anterior orbital rim with apparent hypertelorism was detected occasionally in horses with multiple ocular abnormalities.

In our study only 2 of 21 horses (5.7%) showed MCOA. Both of them had megalocornea, miosis, circling granula iridica and goniosynechiae. In contrast to the findings of *Ramsey* et al. (1999), only in one horse bilateral anterior cortical cataracts were found. In this particular horse a focal cataract in the posterior cortex was also seen in one eye. In one horse a focal pinpoint capsular opacity was noted, as single ocular finding which, due to its localization, was considered as being related to the MCOA complex. In contrast to the findings of *Ramsey* et al. (1999) in both of the horses with MCOA neither ciliary body cysts nor retinal dysplasia were noted. These lesions might not have been present in the European Mountain horses or they might have been overlooked due to extensive and drug-resistant miosis.

The ophthalmic abnormality of Mountain Horses is termed Anterior Segment Dysgenesis (ASD). This is a congenital and inherited defect in the formation and development of the anterior part of the eye during the early embryonic stages, though some horses may also have abnormal development of the posterior part of the eye. The affected horses are already born with this condition, which is not painful and in the majority of cases bilateral. The functional vision in affected animals is usually not impaired; exceptions are foals with extensive retinal dysplasia that are already blind at birth or horses with lens subluxations and progressive cataract formation (*Ramsey* et al. 1999b).

As already mentioned, two different ocular phenotypes of the disease exist: 1) Large fluid-filled cysts arising from the temporal ciliary body and/or peripheral retina and retinal dysplasia, and 2) MCOA consisting of ciliary and/ or retinal cysts, along with megalocornea, dyscoria, nuclear cataracts, lens subluxation, retinal dysplasia, iris hypoplasia, abnormal drainage angles and macropalpebral fissures.

In Rocky Mountain Horses, a codominant autosomal inheritance model of a single gene with incomplete penetrance in approximately 12 percent of the time has been found by analyzing collected pedigree data (*Ewart* et al. 2000, *Andersson* et al. 2008). Due to the inheritance model CBC are expressed in heterozygous animals and the MCOA in homozygous animals (*Ewart* et al. 2000, *Andersson* et al. 2008).

Animals in small breed registries (as the RMH) generally derive from a small gene pool. Many RMH for example descend from a single foundation sire. Gene pools also can be selectively limited by the intensive breeding of individuals with desired characteristics (e.g. coat, mane and tail color). This selection of phenotypic characteristics especially in the Mountain Horses may have resulted in a concentration of the abnormal gene and subsequently a higher incidence of eye abnormalities.

Several studies concerning mainly RMH have been performed. Though the intraocular pressure was not significantly different between RMH with cornea globosa and RMH with clinically normal eyes, corneal thickness in the central and temporal portions were significantly increased in horses with cornea globosa (*Ramsey* et al. 1999a). Horses with cornea globosa refract near emmetropia due to a reduction of the thickness of the crystalline lens and the depth of the vitreous chamber (*Ramsey* et al. 1999b).

Previous studies have shown that horses with chocolate coat color and white or flaxen mane and tail have the highest disease incidence. This popular color combination among the Mountain Horses is related to the "silver dapple gene". It has been hypothesized that ASD is closely linked to a dominant gene at the "Silver Dapple" locus (*Andersson* et al. 2008).

This gene is a color-diluting gene, which dilutes only the black parts of the animal. It will typically dilute a black mane and tail to flaxen and a black body to a shade of brown or chocolate (*Brunberg* et al. 2006). Genetically, a chocolate colored horse is a black horse with the color-diluting gene at the "Silver Dapple" locus. Red-based horses (chestnut, cremello, palomino) may carry the gene but will not show it. Another characteristic of silver dapple is that as the horses mature and age they commonly get more and more dark hair in their manes and tails.

ASD is not unique to the Mountain Horses and has been documented in other horse breeds (Icelandic horses, Shetland Ponies, Miniatures horses) with chocolate coat color as well (*Ramsey* 2004, *Ekesten* et al. 2009). Having this in mind the

question arose whether ASD might exist in the Black Forest Horse, an ancient German cold-blooded horse breed from the Black Forest region, which is characterized by a mainly dark chestnut (chocolate) color and bright mane and tail (Fig. 5). As part of the present study, authors have examined 50 Black Forest Horses. Neither ciliary body cysts nor multiple ocular anomalies could be detected in any of these horses. DNA testing of these horses was not performed.

Descriptions of ASD as a component of MCOA (microphthalmia, retinal dysplasia, etc.) include inherited conditions in Saint Bernard, Akita and Doberman pinscher dogs (*Martin* and *Leipold* 1974, *Peiffer* and *Fischer* 1983, *Laratta* et al. 1985). A case of isolated keratolenticular dysgenesis has been described in a kitten (*Peiffer* and *Belkin* 1983). While individu-

Fig 5 Typical representative of a Black Forest Horse with chestnut coat color and bright mane and tail, which is the reason why they are called "Schwarzwälder Fuchs" (chestnut) as well.

al horses demonstrated a variety of lesions of both the anterior and posterior ocular segments, all the abnormalities are consistent with dysgenesis of the anterior segment as described in humans, mice and rats (*Cook* 1989). The close resemblance between inheritance and lesions observed in mice and rats, humans with congenital aniridia or anterior segment malformation, and horses with anterior segment dysgenesis syndrome supported the conclusion that ASD syndrome in the horse may be similar to ophthalmic anomalies in other species. Due to the similarities and subtle differences between the horse and human regarding the dysgenesis of the anterior segment the ASD syndrome in the horse is looked at as a potentially rewarding disease model for humans (*Ewart* et al. 2000).

The Rocky Mountain Horse Association has published stringent guidelines for breeding to prevent this disease. If there are intentions to breed a horse the ASD gene status should be known in order to select a mate. Homozygous affected horses should not be bred at all. Horses with cysts should ideally be bred only to clinically normal horses. If they are bred to another horse with cysts a 25% chance exists that the foal will be homozygous and will develop MCOA. To date the classification of the breeding stock is by ophthalmic examination only. Although the gene locus has been identified a DNA-Test is not available (*Andersson* et al. 2008). A DNA-Test for the silver dapple gene is available, however (http://www.horsetesting.com/equine.asp).

In conclusion, RMH and KMSH in Europe have the same eye abnormalities as their relatives in the US. Homozygous animals expressing MCOA are relatively rare in Europe; however, the heterozygous horses expressing ciliary body cysts are almost as common as in the US. Due to the small number of breeding animals, mandatory eye exams should be performed in Europe as well. The strict breeding guidelines issued by the Rocky Mountain Horse Association in North America should also be applied in Europe.

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