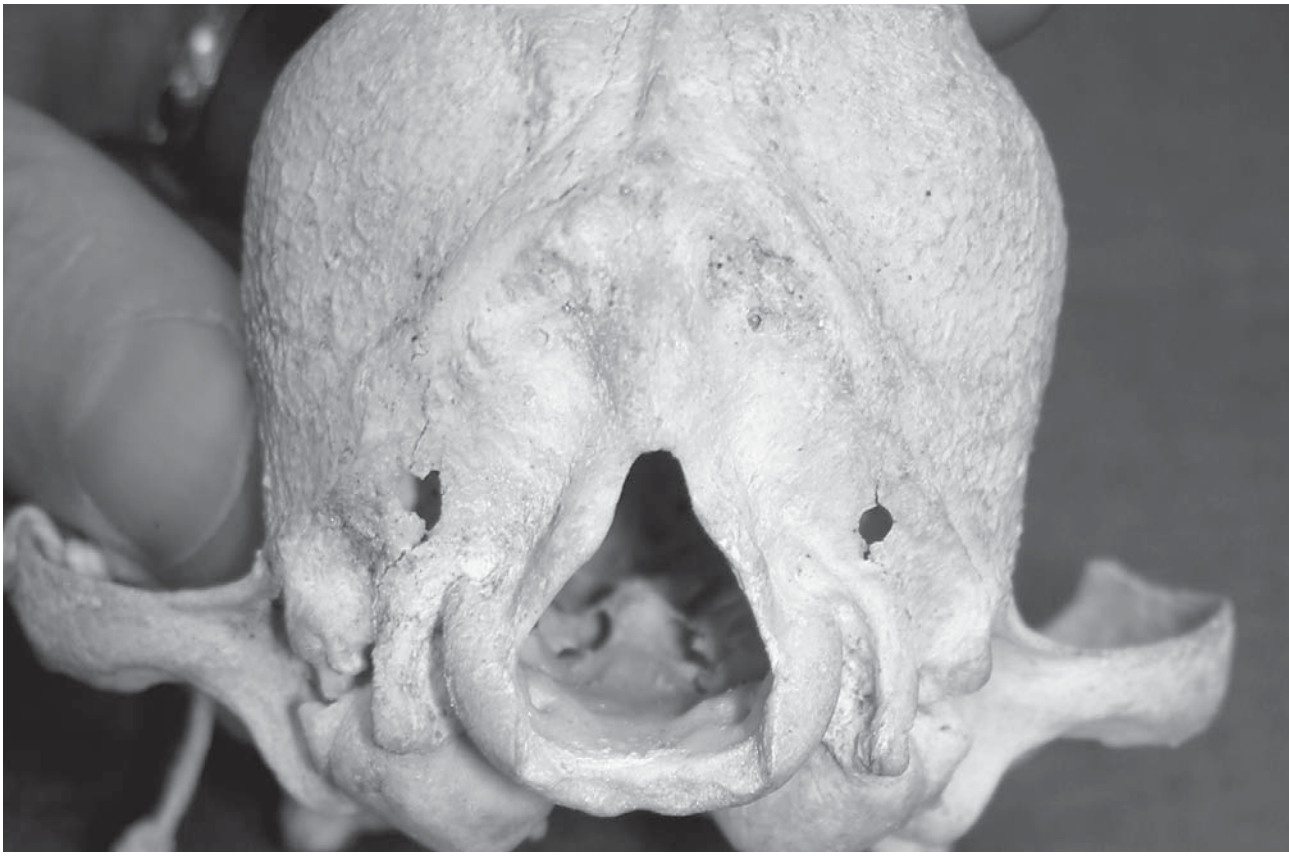

Section 3

Comparison of occipital hypoplasia (Chiari-like malformation) and occipital dysplasia

Chapter 3.1

Co-existence of occipital dysplasia and occipital hypoplasia/syringomyelia in the cavalier King Charles spaniel

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Introduction

The foramen magnum is a ring of bone formed from 4 occipital bone centres; the supraoccipital bone dorsally, the basilar part ventrally and the exoccipitals which bear the occipital condyles laterally (Evans 1993). In addition to providing an exit for the spinal cord, the foramen magnum allows cerebrospinal fluid (CSF) to shunt rostrally and caudally between the head and spine. This rapid efflux and influx compensates for brain expansion and contraction during the cardiac cycle (Oldfield and others 2001). Obstruction to CSF movement can result in development of syringomyelia, a condition whereby fluid containing cavities develop within the spinal cord. The most common cause of syringomyelia in veterinary medicine is occipital bone hypoplasia (Chiari-like malformation) (Rusbridge and others 2000) which is inherited in the cavalier King Charles spaniel (CKCS) and may be seen in other toy breeds (Rusbridge and Knowler 2003, 2004). It is hypothesised that the basi and possibly supraoccipital bone are shortened reducing the volume of the caudal fossa. The cerebellar vermis is often pushed through the foramen magnum and the

medulla is deviated dorsally. Syringomyelia may vary in severity, but for many affected animals is a debilitating neurological disease with clinical signs such as dysesthesia, cervical/occipital pain, paresis, ataxia and scoliosis. One of the most common signs is a tendency to scratch at the shoulder or neck area. The scratching is unlike that seen with ear and skin disease. Generally the dog moves and scratches at the same time; makes minimal or no skin contact; often cries whilst scratches; and initially does it only to one area within the dermatome corresponding to the area of damaged spinal cord.

Other abnormalities of the development of the canine occipital bone are recognised, in particular, occipital dysplasia where there is incomplete ossification of the supraoccipital bone resulting in a widening of the foreman magnum (Parker and Park 1974, Watson and others 1989). This defect varies from a small dorsal notch resulting in a keyhole shaped foramen magnum to a wide midline defect. Watson and others (1989) examined the shape of the foramen magnum in 36 dogs (33 Beagles) and found a considerable variation in the shape of the foramen magnum even within the same breed; the more brachiocephalic the skull the more likely there was occipital dysplasia. In all cases the bony defect was covered by a tough connective tissue membrane which extended as far as the nuchal tubercles so, despite the bony defect, the functional shape of the foramen magnum opening was oval and brain prolapse was prevented. Watson and others (1989) also demonstrated that the presence or absence of the dorsal notch of the foramen magnum is primarily due to variations in the degree of ossification of the ventromedial part of the supraocciptial bone and the authors concluded that it should be regarded as a variation not an anomaly as it did not appear to be associated with any impairment of function. In contrast, Parker and Park (1974) did find neurological deficits in some of the dogs (miniature and toy poodles, Yorkshire terriers, Lhasa apsos, Chihuahuas, Beagle, Pomeranian, Shih Tzu and Maltese) they studied, however it was not established whether these were related to occiptial dysplasia. Some of the dogs had concurrent hydrocephalus. Syringomyelia was not reported as a finding in any of the cases that received a post mortem and in no cases was there any apparent permeant protrusion of the cerebellum into the spinal canal. However not all the dogs had a full post mortem and examination of the spinal cord for syringomyelia. In particular three dogs with a large dorsal notch and a long standing history of ataxia did not have post mortem examination.

In this report two dogs with concurrent occipital dysplasia and hypoplasia with secondary syringomyelia are described. The dogs were related to each other and to other dogs with occipital hypoplasia and secondary syringomyelia but without occipital dysplasia.

Case history

Dog V

A 10 year old male CKCS was presented for examination as the owner was concerned about the possibility of occipital hypoplasia/syringomyelia. This popular stud dog had been identified in a previous study (Rusbridge and Knowler 2003) as being an important ancestor in an extended family of CKCS

with syringomyelia secondary to occipital bone hypoplasia and he was the sire and grandsire of several magnetic resonance imaging (MRI) confirmed cases. The only reported irregularity was he had a mild tendency to scratch at his right mid cervical area. This had been noticed since he was approximately18 months old. There were no other neurological deficits at this time. Ten months after the initial examination the owner reported that he was becoming more sensitive around his right ear and over the following three months he developed tetraparesis more severe on the right with more severe pain. He deteriorated to the stage that the owner elected for him to be euthanatised. Gross post mortem findings are illustrated in Figure 1 a and b.

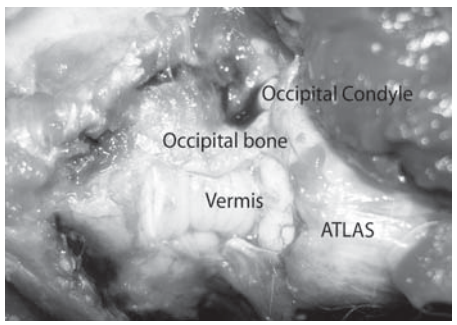

Figure 1a Gross anatomy of the occipital and cranial cervical area in dog V. The neck is flexed. The defect in the occipital bone was originally covered by a tough connective tissue membrane. When the head was in a normal position the cerebellar vermis extended through the foramen magnum.

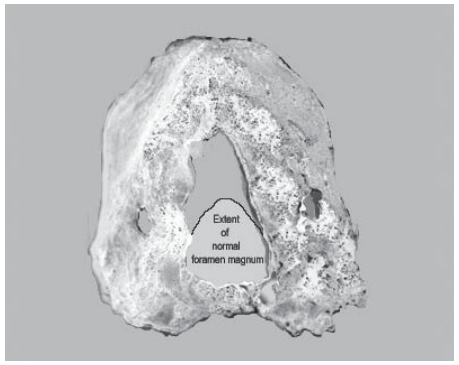

Figure 1b The occipital bone from dog V illustrating the extensive dorsal widening of the foramen magnum

There is a large defect in the supraoccipital bone which was originally covered with a tough membrane that was confluent with the atlantooccipital membrane. In a normal neck position the cerebellum extended into the foramen magnum. There were no other gross lesions that could provide an explanation for the neurological signs and histopathological examination confirmed syringomyelia (Figure 1c).

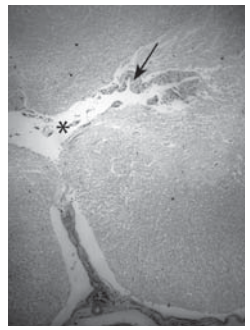

Figure 1c Cross section of the cervical spinal cord from Dog V. There is an irregularly shaped cavitation of the cord (asterixis) centred upon the spinal canal and extending as fissures into adjacent grey and white matter. Focally the cavitation extends to the dorsal and ventral subdural space. The cavities are bordered by frayed parenchyma. There is a little neovascularization, accompanied by a few macrophages, in the wall of the central areas of the cavities (arrow). (Trichome x 40)

A simplified familial relationship between Dog V and other dogs with occipital hypoplasia/syringomyelia is illustrated in Figure 2. For the majority of dogs it is not known whether or not there is concurrent occipital dysplasia as this cannot be readily appreciated on MRI. However there were two descendants where the occipital bone was inspected intraoperatively. Dog T (grandson) and dog O (great grandson) had onset of the signs of syringomyelia at 14 and 28 months respectively. Both had severe signs of pain and neither could be exercised as a consequence. Both were confirmed by MRI and subsequently had a suboccipital craniectomy with atlas cranial laminectomy and durotomy to relieve the obstruction at the foramen magnum. In both dogs there was no dorsal notch to the foramen magnum, i.e. no occiptial dysplasia.

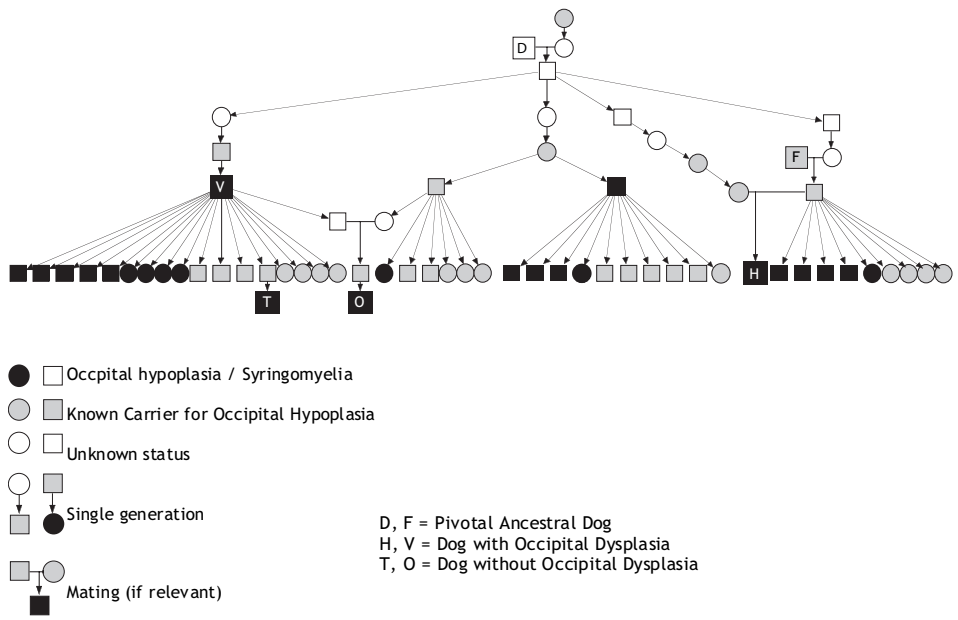

Figure 2 Simplified diagram of one familial relationship between dogs V, H, T and O. The pivotal ancestral dogs had been identified in previous studies (Rusbridge and Knowler 2003, 2004).

Dog H

Dog H was an 8 year old male CKCS with a five year history of brief episodes of unexplained pain. These episodes had become more frequent over the last six months. There was an approximate nine month history of a tendency to scratch at the shoulders (both sides) with a six month history of pelvic limb ataxia and a three week history of thoracic limb weakness. A scoliosis had also been noticed. There had been a partial but not sustained response to 4mg methylprednisolone daily (Medrone; Pfizer) and 200mg gabapentin (Neurontin; Pfizer) twice daily. Neurological examination revealed a bilateral thoracic limb weakness more severe on the right. There was carpal hyperextension of this limb with atrophy of the shoulder muscles and a tendency to stumble. Proprioceptive responses were delayed in the pelvic limbs. A tendency to scratch at the right shoulder was noted. MRI of the brain and vertebral column revealed a small caudal fossa, overcrowding of the foramen magnum and syringomyelia from the level of C1 to L3/L4 (Figure 3).

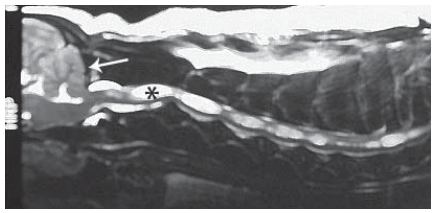

Figure 3 Midsagittal TW2 weighted image of the caudal brain and cervical spinal cord form Dog H. The arrow indicates the occipital bone; the occipital dysplasia cannot be appreciated. The asterixis illustrates the syringomyelia.

The width of the syrinx was variable, and at its maximum at the level of C2, was two thirds of the diameter of the spinal cord. Cervical scoliosis was confirmed. Other than the hypoplasia no other irregularity of the occipital bone was identified. The caudal fossa had been imaged in both sagittal and axial orientations. Due to the rapidly progressive clinical signs and inadequate response to corticosteroids and gabapentin the owner elected for surgical management. A standard approach to the caudal fossa was made. When the supraoccipital bone was exposed it was apparent that it was dysplastic. There was an arch shaped dorsal widening of the foramen magnum. The bony defect was filled by a thick membrane. Above the bony defect there was a band of normal bone (3mm wide) above which was another hole 5mm x 3mm. The bony defect was widened and the thick connective tissue membrane over the defect and the thickened atlantooccipital membrane were removed. The surgery was then continued with a cranial C1 laminectomy which was extended to ~5mm below the tip of the vermis (about 1/3 length of the atlas arch). Finally a durotomy was made from just below the tip of the vermis to the level of the foramen magnum. This allowed further decompression of the cerebellum and allowed the surgeon to ensure that the vermis had been adequately exposed and to facilitate removal of arachnoid adhesions (there were none). The resulting triangular defect was patched with biocompatible collagen matrix (Vet BioSISt ™ ; Cook/ Global Veterinary Products). Closure was routine. Three weeks after surgery the owner reported a marked improvement in demeanour, exercise tolerance, strength and coordination. The scratching behaviour had reduced. The familial relationship of dog H to dog V is illustrated in Figure 2.

Discussion

The occipital bone forms from fusion of the mesenchyme of at least 3 occipital somites (Marin-Padilla 1991). The mesenchyme forms cartilage which in turn undergoes the process of endochondral ossification to form bone. In addition there is membranous tissue caudal to the cartilaginous supraoccipital bone plate which undergoes intramembranous ossification and ultimately fuses to the cartilaginous part. (Matsumura and others 1994). It is proposed that occipital dysplasia occurs when the ventromedial portion of the developing supraoccipital bone fails to ossify (Watson and others 1989). In contrast it is proposed that occipital hypoplasia occurs because of an early paraxial mesodermal insufficiency (Marin-Padilla 1991). Occiptial dysplasia appears not to cause a functional problem because the overall shape and size of the caudal fossa are unchanged. In contrast occiptial hypoplasia results in a reduced volume caudal fossa which in turn can lead to the development of syringomyelia. Occipital dysplasia is common in dogs with a rounded skull shape (Watson and others 1989) and occipital hypoplasia is common in the CKCS therefore it is not surprising that the two conditions should occur in the same dog. What is unusual about dogs V and H is that the progression of the signs of syringomyelia was initially very slow and neither dog displayed severe signs until middle to old age. The majority of dogs with syringomyelia secondary to occipital hypoplasia present with severe compromise before 7 years of age (Rusbridge and other 1997). It is possible that the membrane covering the supraoccipital defect allows for a dynamic expansion and less severe obstruction of CSF movement through the foramen magnum. As a consequence it is possible that syringomyelia could develop more slowly resulting in later onset signs. In other words, dogs with occipital hypoplasia and dysplasia potentially may have a milder phenotype than with occiptial hypoplasia alone.
If this is the case then there are implications for breeding. If the dog is a breeding male, then the mild or subclinical signs may not be recognised by the owner especially when the dog is young. If the dog becomes a popular stud dog then the potential for occipital hypoplasia but not necessary occipital dysplasia may be disseminated widely in the breed. Dog V sired over 50 litters and has hundreds of descendants across the world. Many breeders arrange for their potential breeding stock to have a brain and/or upper cervical MRI with the aim of selecting those without occipital hypoplasia/syringomyelia for at least one half of a mating. If the onset of syringomyelia has been delayed by occipital dysplasia then a dog may be erroneously thought to have a milder phenotype and used for breeding purposes. This is especially important if the screening is done before the age of 18 months. Occipital dysplasia is difficult to identify

on MRI however the foramen magnum can be radiographed in a manner described by Parker and Park (1994) with the dog in dorsal recumbency and with the nose flexed at 25-40º and the x-ray bean centred on the frontal sinus. CT is also likely to be useful. However before recommendations are made for screening it should be established what is acceptable and unacceptable – i.e. what degree of caudal fossa volume reduction leads to syringomyelia.

Conclusion

Occipital dysplasia may be seen in conjunction with occipital hypoplasia possibly resulting in less obstruction of the foramen magnum and later/slower onset of syringomyelia. However, the affected dogs may still pass on a tendency for a more severe phenotype to their descendants. The presence of occipital dysplasia in conjunction with occipital hypoplasia should be taken into account in any future studies on imaging, CSF flow or genotyping and further work is needed to establish whether occipital dysplasia does affect the pathogenesis of syringomyelia.

Acknowledgements

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