



ANIMAL EYE SERVICES

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1st September, 2008

Dear Lynda,

Further to my letter of August 2004 I am pleased to send you an update on our current knowledge of Primary Lens Luxation PLL as it relates to your breed and others.

As I said in 2004 we have been suspicious for some time that PLL is a genetic disease in a number of terrier breeds. We have seen this condition in Jack Russell Terriers, Miniature Fox Terriers, Tenterfield Terriers, Miniature Bull Terriers, Australian Cattle Dogs not necessarily in any perceived order of prevalence. Overseas it has also been reported in many other breeds eg the Tibetan Terrier, Chinese Shar-Pei, Lancashire Heeler, Scottish Terrier, Dandie Dinmont Terrier to name but a few.

Attempts to find the genetic cause of the condition in any breed have been unsuccessful at this point in time despite serious attempts by researchers. Initial attempts involved looking at so called "candidate genes" eg at the University of Missouri in the USA researchers initially started looking for an abnormality (called a "mutation") of a gene called the Fibrillin 1 Gene. Mutations in this gene are responsible for causing a disease in man associated with Lens Luxation called Marfan's Syndrome. More recently researchers at the Animal Health Trust at Newmarket in the UK have been looking for abnormalities in gene called the Tight Junction Protein -1 (TJP-1) gene and while there is still considerable suspicion that they are on the right track nothing is absolutely confirmed at this point in time. The other part of the theory is that whatever the final identity of the gene which causes the abnormality, it seems that it will probably

- turn out to be a common gene across all terrier breeds (ie the gene has a common ancestor because many terrier breeds have common ancestors if one goes back far enough in time) and
- have an autosomal recessive mode of inheritance. This means that we need to be very careful about using potential carriers of the gene in breeding programmes, especially in line breeding situations which will re-unite recessive genes from related dogs to cause the condition again. A recent article in the American Journal of Veterinary Research by a group from the University of California Davis (Inheritance of cataracts and primary lens luxation in Jack Russell Terriers, AM Oberbauer et al AmJVR, February 2008 Vol69 No 2 pp 222-227) has confirmed that cataracts and lens luxation are highly heritable in the Jack Russell Terrier but interestingly that both conditions might not be caused by a single gene.

Unfortunately we do not know the true prevalence (% of total breed numbers affected) of either cataracts or lens luxation in any breed. Any estimates of incidence will always be flawed unless a very large proportion of the total population can be examined for actual physical presence of the condition (we refer to this as the "phenotype" which is different to the "genotype" or actual genetic makeup of the animal at a chromosome level) and this brings me to my second point. Most of you will be aware that there is now an eye scheme in Australia called ACES: the Australian Canine Scheme which offers certification for a range of congenital and inherited eye conditions in dogs. The operation of the scheme is based on similar schemes in Europe, the UK and North America which have been going on for much longer than ACES. It is jointly run by the Australian Veterinary Association, registered specialist veterinary ophthalmologists and the Australian National Kennel Club. The scheme is backed by a quality assurance programme and no individual dog results are made available to anyone apart from the owners of individual dogs. The

results of individual dog examinations are sent to a central database in Canberra where they are “deidentified” and the collective results pooled for statistical purposes. The individual dog information will never be available on a public register unless your breed club decides to do so at a national level (because of natural suspicion between breeders this seems unlikely to happen with any breed club at this point in time!) I would direct you to the Australian Veterinary Association website www.ava.com.au -> Community -> Australian Canine Eye Scheme (ACES) for more information. I would encourage everyone in your breed club to have you dogs examined for evidence of eye disease even if you don't think you have it in your lines: if you never look you'll never know! The signs of PLL can be very subtle at first and can include phenomena such as wobbling of the iris (due to a loose lens behind it) or degenerate vitreous in the anterior chamber of the eye. As an aside I have, in discussion with other ophthalmologists via a private internet bulletin board, recently changed my protocol for examining the eye in PLL susceptible breeds including the JRT. I now have a quick look at the eyes before I dilate the pupils just in case there are already subtle signs of a loose lens. It would be a pity if the lens were already loose and by dilating the pupil we allowed it to move into the front chamber of the eye where it will cause more problems. To put this into perspective however chances of this happening are extremely low. My decision to do this is not based on perceived high incidence of the condition in any breed including yours. My only justification for doing this is that the condition is known to occur at all in any particular breed.

The other reason why I would encourage you to embrace eye certification is that PLL is not the only blinding disease known to occur in your breed. Recently there has been considerable interest overseas in the occurrence of juvenile cataracts in the Jack Russell Terrier. The fact that the disease is perceived to be at nil or at low incidence in your breed in Australia is no reason for complacency. Historically, we have many experiences of eye conditions being observed in Australia for the first time as the result of the importation of UK, European or North American lines where these conditions occur at higher frequency. The only known cause of cataracts in dogs at this point in time is the HSF4 mutation for which there is now a test in the Staffordshire Bull Terrier, the Boston terrier and the French Bulldog. The Davis study suggests the HSF4 mutation is not the cause in the JRT. The absence of a DNA test therefore makes it even more important that you embrace eye certification via the ACES scheme to identify potential eye problems in your breed.

Yours Sincerely



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