

Ectopic ureters in the Entlebucher and the Appenzeller Mountain Dog: Incidence, clinical and genetic characterization

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Key words

Ectopic ureter, Entlebucher mountain dog, Appenzeller mountain dog, phenotype, ultrasonography

Aim of the study

The aim of the study was to evaluate the prevalence of ectopic ureters in the EM and Appenzeller mountain dog (AM) and to improve the characterization and classification of the clinical phenotype.. Furthermore the mode of inheritance is elucidated.

Material and methods

Various European Veterinary Clinics participate in characterisation and classification of the clinical phenotype of Entlebucher Mountain Dogs (EM) and Appenzeller Mountain Dogs (AM). After identification of the dog, a thorough history was taken and the site of ureteral entry is determined by ultrasonographic or CT imaging. To elucidate the mode of inheritance, segregation analysis and variance analysis of collected data were performed.

Results and significance

The clinical phenotype has been classified in a total of 294 EM and of 67 AM by Doppler sonography or B-Flow and computer tomography with contrast medium. 135 EM and 8 AM showed ureteral openings into the bladder neck, while in 52 EM and 3 AM extravesical ureteral openings were diagnosed. 24 EM and 3 AM showed urinary incontinence. In 4 of these dogs, one or both ureters opened into the bladder neck and in 20 dogs into the urethra. While 7 dogs were affected by hydronephrosis, only one of them showed clinical signs of urinary tract disease.

In a pedigree consisting of 346 litters with 170 EM having a clinical diagnosis (uni- or bilateral ectopic ureters), one environmental and four genetic models (general, mixed inheritance, major gene, and polygene) were tested for ectopic ureter by segregation analyses (SAGE program). The analysis failed because in many cases there is only one phenotype known for each litter. We attempt to collect additional data about relatives of affected animals. Molecular genetic analyses are in progress to identify the chromosomal region responsible for this malformation. To identify variants associated with this disease whole-genome association mapping is performed, as this method is suitable for mapping genetic variations across a population and represents a suitable approach to localize loci on dog genome, which predispose diseases with complex background. First results by genotyping 48 affected and 48 unaffected EM using the CanineHD Bead Chip seems promising.

Publications, posters and presentations

Bitterli F.; Schelling C.; Eckrich-Specker C.; Hagen R.; Hungerbühler S.; Dorsch R.; Hittmair K.; Reichler, IM. (2010) Prevalence and inheritance of ectopic ureters in the entlebucher mountain dog. 35th WSAVA Congress Genf 2010 (+ Poster)

Bitterli F.; Eckrich-Specker, C.; Hagen, R.; Hungerbühler, S.; Dorsch, R.; Hittmair, K.; Reichler, IM.; Prevalence of ureteral ectopia in the Entlebucher and Appenzeller Mountain Dogs. 7th EVSSAR Annual Symposium EVSSAR Congress Louvain-La-Neuve (2010) (+Poster)

Reichler, IM. (2010) Ektopische Ureteren - Neue Erkenntnisse. Nephrologie-Kongress Zürich, (19.11.2010)

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