

Ichthyoses in Dogs

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Most diseases that occur in humans are also present in our canine companions. Just as in human patients, there are many different forms of ichthyosis in a variety of dog breeds. All of them have a cornification defect in common that affects the outer layer of the skin. These types of cornification disorders have been observed in a variety of breeds including the Golden Retriever, American bulldog, Norfolk and Jack Russell terriers, Rottweiler, Labrador retriever and Siberian husky, all of which are commonly classified as ichthyosiform dermatoses (Credille et al. 2005; Mauldin et al. 2008; Cadiergues et al. 2008, Guaguere et al. 2009, Mauldin 2013). We have also seen single cases of severe forms in a dachshund, two Dalmatian littermates and an Irish setter. Within each affected breed, the severity, clinical and histological presentation, and molecular characteristics (when known) vary greatly, emphasizing the need for the identification of the causative genetic mutation in affected breeds. To date, the causative mutation has been successfully characterized in a handful of breeds including Golden retrievers (Grall et al. 2012), Jack Russell terriers (Credille et al. 2009), Norfolk terrier (Credille et al. 2005), Cavalier King Charles spaniel (Forman 2012), and American bulldogs (Mauldin, Casal et al.-publication in preparation). In each breed listed above, the clinical presentation resulted from a mutation in a different gene, further demonstrating the diverse nature of the cornification disorders referred to as "ichthyosis" (Mauldin EA 2013).

Currently, the American bulldog is the object of our studies, as we now know the mutation causing this form of ichthyosis, called autosomal recessive congenital ichthyosis. Ongoing detailed studies in these dogs are examining how the barrier functions of the skin are disrupted, which will give us a better understanding of the

disease mechanism. With this new knowledge, novel therapies can be developed for future use in dogs and humans.

References:

- Cadiergues MC, Patel A, Shearer DH, et al. (2008) Cornification defect in the golden retriever: clinical, histopathological, ultrastructural and genetic characterisation. *Vet Dermatol* 19(3):120–9.
- Credille KM, Barnhart KF, Minor J, Dunstan RW (2005) Mild recessive epidermolytic hyperkeratosis associated with a novel keratin 10 donor splice-site mutation in a family of Norfolk terrier dogs. *Br J Dermatol* 153:51-58
- Credille K, Minor J, Barnhart K, et al. (2009) Transglutaminase 1-deficient recessive lamellar ichthyosis associated with a LINE-1 insertion in Jack Russell terrier dogs. *Br J Dermatol* 161:265–72.
- Forman OP, Penderis J, Hartley C, et al (2012) Parallel Mapping and Simultaneous Sequencing Reveals Deletions in BCAN and FAM83H Associated with Discrete Inherited Disorders in a Domestic Dog Breed *PLOS Genetics* 8(1): e1002462
- Grall S, Guague`re E, Planchais S, et al. (2012) PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. *Nat Genet* 44(2):140–7.
- Guaguere E, Bensignor E, Kury S, Degorce-Rubiales F, Muller A, Herbin L, Fontaine J, Andre C (2009) Clinical, histopathological and genetic data of ichthyosis in the golden retriever: a prospective study. *J Small Anim Pract* 50:227-235
- Hargis A, Myers S, Gortel K, Duclos D. (2012) Congenital follicular parakeratosis in the Labrador retriever: Clinical and histopathological characterisation. Submitted for publication.
- Hartley C, Donaldson D, Smith KC, et al. (2012) Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis in 25 Cavalier King Charles spaniel dogs–part I: clinical signs, histopathology, and inheritance. *Vet Ophthalmol* 15:327–32.
- Mauldin EA, Credille KM, Dunstan RW, Casal ML (2008) The clinical and morphologic features of nonepidermolytic ichthyosis in the golden retriever. *Vet Pathol* 45:174-180
- Mauldin EA (2013) Canine Ichthyosis and Related Disorders of Cornification. *Vet Clin North Am Small Anim Pract* 43:89-97