

Questions and Answers

submitted from Dyane Baldwin with response from Dear Dr. Olivry

1. *what are the symptoms/appearance of the PKP1 puppies.*

reply: here is what I wrote for our future article: "In all affected dogs, clinical signs occurred immediately after birth with superficial skin layers sloughing when the neonates were rubbed dry. The skin was described by the breeder to have an abnormal pale and translucent appearance. Three puppies were kept alive for three months; all exhibited waxing and waning superficial skin sloughing with erosions and fissures (crack) at areas of friction (axillae, groin, caudal tarsi, footpads) and mucocutaneous junctions (nasal planum, philtrum, lips, concave ear pinnae, periocular area) (figure 1). Rubbing normal-appearing skin with a pencil eraser led to the formation of erosions (positive direct Nikolskiy sign). Footpads exhibited irregular hyperkeratosis and some claws appeared dystrophic. At three months of age, two of these three dogs were approximately one third of the size and weight of their normal littermates. The hair coat was finer and appeared abnormal compared to that of normal littermates"

2. *Do PKP1 affected puppies die "in utero" and/or be reabsorbed before birth?*

reply: as far as I know from the human disease analog, dogs should not die in utero.

3. *do dogs with PKP1 mutation survive till adulthood?*

reply: they probably could, but they would be severely affected and should probably be euthanized for humane reasons.

4. *if PKP1 live to adulthood, what are their symptoms?*

reply: only 3 dogs were kept till 3 months of age (see #1)

5. *can affected dogs have symptoms that are only part of the disease?*

reply: I am not sure of what you are asking. Can you please clarify?

6. *what expression of symptoms in puppies in a litter are red flags to suggest PKP1 testing of parents?*

reply: pale and translucent skin that sloughs at birth. Tendency to have sores at areas of friction and trauma (see #1)

7. *does PKP1 mutations occur in other breeds?*

reply: at this time, dogs and humans are the only one affected. I am sending a paper summarizing the knowledge of this rare human disease: ectodermal dysplasia - skin fragility syndrome. PKP1 is the name of the gene itself.

I hope this helps. **Thierry Olivry, NC State University**

Dear Dr Olivry Thank-you so much. I will look forward to the paper. I am trying to work with Magi Casals on getting info on the genetic test out to the Chesapeake community.

What I mean by Question 5 is do all signs of the disease always occur. For instance, could a dog only have problems with their footpads ? I ask because I was recently told of a dog who has constant problems with his nails-they get infected; they fall off. Yet he does not have any other symptom as you write or that I saw in the affected pups. So what I wonder is could a dog have a milder form that is misdiagnosed or does it always progress to the severe form I saw in the pups. Looking at human photos-some looked worse than others. What other dog breeds if any have been found with this disease ? I will remember call it by its name and not the gene name. Dyane

Ms Baldwin, There are so few cases diagnosed with this disease that we simply don't know the full spectrum of exhibited signs. As I would expect all affected CBRs to have the same mutation of PKP1, I would expect them to have more or less the same signs.

This disease has only been seen in this breed as far as I know. Dr Olivry

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