Ichthyosis

Affected breeds:
Golden Retriever

Ichthyosis leads to an excessive scaling or dandruff which is particularly noticeable when the dog is brushed. Large flakes of dandruff will be seen. There may also be a dark pigmentation and thickening of the skin – particularly noticeable on the belly – and the skin may be more prone to infection. Ichthyosis is present from birth, but may not be especially noticeable until later in life.

Ichthyosis is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop ichthyosis ("AFFECTED").

Many Golden Retrievers carry the Ichthyosis mutation, and it is important that carriers are not excluded from breeding.

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm Ichthyosis

This test will be reported as:

CLEAR : no evidence of the Ichthyosis mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, and will have Ichthyosis

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

AFFECTED X AFFECTED	= 100% AFFECTED
AFFECTED X CARRIER	= 50% AFFECTED, 50% CARRIER
AFFECTED X CLEAR	= 100% CARRIER
CARRIER X CARRIER	= 25% AFFECTED, 50% CARRIER, 25% CLEAR
CARRIER X CLEAR	= 50% CARRIER, 50% CLEAR
CLEAR X CLEAR	= 100% CLEAR

References

PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. Grall A, Guaguère E, Planchais S, Grond S, Bourrat E, Hausser I, Hitte C, Le Gallo M, Derbois C, Kim GJ, Lagoutte L, Degorce-Rubiales F, Radner FP, Thomas A, Küry S, Bensignor E, Fontaine J, Pin D, Zimmermann R, Zechner R, Lathrop M, Galibert F, André C, Fischer J. Nat Genet. 2012 Jan 15;44(2):140-7. doi: 10.1038/ng.1056.