

# VETERINARY TECHNICAL DATASHEET

Ichthyosis; mutation originally found in Great Dane



Mutation Found In :Great Dane

## Disorder Type

- Skin

## Disease Severity

- Severe

## Background

Ichthyoses are hereditary disorders affecting skin cornification. The genetic background and severity of clinical signs vary between different dog breeds. In Great Danes, the disease leads to severe, generalized hyperkeratosis of the skin promoting secondary infections.

## Key Signs

- Wrinkled and thickened skin
- Scaling skin
- Secondary skin infections

## Clinical Description

Affected puppies present with strongly wrinkled, thickened, dry, inelastic, scaling skin. The region around the eyes and nose are more strongly affected. In some cases, swelling and wrinkling of the skin around the eyes may impede the puppy from opening its eyes. Wrinkles in the exudative part of the skin promote severe secondary skin infections. Affected puppies may also have a displaced and wrinkled auditory canal.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- SLC27A4

## Next Steps

There is no cure for this condition and affected dogs are treated symptomatically. Affected dogs are often euthanized on welfare ground in the early months of life if clinical signs are severe and quality of life impacted.

## References

Metzger J, Wöhlke A, Mischke R, Hoffmann A, Hewicker-Trautwein M, Küch E, Naim H, Distl O. A Novel SLC27A4 Splice Acceptor Site Mutation in Great Danes with Ichthyosis. PLoS ONE 10(10): e0141514, 2015.