

## Dermatosparaxis in goats – a fragile skin disorder

Dear Breeders,

We have had reports of “Australian Miniature Goats” affected by the fragile skin disorder known as dermatosparaxis. We seek your assistance to pursue a DNA investigation to find the defective gene. We require additional samples from affected animals and their parents to progress this study.

**Dermatosparaxis** is an inherited connective tissue disorder characterised by extreme skin fragility caused by abnormal collagen (gives strength) in the skin. The condition has been found in multiple sheep breeds (including Australian Merinos and South African White Dorpers) and identical conditions in humans (Ehlers-Danlos Syndrome Type VIIC) and cattle. The extreme skin fragility or tearing means that this condition is ultimately fatal impacting animal welfare and causing economic loss.

The kids reported with this condition so far have either been still born or died shortly after birth. The affected kids have been larger than expected and have had split skin on their abdomen. Typically the skin has been split between the anus and the umbilical cord. The skin on the kids is fragile and easily torn.

Dermatosparaxis is an inherited condition passed from parents to offspring through the DNA. We predict this condition is recessive in goats, similar to other species, which means that affected animals receive two defective gene copies: one from the buck and one from the doe. Both the buck and the doe are carriers of the defective gene (1 copy) but do not show symptoms.

The defective gene in goats has not been reported previously, hence our investigation. We aim to collect sufficient samples and data to screen the DNA of affected goats to identify the mutation responsible, which will provide an effective management solution for impacted breeders.

Once the gene is identified, a test will be developed. Breeders can then use the test to identify which of their animals are carriers of the gene. This will enable the condition to be managed in the miniature goat population.



Yours sincerely



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