

Gaucher disease

Gene Test



Background

Lincoln University has provided gene tests for the sheep industry for over 17 years. Along with Robert Suter, Senior Veterinary Officer, Agriculture Services and Biosecurity Operations, Department of Economic Development, Jobs, Transport and Resources, Victoria, Australia; we have developed a gene test for Gaucher disease.

Gaucher disease (pronounced Gow-sher) has been described in Australian

Southdown flocks. It has been previously reported in mice and a dog, and is well known in humans.

To date, Gaucher disease has only been identified in purebred Southdown flocks in Australia. The symptoms can include:

- Affected lambs are unable to stand from birth, and die from a combination of hypothermia and hypoglycaemia.

- Lambs exhibit rigidity of the major extensor muscle groups, so are unable to rise, or to hold their heads up to suckle.
- Lambs may have leathery skin, especially in the neck region.
- Farmers also report that some affected lambs will bleat incessantly.

Those lambs that exhibit the disease normally die within one week of birth.



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Sheep that only have one copy of the mutated gene, show no outward sign of the disease, but are termed carrier animals. These sheep when bred with another carrier animal will have an approximately 25% chance of bearing a Gaucher-infected lamb, a 50% chance of bearing a carrier lamb, and a 25% chance of bearing a lamb carrying no mutation.

Presently the only sure way to eliminate this disease is to gene test through Lincoln University, and to remove all carrier animals from your breeding program.

The test

Blood samples are collected by the farmer on our test cards.

In our typing system we will report the results as:

- N,N - non-carrier which is a sheep clear of the disease
- C,N - carrier sheep
- C,C - sheep with Gaucher disease

All sheep have paired alleles, reflecting that they have paired chromosomes and inherit one allele from each parent.

They therefore pass each allele on to their offspring in approximately a 50:50 ratio. This allows farmers to follow pedigrees and identify which families or sheep are passing on the mutation. While not fool-proof, as miss-mothering and recording errors do occur, it does allow for farmers to identify sheep that are the likely source of the mutation.

Disclaimer

Lincoln University and the Lincoln University Gene-Marker Laboratory cannot be held responsible for the outcome of any decisions made by breeders in the breeding of sheep using this DNA-typing technology. The genetic information supplied to breeders may only be used by them on the assumption that they assume responsibility for any loss, damage or consequence resulting directly or indirectly from the use of that information. The liability of Lincoln University and the Lincoln University Gene-Marker Laboratory is limited to re-testing individual sheep where an error has been made at some stage of the DNA testing process.

Breeding for gaucher free flocks

Like other genetic diseases, the sheep industry has a responsibility to eliminate where possible, those genetic diseases that are terminal or produce significant animal distress or production loss.

The simplest and most effective method is to test all animals once hogget selection has been done post-weaning, and after cull ewes have been removed prior to mating. The testing of sale rams and stud sires should also be done at this time. This is generally the most cost effective time to test. Some farmers, because of the high one-off cost, may test ewes in age groups over several years. Stud sires that have been brought or newly bred, must be tested. The key always is to make sure the ram breeder only sells non-carrier rams to their clients. Even if there are Gaucher carrier ewes in the maternal commercial flocks, no Gaucher infected lambs will be born if all sires are tested and are not carriers. Carrier lambs may still be produced.

Getting your sheep tested

Contact the Lincoln University Gene-Marker Laboratory at the numbers listed. We will send out special cards for collecting small blood samples, along with instructions on how to safely and easily collect blood from sheep. Only when your samples are received by the Laboratory can typing be undertaken. If you are outside of New Zealand, an import certificate will be supplied. Multiple tests can be done from the same blood sample, so please have sufficient blood on the card.

Lincoln University Gene-Marker Laboratory Tests

A1/A2 Cattle Genetics	GDF9 Fertility
Booroola	Inverdale
Cold Tolerance	Microphthalmia
Dermatoparaxis	Scrapie
Footrot	T+ Muscling
Gaucher Disease	

Testing costs

A schedule of prices is available on request. Discounts are available for large volumes or multiple tests. For overseas clients we price test and invoice in your currency.



Find out more at
research.lincoln.ac.nz/testing-analytical-services/gene-marker-lab

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