## Testing cost

A separate schedule of prices is available on request. Discounts apply for multiple tests carried out in a calendar year (1 January – 31 December) and for testing done together with other gene tests provided by the Lincoln University Gene-Marker Laboratory.


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## Disclaimer



# Microphthalmia
## Gene Test

Want to find out more?

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## Microphthalmia gene test

Microphthalmia in sheep is found primarily within the Texel breed and crosses made out of that breed. It is characterised by lambs having extremely small or absent eyes and, accordingly, the affected lambs are blind.

In Texel sheep, microphthalmia behaves as a monogenic autosomal recessive trait. This means it is primarily controlled by a single gene (called *PITX*3) that is found on one of the non-sex chromosomes (ovine chromosome 22), and that two faulty or mutant copies of the gene are required for lambs to be affected.

The mutant or abnormal form of the PITX3 gene has a small change in its DNA (described scientifically as c.338G>C, p.R113P). This mutation is in a key part of the gene and can be detected using DNA sequence analysis or gene-typing technologies. Further background on this test is available in the original scientific work, published in a paper at www.plosone.org/article/info:doi%2F10.1371%2Fjournal.pone.0008689.

Selection against this causative mutation could be used to eliminate microphthalmia from Texel sheep and Texel-crosses in sheep production systems and thereby both reduce the current cost of this disease and stop its wider spread into other breeds and national flocks.

The Lincoln University Gene-Marker Laboratory has developed a gene-typing system to detect the mutation in *PITX*3 and is therefore offering a DNA typing service to sheep breeders using Texel or Texel-cross genetics.

## The test

Blood samples collected from sheep can be 'typed' to reveal whether they carry one or two copies, or mutant alleles (C), of the PITX3 gene.

In our typing system we report the results simply as non-carrier, carrier, or affected and blind as follows:

| | | |
|---|---|---|
| **(N, N)** | **–** | **non-carrier** |
| **(C, N)** | **–** | **carrier** |
| **(C, C)** | **–** | **affected and blind** |

Sheep have two alleles, reflecting that they have paired chromosomes and inherit one allele from each parent. They can therefore pass each allele on to their progeny in approximately a 50:50 ratio. The 'flow' of alleles can thus be followed through extended pedigrees of sheep.

The test allows you to identify sheep that carry mutant alleles.

## Breeding with sheep that may be carriers of mutant alleles

This gene test allows breeders and ram-buyers to monitor whether their sheep are carriers of the mutant allele and therefore whether they might produce affected or blind lambs when mated to another carrier.

It is effectively a management tool, as carrier sheep do not necessarily need to be culled, but instead their genetics can be managed to insure that progeny do not inherit two mutant forms of the PITX3 gene and, therefore, be blind.

Whilst eradicating the mutant form of *PITX*3 would be the most desirable outcome across all the world's sheep, that will require time and concerted effort by sheep breeders.

We recommend the following approach when using this technology:

- That breeders place their main emphasis on testing breeding stock, especially rams, as they typically have the most impact on a flock genetically
- That sheep breeders keep precise pedigree records of all genetic testing to be able to follow the mutant form of the gene in their flock
- That the test is used conservatively in the context of the 'golden rule' of genetics, whereby selection for multiple traits limits genetic gain for any given trait. In this context care needs to be taken in culling carrier sheep as they may have genetic merit for other traits. Breeders should always be cautious about overly narrowing their genetic 'base', as this will reduce diversity and may reduce their ability to make genetic gain in other traits
- That carrier sheep should be removed from breeding programmes at the first practical opportunity. For example, ewes that are known to be carriers could be put to terminal sires.

## Getting your sheep tested

Testing will be performed by the Lincoln University Gene-Marker Laboratory. If breeders and farmers contact the testing laboratory at the numbers listed then we will send out special cards for collecting small blood samples, along with instructions on how to easily and safely collect blood from sheep. Only when these cards are returned to us will typing be undertaken.