

Adopting genomic technologies to provide insights on Microphthalmia in Texel sheep

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Application

During 2023, a small number of cases of Microphthalmia were reported anecdotally by pedigree Texel sheep breeders in the UK. This disorder is a welfare issue because it can cause blindness in lambs who therefore do not survive to adulthood. Using an existing database of genomic data, animals that could produce affected lambs were identified. This information was then provided to pedigree breeders along with advice on breeding strategies to enable them to make more effective breeding decisions and reduce the chances of producing affected lambs.

Introduction

Microphthalmia in sheep is an autosomal recessive inherited condition within the Texel breed. It is characterised in affected animals by extremely small or no eyes and accordingly the animals are blind and do not survive to adulthood.

The condition has been reported in Texel populations globally and is associated with a missense mutation (c.338G>C, p.R113P) in the *PITX3* gene that is involved in vertebrate lens formation (Becker et al. 2010; Goncalves et al. 2018).

This single nucleotide polymorphism (SNP) is included in commercially available genotyping SNP arrays and can be reported on simply. Existing data can also be retrospectively analysed to provide information on previously genotyped animals.

The aim of this work was to determine the level of carriage of the affected allele within the registered pedigree male population of British Texels (as only registered animals can be used for pedigree production), develop an effective mechanism of providing this information to pedigree breeders, and support the monitoring and removal of the carrier gene within the realms of Zootechnological legislation.

Materials and Methods

Low and medium density (15-50K) SNP genotypes (Ovine_15K, GGP_Ovine_15Kv2, GGP_Ovine_50K and GGP_Ovine_50Kv2) are stored commercially by Neogen Europe Ltd for all pedigree registered male Texel animals as part of Breed Society policy. The database includes 20,778 low and medium density individual animal genotypes and was retrospectively analysed to provide genotype information on the associated SNP in the *PITX3* gene in 2,746 animals. Data from animals born prior to 2018 were not included in subsequent analyses because information on fewer than 200 animals per year were available.

These were predominantly male animals registered in the preceding three years and represented much of the *in-situ* pedigree male population.

Carrier animals that could pass the condition on to their progeny were identified as heterozygous (C/G); whereas homozygous animals (G/G) were considered resistant. Homozygous (C/C) animals are clinically affected and do not survive.

Additional genotypes were obtained for specific individuals that had been excluded from the initial analysis, to supplement the data. The change in breed registry policy to routine reporting of single-marker information from all commercially obtained genotypes, has provided further information on the prevalence of resistant and carrier animals in the pedigree population.

Results

To date 7,908 animals have a known *PITX3* genotype, including 6,914 males (5,795 registered and 1,119 unregistered) and 994 females (870 registered and 124 unregistered). In the registered pedigree male population, the overall proportion of carrier animals is 2.2%, however when split by year of birth, there is a noticeable increase in the rate of carrier animals born between 2020/2021 (Figure 1).

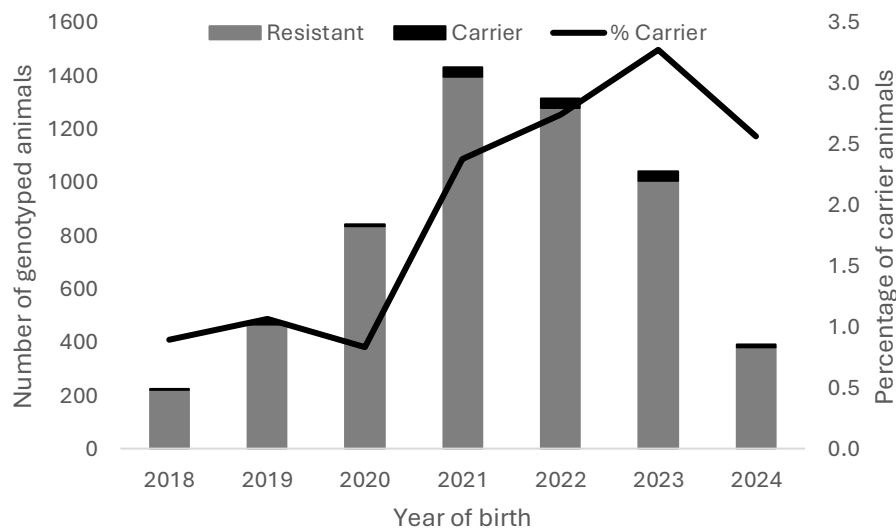


Figure1. Numbers of pedigree registered male Texel animals genotyped and proportion of registered males that are Microphthalmia carriers, by year of birth.

Conclusions

The Breed Society has identified an increase in the proportion of Microphthalmia carrier animals in the pedigree registered male Texel population. This was precipitated by the widespread use of a small number of influential sires that were carriers.

Routine genotyping of all registered sires (approx. 1400-1500 per year) has allowed the Breed Society to rapidly identify this trend. This has also led to the development of reporting systems that inform pedigree breeders of the status of their animals within the confines of Zootechnological legislation. The breed's online public database has been adapted to publish all genotype information available. This enables pedigree breeders and commercial producers to make informed breeding decisions on the use of registered and genotyped pedigree animals.

Breeders are now more aware of the condition than previously and are more informed about their animals and those they may consider purchasing. This also supports a change of

behaviour by breeders influencing the reduction of carrier animals in the male, and potentially the female, population.

The identification of carrier animals for this deleterious gene and the facilitation of improved knowledge transfer by the Breed Society supports a responsible approach, increasing health and welfare in the pedigree population. Improving animal health is a prerequisite to sound animal husbandry, with positive knock-on effects on productivity and efficiency. It is now more likely that fewer lambs will be born blind than would have occurred without this intervention, resulting in lower levels of avoidable lamb losses.

References

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