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## Inheritance of chondrodysplasia in Texel sheep

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### ABSTRACT

A skeletal chondrodysplasia (noncancerous abnormal cell growth affecting cartilaginous tissues) characterized by dwarfism and angular deformity of the forelimbs has been recognized in Texel and Texel cross lambs. Some affected lambs have normal co-twins indicating the disease is likely to be of genetic origin. This study outlines the mode of inheritance of the disorder. Three affected rams were mated to 221 unaffected ewes to generate 123 daughters, which were backcrossed by multiple sire mating with their sires the subsequent season to produce 83 lambs for evaluation. The time course of the morphological progression of the disease was recorded. Of the 46 lambs that were successfully diagnosed, 22 were affected with the condition and 24 were unaffected. This was consistent with a 50:50 ratio expected for recessive inheritance in a backcross between affected and carrier animals. Within sexes, 11 males (24%) and 11 females (24%) were affected with 10 males (22%) and 14 females (30%) were unaffected. This was not different ( $P < 0.05$ ) from a 0.25 proportion for autosomal recessive inheritance. Variation in the age of onset (16 days  $\pm$  7 days) and severity of clinical expression suggests variable penetrance, with semi-lethality.

**Key words:** Texel sheep; chondrodysplasia; inheritance.

### INTRODUCTION

Chondrodysplastic (noncancerous abnormal cell growth affecting cartilaginous tissues) sheep were first recognized in a commercial sheep breeding and lamb finishing property running approximately 1,100 mixed-breed ewes, including Texel, Perendale, and White-faced Marsh breeds, in Southland. Terminal sire rams used included those with Perendale, Poll Dorset, Texel x Romney, Texel x Suffolk, and Texel x Perendale breeding (Thompson *et al.*, 2005). The affected animals were of Texel or part Texel origin.

The affected progeny appeared to be normal at birth, but as early as seven days of age showed evidence of reduced growth rate, shortened legs and neck, angular forelimb deformities, and progressive exercise intolerance. Affected animals usually had a wide-based stance. A further characteristic seen in some affected lambs was a tendency to walk backwards.

Phenotypically, the chondrodysplasia has a spectrum of severity, both morphologically and histologically. Some animals with the condition die as young as two weeks of age while other, less severely affected animals live to breeding age. Thus, there are varying degrees of disproportionate dwarfism. Morphologically, postmortem analysis of the joints of a number of severely affected individuals revealed widespread loss of articular cartilage and exposure of subchondral bone on the weight-bearing surfaces of the leg bones. This indicates a breakdown in the cartilage structure and function.

The aim of this study was to determine the mode of inheritance of the chondrodysplasia.

### MATERIALS AND METHODS

#### Flock background

The dams and sire producing the affected progeny on the Southland property were well-grown and appeared clinically normal, with the rams having been purchased from the same breeder for several years (Thompson *et al.*, 2005). While some of the affected lambs born during the 2002 season had pigmented faces, they were most likely sired by a Texel x Suffolk cross ram that was no longer on the property (Thompson *et al.*, 2005). Parentage was confirmed for all dwarf lambs available for testing on the Southland property in the 2002 season using DNA fingerprinting; a service provided by Signagen®, formally a trading subsidiary of Forest Research Institute, Rotorua, based on microsatellite markers. All affected lambs born had been sired by the same ram (Thompson *et al.*, 2005).

#### Animals

A number of animals from the affected farm were transported to Massey University for this study. These included six affected rams, five affected ewes, 17 putative carrier ewes and a number of younger animals including eight rising two-tooth ewes, and 11 lambs. Several of the younger animals had to be euthanased prior to the breeding season in the first year of the trial due to progression of the chondrodysplasia condition. Thus not all of the animals mentioned above were included in the first mating season. Euthanased animals provided a source for sample collection. Animals were grazed on pasture and offered water and feed *ad libitum*, as the seasonal conditions permitted.

### **Pilot trial**

A small flock of suspected carrier ewes, defined as appearing clinically normal and having produced affected lambs, and affected mixed age ewes were multiple sire mated to affected rams in 2003, to produce lambs to monitor the growth characteristics of the diseased animals.

Five mildly affected two-tooth ewes and 20 mixed age putative carrier ewes were mated with an affected ram over a five week period.

### **Outcross and backcross trial**

In 2003 an outcross trial was also undertaken with 221 unrelated ewes that had never been recorded as producing affected lambs. The ewes were multiple sire mated to a further three affected rams, with the aim of producing daughters for backcrossing to their sires. One hundred and twenty five daughters were then multiple sire mated to their sires in a backcross design the following year in 2004.

### **Data recording**

Newly born lambs were caught in the paddock, tagged and identified to their dam by either assisted delivery or the lamb suckling from the mother. Lambs were sprayed with the corresponding tag number on their sides, to allow identification and monitoring from a distance. The birth date was recorded and used along with the date of first visual detection of disease symptoms to provide a time course of the morphological progression of the disease. Data collected for new born lambs included weight, sex, birth rank, dam number and morphological measurements of crown-rump length, front leg length, back leg length, and girth. Photographs were taken of lambs as they aged, at two day intervals. Affected status was assigned based on development of dwarfism, wide-based stance, exercise intolerance and skeletal measurement analysis (S. Piripi, Unpublished data). Weight measurements at time of onset were not suitable criteria for distinguishing the phenotype.

### **Sample collection**

Offspring from the affected ewes were of primary interest for tissue sample collection from birth. Samples from deceased and euthanased animals included articular, physeal, and tracheal cartilage samples. Some of which were frozen at  $-18^{\circ}\text{C}$  for biochemical analysis, and some were stored in 10% buffered formalin for histological analysis. These samples, combined with visual assessment of the animals when alive, assisted in assigning affected status. Tissue samples including muscle, spleen and blood samples were collected to provide DNA for genetic analysis. Normal tissue for use as a control was collected from an external source in the earlier stages of lambing because affected and unaffected lambs could not be distinguished

reliably during the first seven days of life. After identification of affected animals, control samples were taken from within the existing flock from carrier animals that were not affected by the disease.

### **Statistical analysis**

Chi square analysis was used to test the "goodness of fit" of the recorded data to the expected distribution for autosomal recessive whereby an affected (*cc*) ram mated with carrier ewes (*c+*) should give 50:50 *cc:c+* genotypes in the offspring, with no difference in segregation expected between male and female offspring.

## **RESULTS**

### **Pilot trial (2003)**

The 25 ewes mated with an affected ram produced 38 (19 male, 19 female) progeny. All lambs born to a cross between an affected ram and an affected ewe were dwarves.

### **Outcross trial (2003)**

The 221 mixed age ewes were pregnancy scanned 46 days after the last day of mating.

The ewes scanned at a rate of 140 lambs per 100 ewes (310/221). These ewes later produced 95 male and 119 female lambs, born over a six week period beginning on 2 September. Thus, a total of 125 females (119 from the outcross trial and 6 from the pilot trial) were potentially available for backcrossing the following year.

### **Backcross (2004)**

One hundred and twenty three of the 125 available female hoggets survived to mating. Pregnancy scanning was carried out 46 days after the end of mating. Of the 78 hoggets which were deemed to be pregnant, 68 were scanned as carrying one foetus and 10 as carrying two foetuses.

A total of 83 lambs were born over a four week period beginning on 26 September. Of these lambs, 23 died at birth and 14 died within the first 14 days of life. The disease status of these animals was thus not able to be recorded by clinical phenotype alone with these 37 animals excluded from the chi square analysis. There may be the opportunity to categorize these animals as affected or not affected by clinical phenotype (S. Piripi, Unpublished data).

The observed disease status and sex results for the 46 live offspring that survived more than 14 days, produced by the carrier hogget ewes are presented in Table 1.

The observed proportion of affected lambs was 0.478 (22/46), while the expected proportion was 0.5. The observed proportion of male lambs was 0.456 (21/46), while the expected proportion was 0.5. The chi square of 0.376 ( $P < 0.05$ ) was not significantly different from the expected 50%. This outcome provides support that the disease is

inherited in an autosomal recessive manner. However, caution should be exercised in interpreting results of small samples (Sokal and Rohlf, 1969).

**TABLE 1:** Number of male and female progeny classified as being either affected or not unaffected with the Chondrodysplasia condition following crossing an affected ram with carrier ewes.

Sex of progeny	Chondrodysplasia condition in progeny		
	Affected	Carrier	Total
Male	11	10	21
Female	11	14	25
Total	22	24	46

## DISCUSSION

The animals born over the two year period were assessed for development of the condition and monitored as they grew. The pilot trial provided an opportunity to assess the technical aspects of lamb monitoring. The outcome of the backcross trial indicated an autosomal recessive disease.

The results suggest that the condition under study is a recessively inherited chondrodysplasia of the Texel breed. In all affected animals there were Texel genes on both the sire and dam sides of the pedigree. Some affected individuals had normal cotwins (T. Byrne, Unpublished data). Parentage testing results identified one ram as the sire of all affected lambs from the 2002 season, providing further evidence for a genetic aetiology (Thompson *et al.*, 2005).

Given that the Texel breed in New Zealand is based on a relatively small number of founder animals (Tervit *et al.*, 1986), it is not surprising that with a condition due to simple inheritance being present in the breed stock a number of clinical cases have been reported. With the continued use of a carrier ram, carrier ewes eventually became part of a flock and are joined with carrier rams to produce affected progeny (Thompson *et al.*, 2005). Key observations that support an autosomal recessive

mode of inheritance include the following: no normal lambs were born to a cross between affected parents; affected lambs have been produced from a cross between phenotypically normal parents; a cross between a phenotypically normal parent and an affected parent produces both normal and affected progeny; and no third phenotype has been identified at this stage.

The proportions of normal and affected lambs, from the putative carrier ewes crossed with an affected ram in the pilot trial provide support for the hypothesis that the chondrodysplasia was indeed caused by a single recessive gene. In addition to this, proportions of normal and affected lambs in the backcross trial in year two, with a larger number of animals, were consistent with the expected proportions assuming recessive inheritance. The equal number of male and female lambs born that were affected with chondrodysplasia in the backcross trial indicated that the inheritance was not sex-linked.

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