

Incomplete reduction of branchial clefts in Mutton Merino lambs

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Dates:

Received: 26 Nov. 2012

Accepted: 20 Dec. 2013

Published: 19 May 2014

How to cite this article:

Leask, R., Pettey, K.P. & Bath, G.F., 2014, 'Incomplete reduction of branchial clefts in Mutton Merino lambs', *Journal of the South African Veterinary Association* 85(1), Art. #964, 3 pages. <http://dx.doi.org/10.4102/jsava.v85i1.964>

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Congenital malformations of the branchial arches, clefts and grooves have not been previously reported in sheep. These defects may be due to infectious agents (especially viruses), toxins or genetic abnormalities. Defects were reported in two of a set of quadruplet lambs born prematurely to an eight-tooth Mutton Merino ewe. The lambs weighed between 2.0 kg and 2.5 kg; this is below the normal expected birth weight of 3.5 kg for quadruplet lambs, below which viability is compromised. The firstborn lamb was severely affected by bilateral orauricular fistulae. The second lamb was unilaterally affected on the right, less severely than the first. The third lamb was normal and the fourth was mummified. The occurrence of another case in this small flock almost a decade earlier indicates that there could be genetic involvement.

Introduction

Congenital malformations of the branchial arches, clefts and grooves have not been previously reported in sheep (Aitkin 2007; Bath & de Wet 2001; Brightling 1988; Fraser & Stamp 1987; Hindson & Winter 1990; Kimberling 1988; Plant 1992; Radostits *et al.* 2000). Venzke and Grossman (1951) reported a case of patency of the first pair of pharyngeal pouches in a calf. They recorded bilateral patency of the external auditory meatus and the absence of buccal tissue. This differs from incomplete closure, where only a small portion of the pouch remains patent, resulting in either a sinus or fistula (Nazario & Vermeulen 2010; Parke & Settles 1991; Tham & Low 2005).

In humans, branchial arches, clefts or grooves and pouches are formed during the fourth week of gestation (Acierno & Waldhausen 2007; Arey 1965), as can be seen in Figure 1 and Figure 2. The first arch forms mandibular and maxillary processes, parts of the sides and floor of the mouth (Arey 1965), as well as parts of the inner ear (Acierno & Waldhausen 2007). The first cleft or groove forms the external auditory meatus (Arey 1965), while the first pouch forms the auditory or Eustachian tube, mastoid cells and the middle ear cavity (Acierno & Waldhausen 2007; Arey 1965). The second arch grows over and obscures the other arches and is also responsible for the formation of the proximal third of the tongue (Arey 1965; Gerneke 1985). Branchial defects can present as one of three conditions: cysts, sinuses and fistulae (Acierno & Waldhausen 2007). Branchial defects have been previously reported in a calf (Venzke & Grossman 1951), horses (Lane 1993) and humans. In a retrospective study on humans, Rattan *et al.* (2006) reported that second branchial cleft defects were three times as common in males and were predominantly unilateral in occurrence.

The chief aetiological categories of congenital defects are infectious agents (especially viruses) (Coetzer, Thomson & Tustin 1994), toxins (Kellerman *et al.* 2007) and chromosomal abnormalities (Arey 1965; Gerneke 1985). The last group of causes is of particular concern if the defect is linked to a recessive gene or one with incomplete expression. Venzke and Grossman (1951) recorded that there was no evidence that the problem they reported had occurred previously in the herd. This differs from this case where there was recollection that an adult ewe had presented with a similar condition some years previously.

Case history

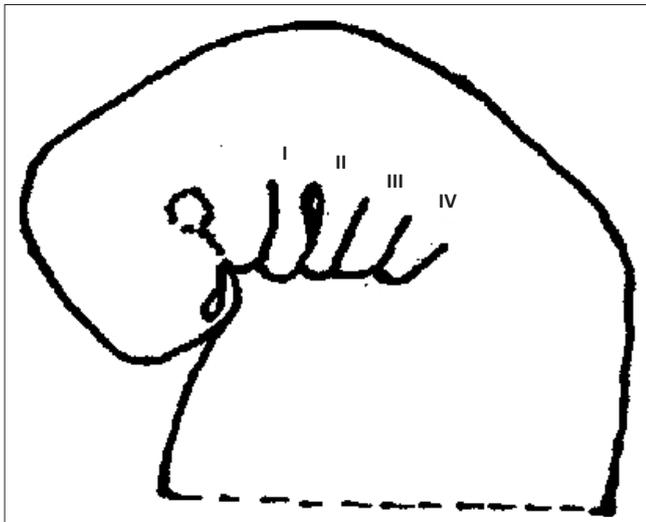
An eight-tooth Mutton Merino ewe gave birth to quadruplets. Two of the lambs (both males) were affected by incomplete closure of the branchial arches, one lamb was normal and one lamb was mummified. The lambs were born prematurely and weighed between 2.0 kg and 2.5 kg. On examination of the oral cavity the teeth had not yet protruded through the gums. This can be expected with multiples, as the lambs are often smaller and born earlier than the expected date of partus due to limited available space *in utero*. However, normal birth weights of quadruplets would be approximately 3.5 kg; viability is compromised in those weighing less than 3.5 kg at birth. This was the only ewe within the flock to give birth to lambs with congenital defects at the time. The ewes were mated over a period of six weeks and all procedures such as vaccinations,

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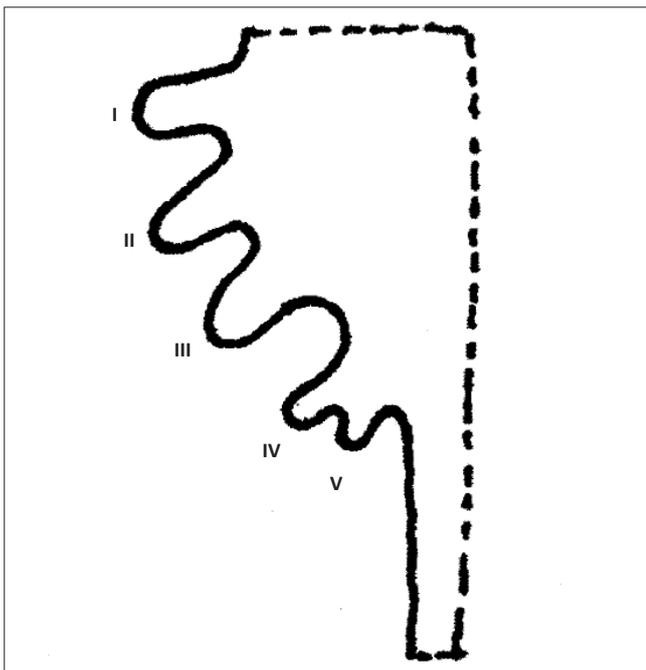
dipping and drenching were performed simultaneously on the flock. The flock grazed irrigated kikuyu pastures and was always kept together as a group in the same camp. Toxic plants have not been identified in any of these camps.

The first-born lamb (Lamb 1) weighed 2.22 kg and was severely affected by bilateral oro-auricular fistulae (Figures 3 and Figure 4). The second-born lamb (Lamb 2) weighed 2.46 kg and was less severely affected than the first (Figure 5) with what appeared to be a unilateral incomplete fistula on the right. All three of these fistulae opened just ventral to the external ear and the tissue that made up the duct was macroscopically consistent with oral mucosa. The third lamb (Lamb 3) was born later that evening and was unaffected.



Source: Arey (1965).

FIGURE 1: External view of branchial arches I, II, III and IV separated by branchial grooves.



Source: Arey (1965).

Note: Each pouch eventually comes into contact with the branchial grooves.

FIGURE 2: Internal view of pharynx indicating pharyngeal pouches I, II, III, IV and V.

The last-born lamb (Lamb 4) was mummified and was only discovered in the straw the following day. Lamb 1 showed left unilateral involvement of the first branchial groove or pouch and the second branchial arch (Figures 3 and 4),

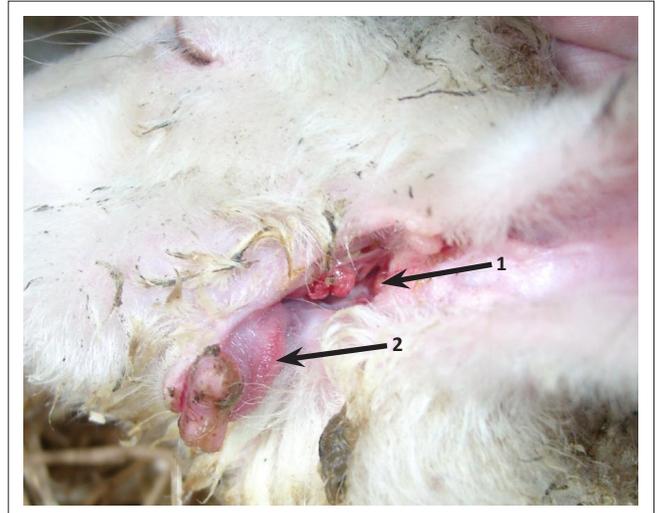


FIGURE 3: (1) Lamb 1 showing oro-auricular fistula and (2) presence of tongue tissue.



FIGURE 4: Lamb 1 with a less severe oro-auricular fistula in the right ear.

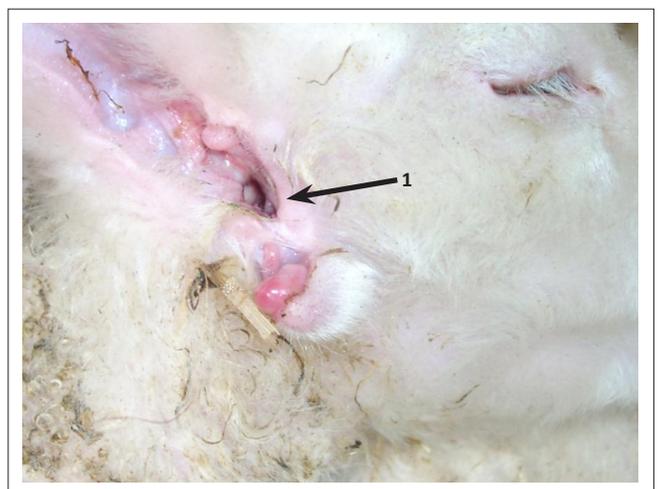


FIGURE 5: (1) Lamb 2 showing oro-auricular fistula.

where only involvement of Pouch one was evident on the right (Arey 1965; Gernecke 1985). Although Lamb 1 was able to suckle, milk spilled out of the openings and the lamb died three days later. There appeared to be tongue tissue forming part of the malformation on the left (Figure 3). On suckling the tissue that looked like tongue moved up and down in unison with the tongue. Lamb 2 suckled normally and no milk was seen to spill out of the fistula, but it also died a few days later. It is believed that the single case of mummification in this set of quadruplets (Lamb 4) was an unrelated finding.

Discussion and conclusion

The sheep flock consisted of commercial Mutton Merinos where, according to records, some inbreeding had taken place. Although not recorded or investigated at the time, another case of incomplete branchial closure had occurred some eight years prior to these two cases. The affected ewe grew to maturity and had several normal lambs until culled for age. The fistula opened just below the auricle and was lined with tissue that was macroscopically identical to buccal mucosa. The external meatus communicated with the oral cavity and saliva kept the fistula moist. This shows that these animals may survive to adulthood. Since the flock was always small, between 20 and 40 breeding ewes, it is possible that this ewe's normal offspring maintained the genetic defect in the intervening period.

The location of the incomplete closure and the fact that both lambs were male is in agreement with the findings of Rattan *et al.* (2006) who performed a study in humans, also with involvement of the second branchial cleft. What is intriguing about this case is that only two out of the four lambs born were affected, suggesting that if, as was suspected, the defect was genetic in origin the lambs arose from separate ova and thus also different sperm. The occurrence in a small flock of another case almost a decade earlier indicates that there may be a genetic link. If this is the case, a recessive gene or one with incomplete expression could be involved.

Ignoring such cases and continuing to breed with possible carriers can lead to a gradual buildup of deleterious genes, and in turn an increasing prevalence of cases, with consequent negative impact on profitability.

Acknowledgement

The authors would like to thank Ms A.M. Botha for her assistance with the digital images.

Competing interests

The authors declare that they have no financial or personal relationship(s) which may have inappropriately influenced them in writing this article.

Authors' contributions

R.L. (University of Pretoria) wrote the manuscript and collected the data on the clinical cases. K.P.P. (University of Pretoria) contributed towards the information in the introduction and discussion. G.F.B. (University of Pretoria) contributed towards the abstract wording, information in the introduction and discussion, as well as final editing.

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