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# Management of Infertility due to Unilateral Segmental Aplasia of the Paramesonephric (Müllerian) Duct in Holstein Friesian Cattle – A Case-Based Review and Update

**Christopher B Riley,** *BVSc, PhD, Dipl ACVS*[1]; **Kim Kulik,** *DVM*[1]; **Melvin Crane,** *DVM*[2]; **Frank Robblee,** *DVM*[2]; **Shawn BL McKenna,** *DVM, PhD*[1]

[1] *Atlantic Veterinary College, University of Prince Edward Island, 550 University Avenue, Charlottetown, PE, C1A 4P3*

[2] *Kensington Veterinary Clinic Ltd, 11 Park Road, Kensington, PE, C0B 1M0*

*Corresponding author: Christopher Riley; email criley@upei.ca*

*Dr Kulik's current address: 806 Belle Fourche Dr, Gillette, WY 82718*

## Abstract

Segmental aplasia of the paramesonephric (Müllerian) duct (SAP) is an infrequently reported cause of infertility, with few cases reported in North America in the last 30 years. The objectives of this paper are to report the findings and outcome of 17 Holstein Friesian cows recently diagnosed with this condition, and to review and discuss the current genetic and economic considerations in managing this condition in breeding dairy cattle. The possible genetic basis of this condition in the Holstein and other breeds is reviewed, as are the possible molecular etiologies. Management challenges and options are discussed, as are the needs for centralized reporting and elucidation of SAP in North American Holsteins.

**Keywords:** bovine, infertility, segmental aplasia

## Résumé

L'aplasie segmentaire des canaux (Müllerien) paramésonéphriques (SAP) est une cause d'infertilité rarement rapportée. En Amérique du Nord, seulement quelques cas ont été rapportés dans les 30 dernières années. L'objectif de cette étude était de rapporter les conclusions et l'issue de 17 cas impliquant des vaches Holstein-Friesian récemment diagnostiquées avec la condition. Une revue et discussion des considérations économiques et génétiques actuelles sont aussi présentées dans le contexte de la régie des vaches laitières en reproduction. Le fondement génétique probable de cette condition chez les vaches Holstein et chez d'autres races est considéré de même que les étiologies moléculaires envisageables. Les défis et les options de régie sont discutés de même que le besoin de développer un système centralisé de signalement et le besoin d'élucider le SAP chez les vaches Holstein de l'Amérique du Nord.

## Introduction

Infertility is a frequent problem in dairy cattle that may be caused by a wide variety of conditions, including those due to developmental and heritable etiologies. Segmental aplasia of the paramesonephric (Müllerian) duct (SAP) is an infrequently reported cause of infertility in the cow and other food animal species.[5-8,20,25,32,35] In humans the equivalent condition is described as Müllerian aplasia (also called Mayer-Rokitanski-Küster-Hauser syndrome), and may result in anomalies restricted to the tubular portions of the genital tract, or in anomalies affecting several body systems.[2,31] In cattle, SAP is thought to be limited to focal developmental defects of the paramesonephric ducts, resulting in partial or total developmental failure of either or both uterine horns, or the uterine body.[5,8,20,25,34] Ovarian development is not usually affected by this condition, but similar to the disease in humans, it may also cause aplastic portions of the cervix and anterior vagina, and a complete or partial persistent hymen.[5,31] This defect was first described scientifically in the early 1900s in white-bred Shorthorn cattle.[5,8] Once a rarely reported disease, an incidence of 0.15-0.45% in various cattle subpopulations around the world has been reported.[1,5,7,9,13,19,25,26] The condition has only been reported in the scientific literature in two North American Holstein cows since 1973.[5,6,20] The objectives of this paper are to report the findings

and outcome of 17 Holstein Friesian cows recently diagnosed with this condition, and to review and discuss the current genetic and economic considerations in managing this condition in breeding dairy cattle. It is hoped that this report will serve as a comprehensive review for veterinarians and producers, and that it will encourage further reporting and study of the condition in the Holstein Friesian population.

## Case Presentations

A four-year-old, 1320 lb (600 kg), Holstein Friesian heifer (case 1) was referred to the Atlantic Veterinary College (AVC) for unilateral ovariectomy with a tentative diagnosis of right uterine horn aplasia (uterus unicornis). The owner had requested examination by the referring veterinarian (RDVM) prior to using the heifer as an embryo donor. On general physical examination, findings were within normal parameters. On rectal palpation, the left uterine horn and ovary were within normal limits and follicular activity was present. The right ovary was palpated within the abdomen alongside the right body wall without apparent uterine horn attachment. Approximately 2.5 inches (6 cm) of right horn was present beyond the uterine body before ending abruptly, confirming the diagnosis of the RDVM. At the owner's request a right ovariectomy was performed using an ecraseur via a right flank approach. Following routine preoperative preparation of the right flank, a right paravertebral local anesthetic block was performed prior to a standing laparotomy. The preoperative diagnosis was confirmed, and the ovary removed without significant hemorrhage. The heifer recovered uneventfully and was discharged two days postoperatively.

A three-year and 10-months-old, 1390 lb (632 kg), first-calf Holstein Friesian heifer (case 2) was referred to the AVC from the same practice as case 1 (two months later) for unilateral ovariectomy. The heifer had been exposed to a bull for six months before conceiving the first time, and following calving was used for successful collection of an embryo. Following embryo collection the heifer suffered from a lack of ovulations on the right ovary, multiple episodes of cystic ovarian disease, and the RDVM was unable to pass an artificial insemination pipette completely down the right uterine horn. The RDVM had administered prostaglandin and GnRH to induce estrus approximately three weeks prior to presentation to the AVC. On general physical examination, findings were within normal parameters. On transrectal palpation and ultrasound evaluation, the left ovary was normal with no follicular activity present. The left uterine horn was slightly edematous, but otherwise normal. There were two enlarged follicular cysts present on the right ovary, and the right uterine horn was present with hyperechoic areas (representing fibrous or scar tissue), separating it from a large dilated area with hypoechoic fluid inside the horn lumen (mucometra). No luteal tissue was identified. Right ovarian follicular cysts, segmental stricture or fibrous occlusion of the right uterine horn and dilation of a cranial portion of the right horn with mucometra was diagnosed. Partial SAP of the right uterine horn was considered the most likely etiology.

Following aseptic skin preparation, case 2 underwent a right ovariectomy by laparoscopy via a standing right flank approach. Prior to the procedure, food was withheld for 27 hours and water for 14 hours to accommodate visualization within the abdomen, and a paravertebral local anesthetic block was performed. The laparoscopic procedure allowed visual confirmation of partial segmental aplasia of the right uterine horn. The defect was located approximately mid-horn, with the distended portion containing an accumulation of fluid anterior to the aplastic portion (Figure 1). The ovary attached to the affected segment of the right horn was removed under laparoscopic guidance.

Both heifers recovered without complications following surgery. Case 1 returned to normal cycling with follicular activity of the left ovary without exogenous drug administration postoperatively, and became pregnant after two artificial insemination attempts. Case 2 had not returned to normal cycling by six weeks postsurgery, and the RDVM found the left ovary to be cystic. Over the 12 months following surgery the heifer has remained infertile, and has had repeated episodes of cystic ovarian disease in her remaining ovary. A re-

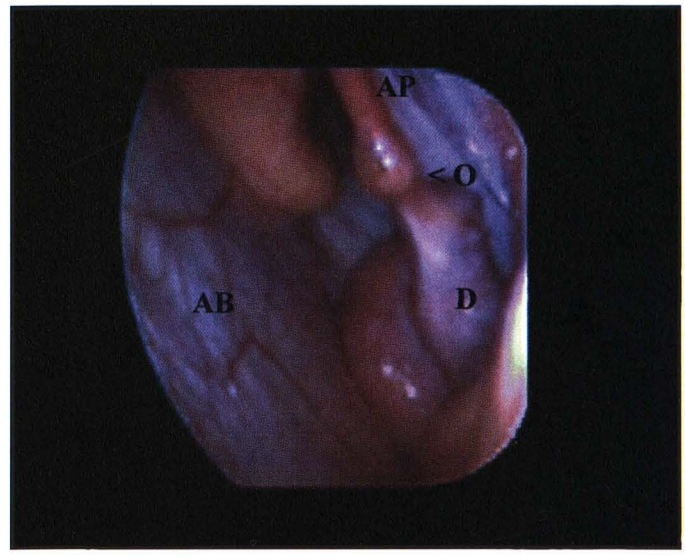


**Figure 1.** Laparoscopic view of a portion of the right uterine horn in case 2. O = start of occluded portion of right uterine horn; AP = aplastic segment of right uterine horn; D = caudal portion of right uterine horn; AB = abdominal wall.

cipient cow that had received an embryo from case 2 prior to referral had delivered an apparently normal heifer calf six months after referral of the donor.

The lack of recent information available about SAP, and the absence of clear guidelines on appropriate management for the condition in a modern dairy herd, prompted a review of records at the AVC and the Kensington Veterinary Clinic (KVC). The review found 15 additional cases had been diagnosed by veterinarians from the practices within the last five years (2001 – 2005), including six within the last six months of 2005. Signalment, relevant case history, clinical findings and outcomes for all 17 cases are summarized in Table 1. Mean age at the time of diagnosis was 26 months (data available for 15 of 17 cases). Six heifers had delivered live offspring, including five prior to diagnosis; another had produced a live offspring via embryo transfer (case 2). For nine heifers, anestrus was reported as a presenting complaint. None of the animals that had conceived were reported with a history of anestrus, however, four or more breedings were required for conception. Ovarian cystic disease was uncommon (three cases). All cases were unicornal, including 14 with SAP of the right and three with a lesion affecting the left. Aplasia was partial unicornal in six cases, and affected an entire horn in 11 cases. Two cases (described above) were managed surgically (one had calved), nine were culled (one of these had been treated medically without success), and five others kept in the herd at the time of writing (one of these had calved). Two pairs of affected animals were closely related.

## Discussion and Review

***Clinical considerations.*** History and signalment of the affected cows were typical of previously reported cases.[5,20,34] In the case of cow 2, heifer management practices may have contributed to a delay in diagnosis until

**Table 1.** Signalment, relevant case history, clinical findings, management and outcomes for 17 cases of segmental aplasia of the paramesonephric duct (SAP) in cattle diagnosed at Atlantic Veterinary College or Kensington Veterinary Clinic.

| Case | Age (yr) | Animal source | | | Parity | Anestrus reported | Breedings to conception | Ovarian cysts | Aplastic segment | Management and outcome |
|---|---|---|---|---|---|---|---|---|---|---|
| | | Cow | Sire | Dam | | | | | | |
| 1 | 4 | Farm | CAN | Farm | 0 | No | ≥ 4 | No | Right horn | Surgery –calved |
| 2 | 3 | Farm | CAN | Farm | 1 | No | ≥ 4 | L & R | Right horn (50%)[D] | Surgery – FTC |
| 3 | 4.5 | Sale | UK | UK | 2 | No | ≥ 4 | L & R | Right horn | Medical – culled |
| 4 | 3.5 | ET | USA | Farm | 1 | No | ≥ 4 | No | Right horn | None – culled |
| 5 | 1.5 | Farm | Farm | Farm | 0 | Yes | NB | No | Left horn (50%)[D] | None – kept |
| 6* | 1.5 | Farm | NR | Farm | 0 | Yes | FTC | No | Right horn (50%)[D] | None – culled |
| 7* | 1.5 | Farm | NR | Farm | 0 | Yes | FTC | No | Right horn (50%)[D] | None – culled |
| 8 | 2 | NR | NR | NR | 0 | No | > 4; FTC | No | Right horn | None – culled |
| 9 | 1.5 | Farm | UK | UK | 0 | Yes | NB | No | Right horn | None – kept |
| 10 | NR | Farm | UK | UK | >1 | No | † | No | Right horn | None – UK |
| 11 | 1.5 | Farm | UK | UK | 0 | Yes | NB | No | Right horn (25%)[D] | None – culled |
| 12** | NR | Farm | NR | NR | >1 | Yes | ≥ 4 | No | Right horn | None – culled |
| 13** | 1.5 | Farm | Local farm | NR | 0 | Yes | FTC | No | Right horn | None – culled |
| 14** | 1.5 | Farm | NR | NR | 0 | Yes | UK | No | Right horn | None – calved |
| 15 | 1.2 | Farm | UK | UK | 0 | No | FTC | No | Left horn | None – kept |
| 16 | 2.3 | CAN | CAN | | 0 | No | ≥ 4 | No | Left horn (>50%) | None – culled |
| 17 | 1.7 | Farm | Farm | Farm | 0 | Yes | FTC | Yes | Right horn | None – kept |

*Maternally related (full or maternal sisters)
**Animals from same farm (case 12 was the dam of case 13)
†Used for embryo collection – fertilization unsuccessful
CAN = semen from Canadian breeding company
USA = semen from US breeding company
FTC = failed to conceive
NB = not bred
UK = unknown
NR = not recorded
D = distal segment distended with fluid with percentage of horn affected in parentheses.

artificial breeding was attempted. Several others were not diagnosed until after conception. This would be expected to be more common in herds which breed heifers by natural cover, or breed older animals experiencing delayed conception by natural cover, prior to a thorough reproductive examination by an experienced veterinarian. Cases described in the current report that had reproduced confirm the findings of others that a previous history of successful conception and parturition does not preclude a diagnosis of unilateral SAP.[5,20,34] Cases in which there is SAP of both uterine horns, the uterine body, the cervix, or the vagina are unable to conceive.[5,25] As will be discussed below, the heifer offspring of heifers with SAP may be expected to be genetic carriers of the disease. The authors suggest that reproductive tracts of heifers (particularly in the affected herds) be examined as early as possible to determine if they are anatomically normal prior to breeding, and that all cows suffering from delayed conception be similarly examined.[27]

Although most cases may be diagnosed by simple transrectal palpation, there are some mild forms of the anatomic anomaly that may require ultrasonography or attempted passage of an insemination catheter to make an antemortem diagnosis.[5,20,25] In the case of partial uterine SAP, as seen in many of the cases described in this report, the more cranial portions of the affected horn may distend and enlarge with fluid over time, potentially resembling an early pregnancy on palpation or a paraovarian cyst, thus delaying or impeding a definitive diagnosis.[34] In some cases this sequestered fluid may become thick and firm on palpation.[34]

The most common differential diagnosis for the tubular anatomic defects of the female bovine reproductive tract is freemartinism.[23] Historical information and vaginal examination allow rapid identification of most freemartins.[23] However, a small proportion are born as singletons or have near-normal reproductive tracts, and may require laboratory testing for confirmation of the diagnosis.[23] None of the present cases displayed findings consistent with freemartinism, as all had an intact vagina and uterine body.[23] The fluid accumulation detected in some of the animals in Table 1 with partial SAP may also occur following other causes of uterine obstruction, including luminal adhesions or neoplasia, intramural neoplasia, extramural obstruction or from a primary mucometra or hydrometra.[13] In the current cases with partial SAP, concurrent mucometra may have resulted from secretions of the glandular epithelial tissue anterior to the luminal obstruction.[5,20,34] Although not observed here, rupture of the portion of uterus affected by mucometra is a possible complication of SAP, resulting in peritonitis.[30,37]

In cases of SAP, including those described in this report, ovarian development is normal and estrus cycles may be maintained in cows, ewes and women.[8,20,25,31,32,34] However, infertility and decreased reproductive performance are common signs associated with SAP. In cases of unilateral partial uterine SAP, as observed in cases 2, 5-7, 11 and 16 (Table 1), there is a failure of lumen development of a portion of the horn between the uterine body and the tip of the horn. This anomaly prevents the expulsion of endometrial secretions associated with normal estrus cycling from the segment (if patent), therefore resulting in mucometra.[20,34] It has been hypothesized that chronic distension of the uterine horn segment damages the endometrial lining, interfering with the local production of endogenous prostaglandin F2$\alpha$.[20,36] Luteolysis of the ipsilateral corpus luteum (CL) is inhibited and this may result in persistent anestrus.[36] Caution should be exercised in attempting to resolve the problem pharmacologically as attempted in case 2, as there is risk of subsequent rupture of the segment and metroperitonitis.[30,37] In cases of complete unilateral uterine horn SAP, the ovaries remain sensitive to exogenous prostaglandin, and its administration is more likely to induce CL regression on the defective side and induce estrus. As documented in several of the cows in this report, ovulation on the unaffected side may result in a normal estrous cycle and permit conception.

***The genetic basis of SAP.*** In cattle, the management recommendation for many abnormalities believed to be heritable (including SAP) has traditionally been the slaughter or euthanasia of affected animals, eliminating them from the gene pool.[5,20,25,34] This practice may be effective for removing single affected animals, but does little to remove undesirable heritable traits within unaffected heterozygous Holstein Friesian carriers from the population at large. In the current report, an affected heifer was successfully treated surgically and has been returned to use for breeding and milking. The request for surgical treatment of cases 1 and 2 placed the authors in a dilemma in providing appropriate advice on the breeding management of these valuable and high-producing animals, prompting the preparation of this case-based review. As observed here, many animals remain undiagnosed before having one or more offspring (usually unaffected, however case 12 had an affected offspring), possibly after previous medical intervention, or they may not be diagnosed until slaughter.[1,9,13,20,25,26,34] Thus, causative genetic mutations may persist at a low frequency[5,10] in a herd or offspring of heterozygous or homozygous animals. Any decision on treatment and management of Holstein Friesians with a disease that is suspected to be heritable requires in-depth knowledge of the biochemical and genetic basis of that disease, influence of environmental factors, prevalence of disease in the population and production costs (both short- and long-term) of eliminating that disease.[10]

There are approximately 350 inherited diseases in cattle, most of which occur at such low prevalence that they are generally not detected in progeny tests.[10] Currently this appears to be true for SAP in North America, with only one published investigation of disease transmission by a Holstein Friesian bull to his female offspring in over 30 years, and two case reports.[5,6,20] More recent abattoir studies of mixed-breed populations (one including Holsteins) have reported incidences of 0.45% and 0.24% in slaughterhouses in Turkey and Nigeria, respectively.[9,13] To the authors' knowledge, similar recent data have not been reported in North America for the Holstein, or for any other breed. At least two pairs of cases in this report were closely related, providing some support for a heritable etiology in Holstein Friesians in the Atlantic region of Canada.

The disease has specifically been diagnosed in Belgian Blue, Black Pied, Brahman, Highland, Holstein Friesian, Senepol, Shorthorn, Zebu and crossbred cattle.[3,5,8,12,16,18-20,25,30,34] However, heritability of SAP has only been systematically investigated in Holstein Friesian, Belgian Blue and Shorthorn breeds.[5,8,19] There is little in-depth information on the mode of inheritance of SAP specifically in the Holstein Friesian cattle population at-large.[5] The most recently published investigation (over 30 years ago) found that in an inbreeding study of one suspected SAP carrier Holstein bull, the sex-limited (female-associated) trait was inherited as an autosomal recessive, with females affected when they were homozygous for the trait.[5] These authors hypothesized that there were separate causative genes for the left and right horns, but this hypothesis has not been substantiated in any species to date. The causative gene or genes were not identified. If this mode of inheritance holds true for the heifers in the current report, it would imply that carrier bulls are also present. Limited information on familial lines was available for this report, but sires of the heifers included on-farm sires as well as high-profile, registered sires from the United States and Canada.

Recently the mode of inheritance in the Shorthorn and Belgian Blue breeds has been more clearly established based on the high frequency of association of SAP with the white-coated cows phenotype (thus incurring the name of "white heifer disease") in cattle populations segregating for the roan locus.[8] Generally, few red-spotted Shorthorns are affected, but a greater percentage of roan (red and white hairs intermingled) spotted cows are. The Belgian Blue breed has a similar coat color trimorphism of black-spotted, blue-spotted (white and black hairs intermingled) and white, with a percentage of blue-spotted cows affected in much the same manner as the roan Shorthorns.[8] Coat color variation in these two breeds is under the influence of a single autosomal locus (the roan locus on chromosome 5) characterized by co-dominant alleles with incomplete penetrance, the latter explaining the less frequent occurrence of SAP in non-white cows.[8] Neither the roan locus nor other coat color associations with SAP have been demonstrated in Holstein Friesians.[8] Based upon this difference, we contend that when SAP occurs in this breed it should not be referred to as white heifer disease. In humans, sex-limited (female) autosomal dominant inheritance in a group of Saskatchewan families has been described, but more recently non-genetic causes and polygenic/multifactorial inheritance have been hypothesized to account for most cases in women.[31]

***The molecular basis of SAP.*** There has been limited work in cattle to elucidate the molecular basis of SAP, with the only supportive studies performed in the Belgian Blue population.[8,29] A missense mutation of the bovine mast cell growth factor gene (MGF; also called stem cell factor) has been associated with the roan phenotype in the Belgian Blue and Shorthorn breeds.[29] This work was based on the predicted nucleotide sequence for bovine MGF, for which the full gene (including introns) has not been elucidated to date.[4] Although their work alludes to a link with SAP, these workers did not attempt to demonstrate a direct relationship between this mutation and SAP.[8,29] The authors of the current manuscript conducted a search of the National Center of Biotechnology Molecular Biology Database Collection (NCBI-MDB) and found that sheep, goats and pigs, in which SAP has also been reported, have stem cell factor mRNA homologies of 98, 98 and 96%, respectively, with bovine MGF.[11] It is possible that these species may serve as models to determine the significance of MGF in bovine SAP.

In human and bovine developmental biology fields, there has been a significant focus on the role of anti-Müllerian hormone (AMH, also called Müllerian inhibiting substance) and its receptors in the development of genital malformation.[17,22,24,28] In cattle the paramesonephric ducts develop during the sixth week of gestation in the fetus, giving rise to a pair of tubular structures, each containing a lumen.[21] In the male fetus AMH is produced by the Sertoli cells, inducing regression of the paramesonephric ducts.[17] In Holstein heifer calves, low levels of AMH are produced by the granulosa cells that are present at birth, and are maintained at a consistent level over the first year of life.[28] In males and freemartins, initial levels at birth are seven-fold higher than those of the heifers, with levels in freemartins declining within the first two weeks of life to a level not significantly different from the heifers, but markedly lower than that of the young bulls.[28] It is possible that SAP in cattle occurs as a result of early *in utero* release of ovarian AMH, or due to a genetic aberration of AMH or its receptor molecules in the female

fetus. Although there are no data to support the first hypothesis in the cow, bovine values for AMH are markedly higher than those reported in humans.[28] In humans, the female fetus does not express AMH until the last month of gestation.[24] To the authors' knowledge, fetal levels in the bovine female have not been reported. Human studies have failed to demonstrate a role for aberrant AMH expression in cases of Müllerian aplasia (SAP).[22,40] However, a mouse model in which a mutated gene for bone morphogenic protein receptor 1A (BMPR-1A), an AMH type I receptor, was associated with uterine aplasia.[17] In a search of the NCBI-MDB we found that although this gene has not been sequenced in cattle, several genetic variants have been predicted by automated computational analysis to occur on bovine chromosome 28.[11] This may provide a focus for future research into the molecular basis of SAP in the Holstein.

***Management considerations for SAP.*** The choice of management options for SAP in milking Holsteins depends upon production and breeding goals, and the financial impact of the disease on the producer. Unilaterally affected animals may be medically managed or surgically corrected to allow conception. Medical management would consist of cycle-to-cycle management, including prostaglandin administration when needed. Surgical correction consists of removing the ovary on the aplastic side, therefore removing the potential for an ovulation from the non-patent side. A pregnancy in the unaffected horn is possible with both forms of treatment, or as described in this report and others, without treatment at a decreased conception rate.[34] In previous reports of similarly affected cases euthanasia has been recommended, irrespective of the parity of the affected cows.[5,20] To the authors' knowledge, this paper constitutes the first report of surgical management of this condition. In the two surgically managed cases described in this report, both cows were valuable high-producing animals initially intended as embryo transfer donors. One animal has been successfully returned to reproductive activity and milk production. As observed in case 1, the normal ovary is capable of maintaining cyclic activity.

If the owner chooses to maintain heifers with SAP in the herd, the authors recommend that their genetic contribution to the herd be limited by avoiding their use as embryo transfer donors, and minimizing breeding to the same or related sires.[16] A better alternative, given the lack of a genetic test for SAP, may be to breed to a beef sire and divert the offspring to meat production or to use as embryo recipients. This would enable the producer to optimize milk returns on the animal, and allow more time for obtaining a herd replacement. Consideration should be given to notifying owners of the bulls used to sire affected heifers. Alternatively, veterinarians encountering SAP may choose to submit a "Report Form for Abnormalities" available from various dairy cattle associations, including the Holstein-Friesian Association of America. The current data available for SAP in the Holstein suggests it is a heritable condition, however it is impossible to state with certainty that the mutation is these cases is heritable or due to nongenetic factors without a test for the genetic defect. The authors suggest that North American Holstein breed associations establish a database of SAP cases and actively solicit and collect pedigree and other information. This information could then be used to determine if SAP in the Holstein breed is heritable or due to non-genetic factors, and provide support for genetic studies of the disease if indicated.

Currently there are commercial tests available for identifying Holstein carriers (heterozygous) of the inherited diseases citrullinemia, deficiency of uridine monophosphate synthase (DUMPS), bovine leukocyte adhesion deficiency (BLAD), and complex vertebral malformation (CVM).[10,14,35] For each of these conditions, a major high-profile Holstein sire or sires has been implicated which, in addition to the recessive genetic defects, carry highly desirable production traits.[14] Consequently, a testing strategy has been adopted by the Holstein Association USA whereby genes for the most recent high-profile defect (CVM) are maintained in the national herd, and tested animals heterozygous for CVM are not bred to other heterozygotes.[16] However, strategies of this type may become more difficult over time as the North American Holstein Friesian gene pool becomes smaller based on specific production traits, and the distribution of recessive genes becomes more widespread in the population.[38] Other more complex strategies have been developed for reduction of recessive anomalies in Holstein cattle, but until a genetic marker for SAP in this breed has been determined, their usefulness is limited.[33,38] The World Holstein Friesian Association has developed comprehensive guidelines for reporting recessive genetic anomalies, and a research plan to investigate and classify these defects.[39] However, in a recent communication between one of the authors (Riley) and an officer of the organization, it was indicated that SAP was not currently considered a problem in the breed. The authors hypothesize that under-reporting of the disease may be occurring due to economic and reputation concerns of breeders and producers, or due to failure to diagnose this condition. It is clear from the current limited retrospective review of cases in a small region of North America that the disease is present, and that affected heifers continue to produce viable offspring that perpetuate the carrier state for the disease. The current report demonstrated that the means exist to return heifers affected with SAP to production. However, owners should be counseled on the

possible genetic impact of maintaining these heifers in their breeding lines due to the likely heritable nature of SAP. An aplastic cow (homozygous) may produce carriers or affected offspring. Breeding to a carrier bull will increase the chances of producing aplastic heifers. The owners of cases 1 and 2 were advised of the probable heritable nature of the condition, but because of the high value of the animals, they opted for surgery to maintain them in the breeding lines of the representative farms. The traditional management option for many genetic diseases in animals has been the culling of the affected animals. In the case of SAP, this would require ending the animal's life for a defect that is not life threatening, but has negative effects on production. This approach is simplistic in that it fails to address the carrier state of the disease, which is especially difficult as there is currently not a screening test for SAP available. The authors suggest that the Holstein Friesian breed associations in North America undertake the development of a screening test for SAP, enabling both an estimate of disease prevalence, and to provide a tool for control of SAP either by culling or management practices similar to those already in place for other heritable diseases in North American Holstein Friesians.

## Conclusions

Uterine aplasia should be considered as a differential diagnosis in cases of recurrent anestrus or delayed conception, especially in animals with a distended uterus. Unilaterally affected animals may be medically managed or surgically corrected to allow normal pregnancies, although some affected females may have delivered viable offspring prior to diagnosis. The authors recommend limiting the genetic contribution of SAP-affected animals to the herd, and avoiding their use in embryo transfer or their offspring as replacements. Heterozygous and some homozygous heifers with SAP will continue to affect the North American Holstein Friesian herd until such time as screening test and accompanying management plans have been implemented. Identification of the causative genetic anomaly, development of screening tests to identify heterozygous carriers and a recording of all cases of SAP in Holstein Friesians in a centralized database are all necessary steps to restrict the occurrence of SAP in this breed.

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# Abstract

## LAMECOW at The University of Warwick: A Study of Lameness in Dairy Cattle

Amory J.R., Barker Z.E., Wright J.L., Mason S.A., Blowey R.W., Green L.E.

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This paper describes some of the work carried out by the University of Warwick as part of the EU project LAMECOW. A study was undertaken on 49 dairy farms in England and Wales to determine risk factors for impaired locomotion measured using a 3 point score based on back posture. Factors associated with increased mean locomotion score (*i.e.* poor locomotion) included variables related to management of dry cows and heifers, housing design, diet and hoof trimming. An intervention study monitored the uptake and success of standardized recommendations given by a veterinarian to 25 treatment farms compared with 24 control farms. Farmers were prepared to adopt only some of the recommendations made, typically those with least cost. At the end of a one year study there was a reduction in lame cattle (score 3) on treatment farms and a net reduction in sole ulcers compared with control farms.

A multivariable statistical analysis of milk recording data and hoof lesion incidence data from 30 farms indicated that there were significant milk losses associated with the occurrence of sole ulcer and white line disease, but not associated with digital dermatitis. These clarify earlier findings on which lesions are associated with reduced milk yield. We conclude that an important part of reducing lameness is in improving the uptake of recommendations for best practice management that would be aided by further quantification of economic cost.