

Panel discussion 1: opportunities and difficulties in multi-disciplinary and multi-actor research

Jaap J. van Milgen, Marie-Hélène Pinard-van Der Laan

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	Combining clinical mastitis with somatic cell indicators for udder health selection in Spanish cows M.A. Peréz-Cabal and N. Charfeddine	535
	Investigations of the variation of horn phenotypes and the genetic architecture of scurs in cattle L.J. Gehrke, D. Seichter, I. Ruß, I. Medugorac, C. Scheper, S. König, J. Tetens and G. Thaller	536
	Relationships between subclinical ketosis, BCS, fat-protein-ratio and other diseases in Fleckvieh B. Fuerst-Waltl, A. Köck, F. Steininger, C. Fuerst and C. Egger-Danner	536
	Poster Session 55	
	Thermal comfort indexes and physiological variables of Caracu cattle under thermal stress S.B.G.P.N.P. Lima, A.P. Freitas, N.T. Bazon, R.P. Savegnago, R.H. Branco and C.C.P. Paz	537
	Correlation between heat tolerance and residual feed intake in Caracu cattle under thermal stress S.B.G.P.N.P. Lima, A.P. Freitas, R.P. Savegnago, N.T. Bazon, J.N.S.G. Cyrillo, J.A. Negrão and C.C.P. Paz	537
	Combined selection for milk and weigth in cattle in the tropical environment R.R. Rizzi, M.O. Oropeza, F.C. Cerutti and J.C.A. Alvarez	538
	Heat tolerance or extensive ability to acclimate A. Geraldo, F. Silva, C. Pinheiro, L. Cachucho, C. Matos, E. Lamy, F. Capela E Silva, P. Infante and A. Pereira	538
	Session 56. Multidisciplinary approaches for improving sustainable livesto production: research needs, opportunities and difficulties	ck
	Date: Thursday 30 August 2018; 8.30 – 12.30 Chair: J. Van Milgen / M.H. Pinard-Van der Laan	
	Theatre Session 56	
	Multidisciplinary approaches to livestock production J. Van Milgen, M.H. Pinard-Van Der Laan, E. Schwartz, Ç. Kaya and V. Heuzé	539
d	Twists and turns of interdisciplinary work in resarch projects: which conditions and achievements ? M. Cerf	539
	Detection and characterization of the feed intake response of growing pigs to perturbations <i>H. Nguyen Ba, M. Taghipoor and J. Van Milgen</i>	540
	Layers response to a suboptimal diet through phenotype and transcriptome changes in four tissues F. Jehl, M. Brenet, A. Rau, C. Désert, M. Boutin, S. Leroux, D. Esquerré, C. Klopp, D. Gourichon, A. Collin, F. Pitel, T. Zerjal and S. Lagarrigue	540
	What potential of genome-wide integrative approaches to predict vaccine responses? F. Blanc, T. Maroilley, M.H. Pinard-Van Der Laan, G. Lemonnier, J.J. Leplat, E. Bouguyon, Y. Billon, J.P. Bidanel, B. Bed'hom, J. Estellé, S. Kim, L. Vervelde, D. Blake and C. Rogel-Gaillard	541
	Immune responses after administration of innovative <i>Myocoplasma hyopneumoniae</i> bacterins in pigs D. Maes, A. Matthijs, G. Auray, C. Barnier-Quer, F. Boyen, I. Arsenakis, A. Michiels, F. Haesebrouck and A. Summerfield	541
	Effect of heat stress on faecal microbiota composition in swine: preliminary results M. Le Sciellour, I. Hochu, O. Zemb, J. Riquet, H. Gilbert, M. Giorgi, Y. Billon, JL. Gourdine and D. Renaudeau	542

invite

	The socio-economic evaluation of vaccines in livestock systems C. Bellet and J. Rushton	542
	Panel discussion 1: opportunities and difficulties in multi-disciplinary and multi-actor research <i>J. Van Milgen and M.H. Pinard-Van Der Laan</i>	543
	Session 57. Overcoming technological barriers in sheep and goat production and breeding	on
	Date: Thursday 30 August 2018; 8.30 – 11.30 Chair: J. Yates	
	Theatre Session 57	
	Development of and Imputation with a SNP map derived from the latest reference genome sequence X. Yu, J.C. McEwan, J.H. Jakobsen and T.H.E. Meuwissen	543
	Light-treated rams and bucks abolish reproductive seasonality in sheep and goats <i>P. Chemineau, J.A. Abecia, M. Keller and J.A. Delgadillo</i>	544
	Evaluation of accelerometers as an effective tool to measure sheep behaviour in a pastoral context <i>P.G. Grisot, A. Philibert, F. Demarquet and A. Aupiais</i>	544
	Low cost portable microwave system for non-destructive measurement of carcass fat depth J. Marimuthu and G.E. Gardner	545
	Benefits for sheep farmers of monitoring grass growth, quality and utilisation A.E. Aubry	545
	Investigating factors affecting lifetime performance in ewes on a network of commercial farms <i>E. Genever, H. King and N. Wright</i>	546
	Use of electronic identification (EID) associated technologies in marginal sheep farming systems C. Morgan-Davies, J.M. Gautier, B. Vosough-Ahmadi, P. Creighton, A. Barnes, R. Corner-Thomas, S. Schmoelzl and D. McCracken	546
	The value of information from commercial livestock in multi-tier sheep breeding schemes B.F.S. Santos, J.H.J. Van Der Werf, T.J. Byrne, J.P. Gibson and P.R. Amer	547
	Poster Session 57	
	Productive and selenium status in lambs affected by selenium biofortified corn-preliminary results <i>Z. Antunovic, Z. Klir, Z. Loncaric and J. Novoselec</i>	547
	Session 58. Non-invasive biomarkers in nutritional studies	
	Date: Thursday 30 August 2018; 8.30 – 11.30 Chair: R.M.A. Goselink	
	Theatre Session 58	
invited	Faecal biomarkers for intestinal health in nutritional studies T.A. Niewold	548
	Development of a new ELISA test for pancreatitis associated protein detection in pig <i>E. Mariani, G. Savoini and T.A. Niewold</i>	548
	TARD ooth A LIM II D. I. II Ooto	

Session 56 Theatre 9

Panel discussion 1: opportunities and difficulties in multi-disciplinary and multi-actor research

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Panel discussion with stakeholders and interaction with participants on the opportunities and difficulties of multidisciplinary and multiactor research.

Session 57 Theatre 1

Development of and Imputation with a SNP map derived from the latest reference genome sequence

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SNP chips of different densities are often used together on farm animals for economic reasons. The missing genotypes on the low-density (LD) chips can be imputed with genotype results of high-density (HD) chips and a linkage map of HD loci. Genome references and SNP chips are under continuous development. This may introduce several problems. For example, different chips may be based on different versions of the reference sequence. The SNP names can come from different naming systems. There are also typically many duplicated loci, because of SNP types and importance. Any one of these problems can increase imputation errors. One solution for these problems is to derive a SNP map from the most up-to-date reference sequence. The probes that come with the manifest of a chip-design can be used to position the SNPs on this map. Each of these probes is 50 base pairs (bp) in length, and can uniquely define a mutation position. Because the ends of the probes may also be the SNP, one bp was trimmed from both ends of a probe. All possible 48-bp sequences of a chromosome were then indexed by their positions and sorted in alphabet order. This can speed up the search procedure thousands of times, for sequential searches were converted to binary ones. This method was tested on data from Norwegian White Sheep genotyped with an 8k LD chip and a 600k HD chip. Random animals who were genotyped with the HD chip were masked at the missing loci on the LD chip. Only autosome loci were considered. Using the provided 600k linkage map, the correct allelic imputation rate was only 85%. Using the map derived from the sheep genome reference version 3.1.91, the imputation rate increased to 92%. When the number of animals with known HD genotypes increased from 120 to 617, the imputation rate increased to 94%, indicating the accuracy improvement was mainly from the new map. In conclusion, using a SNP map derived from the latest reference can greatly increase imputation rate. The described binary search algorithm makes such map construction feasible with limited computation costs.