



# Advances in Fibre Production Science in South American Camelids and other Fibre Animals

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Martina Gerken, Carlo Renieri, Daniel Allain, Hugh Galbraith, Juan Pablo Gutiérrez, Lisa McKenna, Roman Niznikowski, Maria Wurzinger (eds.)

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## **Preface**

Animal fibres from South American camelids and other fibre or wool bearing species provide important products for use by the human population. The contemporary context includes the competition with petrocarbon-based artificial fibres and concern about excessive persistence of these in the natural environment. Animal fibres present highly valuable characteristics for sustainable production and processing as they are both natural and renewable. On the other hand, their use is recognised to depend on availability of appropriate quality and quantity, the production of which is underpinned by a range of sciences and processes which support development to meet market requirements.

Such support includes the efforts of the Animal Fibre Working Group (AFWG) of the European Federation of Animal Sciences (EAAP) which was instituted in 2007 and tasked with creating a network for investigation and dissemination of information in Europe and internationally. One task has been the organisation of scientific meetings, and continuing the tradition of previous European Symposia on South American camelids. These include the recent 5<sup>th</sup> Meeting in Sevilla (Spain: 2010) and 6<sup>th</sup> Meeting at EAAP, Nantes (France: 2013). References to these and other meetings, workshops and publications may be found on the AFWG website: <http://www.eaap.org/presentation/scientific-structure/commissions-working-groups/animal-fiber-working-group/>.

The present publication derives from the 7<sup>th</sup> European Symposium on South American Camelids and 3<sup>rd</sup> European Meeting on Fibre Animals (<http://www.sympcam.org/>). This meeting was held in the conference facility of the Domus Pacis Hotel, Assisi, Italy, on 12 to 14 June 2017. It was organised by Prof Dr Carlo Renieri and his colleagues Dr Attilio De Cosmo, Dr Francesco Fantuz, Dr Antonietta La Terza, Prof Alessandro Valbonesi (University of Camerino), Dr Marco Antonini (ENEA), and Maurizio Gubbiotti (University Marconi, Roma) with support from the scientific board comprising AFWG colleagues. We wish to thank Dario Pediconi, Cristina Nocelli, Irene Pazzaglia, Stefano Pallotti (University of Camerino) who helped us during the symposium. We also thank all participants who readily agreed to chair sessions or to participate in the Round Table.

We are very grateful to Loro Piana (<http://www.sympcam.org/loropiana/>) for generous funding support which enabled the attendance of international speakers and provided scholarships for three young scientists from Latin American countries.

Individual papers and abstracts, where full papers were not available, were printed from the manuscripts supplied by the authors. The assistance of Marvin Heuduck (Göttingen University) and the editors of Göttingen University Press in the editorial process is acknowledged.

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The editors



# Table of contents

Preface.....	5
<b>Sustainable Development, Climate Change and Biodiversity</b>	
Sustainable Development of Livestock Production: What and how can Research Contribute?.....	15
<i>M. Wurzinger</i>	
Animal Fibre Production in Europe: Biology, Species, Breeds and Contemporary Utilisation.....	23
<i>H. Galbraith</i>	
Effect of Technological Alternatives in the Mitigation of Climate Change in the Aging of Alpacas above 4.000 msnm Puno-Peru .....	43
<i>T. Huanca, R.H. Mamani-Cato, M. Naveros and M. Gonzales</i>	
Collection of Diversity – Preserving Rare Indigenous Sheep Breeds in Germany.....	47
<i>N. Ketterle</i>	
<b>Breeding and Genetics</b>	
Advances in Llama ( <i>Llama glama</i> ) Coat Color Genetics.....	57
<i>M.S. Daverio, M. Anello, L. Vidal-Rioja and F. Di Rocco</i>	
Characterization and Expression Analysis of SLC7A11 in Llamas.....	63
<i>M. Anello, E. Fernandez, M. Silbestro, F. Veiga, L. Vidal Rioja and F. Di Rocco</i>	
PCR-RFLP Method for Testing ASIP EXON 4 Mutations in Llamas.....	71
<i>M.S. Daverio, V. Alcoela-Ersinger, M. Anello, L. Vidal-Rioja and F. Di Rocco</i>	
Heredabilidad estimada de fibras meduladas en alpaca huacaya.....	77
<i>R. Pinares, A. Cruz, R. Morante, I. Cervantes, A. Burgos, G. Gutiérrez, J.P. Gutiérrez</i>	
Performance Evaluation of Llama, Alpaca and Sheep Herds of a Community in Pasco, Peru.....	83
<i>D.M. Pizarro, G.A. Gutiérrez, J.A. Ñaupari and M. Wurzinger</i>	
The Camelid Registry LAREU: What Are We Breeding In Europe?.....	97
<i>C. Kiesling</i>	
Comparación de los criterios de selección de los productores con el reglamento oficial para llamas en el Perú .....	111
<i>D.Y. Calderon, M. Wurzinger, J.G. Mendoza and G.A. Gutiérrez</i>	

Selection and Evaluation of Fiber Characteristics of an Extreme Fine Alpaca Strain at Victory Farm in Missouri .....	121
<i>T. Wuliji</i>	
Merino Breeding Program Improves Wool Quality in US Wool Sheep Flocks ....	135
<i>T. Wuliji, L. Wuri, H. Glimp and T. Filbin</i>	
Selection Strategies for Fiber Quality in Alashan Cashmere Goat.....	149
<i>M. Antonini, P.R. Tang, F. Panella, G. Attard, E. Lasagna, S. Ceccobelli and F.M. Sarti</i>	
Interaction between ASIP and MC1R in Black and Brown Alpaca.....	163
<i>C. Bathrachalam, C. Nocelli, I. Pazçaglia, S. Pallotti, D. Pediconi, A. La Terça and C. Renieri</i>	
Alpaca FGF5: Hypothetical Post-Transcriptional Readthrough Regulation in Skin Biopsies.....	171
<i>Pallotti S., Pediconi D., Morelli M.B., Dbaraneedharan Subramanian, Molina M.G., Antonini M., Renieri C. and La Terça A.</i>	
Alpines Steinschaf (Alpine Stonesheep) .....	185
<i>Christian Mendel, Isabelle A. Ketterle</i>	
<b>Reproduction and Pathology</b>	
The Alpaca Cria, Clinical and Immunological Aspects.....	195
<i>P. Walter Bravo</i>	
Addition of Seminal Plasma to Frozen-Thawed Llama Spermatozoa does not Preserve Sperm Motility.....	201
<i>Fumuso, F.G., Carretero, M.I., Chaves, M.G., Neild, D.M., Miragaya, M.H. and Giuliano, S.M.</i>	
Alpaca Semen Quality throughout the Breeding Period.....	213
<i>P. Walter Bravo, W. Garcia and V. Alarcon</i>	
The Sperm Chromatin Dispersion Assay (HALO Test) Correlates with the Tunel Technique in Llama Sperm.....	221
<i>M.I. Carretero, F.G. Fumuso, S.M. Giuliano, D.M. Neild, P. Cetica and M.H. Miragaya</i>	
Teeth in Camelids: Myths, Facts and Problems .....	229
<i>I. Gunsler</i>	
<b>Nutrition</b>	
Advances in Nutrition on Chinese Cashmere Goat: A Review.....	239
<i>Sun Haiçhona, Li Shenglia, Zhang Chongçhia, Jin Lua, Sang Dana and Zhang Chunbuaa</i>	

Alfalfa Hay Supplementation to Improve Llama Meat Production for Smallholders in Pasco Region, Peru.....	255
<i>G. Gutierrez, A. Corredor, R. Robles, J. Mendoza, V. Hidalgo and M. Wurzinger</i>	

Water Metabolism in South American Camelids .....	267
<i>M. Gerken, L. Brinkmann, R. Amin Runa and A. Riek</i>	

### **Meat and Fibre Production, Fibre Metrology**

Carne y charqui de llama .....	279
<i>C. Ayala, G. Condori, C. Renieri, S. Pilco and J.L. Quispe</i>	

Wool Scouring in Europe: Urgent and Ecological Solutions.....	301
<i>M.T. Chaupin</i>	

Proteomic Method for Determination of Animal Hair Fibres .....	305
<i>C. Tonetti, S. Paoletta, D.O. Sanchez Ramirez, R.A. Carletto, C. Vineis, A. Varesano and S. Sforza</i>	

The Use of Near-infrared (NIR) Reflectance Spectroscopy to Predict Mohair Quality in Greasy Fleece Samples of Angora Goats .....	313
<i>D. Allain, S. Brenot, G. Awinet, B. Pena-Arnaud and P. Martin</i>	

Variability of Fiber Quality of Chinese Alashan Left Banner White Cashmere goat .....	325
<i>S. Pallotti, J. Wang, P. Tang, M. Antonini, Y. Lou, C. Pieramati, A. Valbonesi and C. Renieri</i>	

Effects of Year and Sampling Site on Mean Fibre Diameter of Alashan Cashmere Goat .....	333
<i>Marco Antonini, Jun Wang, Yujie Lou, Peirong Tang, Carlo Renieri, Irene Pazzaglia, Alessandro Valbonesi</i>	

### **Abstracts**

Sustainable Cashmere, Pastoralism, and Coexistence with Predators in Europe ..	341
<i>N. Kravis</i>	

Efecto de la precipitación pluvial en la seja de selva y la zona alto andina de la región Puno sobre la producción ganadera de altura .....	342
<i>Pineda B., Zeballos J., Mamani R. and Huanca T.</i>	

Evaluation of Population and Social Composition of Vicunas ( <i>Vicugna vicugna</i> ) in Different Environment Sites of the Laguna Blanca Biosphere Reserve (Catamarca, Argentina) .....	343
<i>Riva de Neyra, L. A., Hick, M.V.H. and Frank, E. N.</i>	

Animal Welfare Problems in South American Camelids Kept in Europe .....	344
<i>Gauly, M.</i>	

Breeding Objectives for Alpacas of the Highlands Central of Peru .....	345
<i>Candio, J.R. and Gutiérrez, G.A.</i>	
Vicugna Pacos As1-Casein: Identification of New Polymorphisms at the Csn1s1 Gene .....	346
<i>Erhardt, G., Gu, M., Wagner, H., Di Stasio, L. and Paucillo, A.</i>	
Estimación de la heredabilidad de seis caracteres de calidad de fibra de alpacas huacaya del INIA Puno .....	347
<i>Mamani-Cato, R.H., Huanca, T., Pineda, M., Naveros, M. and Gallegos, R.</i>	
Effect of the Brown Coat-Coding Gene (Typr-1) on Wool and Skin Color of Żelaźnińska and Wrzosówka Sheep .....	348
<i>Niżnikowski, R., Świątek, M. and Zymańska, Z.</i>	
Relationship between Classes Assigned by Visual Appraisal and a Selection Index in Function of Live Weight, Fleece Weight and Fiber Diameter in Huacaya Alpacas from Pasco.....	349
<i>Corredor F.A. and Gutiérrez G.</i>	
Preliminary Comparative Analysis and Localization of <i>Bos Taurus</i> SNPS on <i>Vicugna Pacos</i> Chromosome 10 (Vpa10).....	350
<i>Farfán K.A., Gutiérrez G.A. and Ponce de León F.A.</i>	
Innovative Andrological Evaluation to Optimize the Selection of Fiber Animal.....	351
<i>Stelletta, C.</i>	
Use of Seminal Plasma on Interval to Ovulation, Susceptibility of Corpus Luteum to Prostaglandin and Improving of Reproductive Performance in Alpacas ( <i>Vicugna Pacos</i> ) under Peruvian Highland Conditions .....	352
<i>Huanca, W., Turin, J., Huanca, W.F., Mamani, C., Sanchez, S. and Cordero, A.</i>	
Induction of Superovulation in Alpacas According to the Number of Follicles Recruited to the Emergence of Follicular Wave .....	354
<i>Pozo A., Vásquez A., Zevallos J., Olivera L., Cordero A. and Huanca W.</i>	
Farmers Wool and Traceability .....	355
<i>Thompson, N.</i>	
Feed Intake and Animal Behaviour of Alpaca and Llamas Co-Grazing on Andean Highlands in Peru.....	356
<i>Hoehn D., Castro-Montoya J., Gomez C. and Dickhoefer U.</i>	
Daily and Seasonal Changes in Body Temperature and Activity Patterns of Llamas in the High Andes of Peru .....	358
<i>Riek, A., Stölzl, A., Marquina Bernedo, R. and Gerken, M.</i>	

Blood Levels of Phosphorus in Pubescent Alpaca ( <i>Vicugna Pacos</i> ) and the Effect of Dietary Phosphorus on Growth of Female Alpacas Post Weaning in Peruvian Andes .....	360
<i>Quispe, C.E., Ancco, E., Van Saun, R. and Gomez, C.</i>	
Digestibility of Bean Pulp Granulated in Rabbits .....	362
<i>Arce, O., Alagón, G., Ródenas, L., Martínez-Paredes, E., Moya, V.J., Pascual, J. and Cervera, C.</i>	
Correlation between Diameter of Fiber, Medulation and Ancestrality in Alpacas.....	363
<i>Melo, C., Zapata, C. and Bravo, W.</i>	
Apelin, a New Adipokine Acting on Hair Follicle: an Immunohistochemical Study on Ovine Skin.....	364
<i>Mercati, F., Dall'Aglio, C., Guelfi, G., Scocco, P. and Ceccarelli, P.</i>	
ICAR – Guideline for the Animal Fibre Production in Alpaca and Cashmere and New Rules for the Organization of the Fibre and Fleece Collection Centers .....	365
<i>Antonini, M., Pazzaglia, I., Nocelli, C., Lou, Y. and Thompson, N.</i>	
Technological Characteristics of White and Colourised Huacaya Alpaca Fibre in Apurimac, Perú .....	366
<i>Corredor, F.A., Bustinza, V., Machaca, V., Paucara, V., Paúcar, R. and Quispe, E.C.</i>	
The Prickling Issue in Fabrics Made of Camelid Fibres: Possible Mechanical or Genetic Solutions .....	367
<i>Frank E.N.</i>	
Determination of the Optimal Number of Runs Using AM2 Dehairing Technology in Fibers of Patagonian Goats (Patagonian Cashmere) .....	368
<i>Frank, E.N., Hick, M.V.H., Castillo, M.F. and Frondizj Seghetti, D.G.</i>	
Dehairing of Alpaca Fibres Top with Am2 Dehairing Technology .....	369
<i>Frank, E.N., Frondizj Seghetti, D.G., Hick, M.V.H., Castillo, M.F., Burgos, A. and Cruz, A.</i>	
Modelación de curvas de crecimiento de llamas q'ara utilizando modelos de crecimiento no lineales .....	370
<i>Mamani-Cato, R.H., Huanca, T., Naveros, M. and Gallegos, R.</i>	
Genetic Basis of Early Activation of Hair Follicle in Cashmere Goat: An Approach with Candidate Genes .....	371
<i>Pazzaglia, I., Mercati, F., Antonini, M., La Terza, A., Nocelli, C., Pallotti, S., Pediconi, D. and Renieri C.</i>	

# Advances in Llama (*Llama glama*) Coat Color Genetics

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**Abstract.** Domestic species present a wide phenotypic diversity. As their genomic sequences become available, finding the connection between the genotype and the observed phenotype is a goal possible to achieve. In several species, the genes and mutations responsible for the different coat colors and patterns have already been identified. Color is a Mendelian trait, with little or no environmental influence. Moreover, the biochemical pathway of pigment synthesis is well-known and highly conserved among mammals. Thus, in species such as llama whose complete genome has not yet been sequenced, the study of candidate genes results a valid alternative to identify the genetic variation responsible for different phenotypes. The objective of this work is to present the recent advances in the knowledge of the genes that control the coat color in llamas and compare our results with those published for alpaca.

**Resumen.** Las especies domésticas presentan una amplia diversidad fenotípica. A medida que sus secuencias genómicas están disponibles, encontrar la conexión entre el genotipo y el fenotipo observado es una meta posible de lograr. En varias especies, ya se han identificado los genes y mutaciones responsables de los diferentes colores de capa. El color es un carácter mendeliano, con poca o ninguna influencia ambiental. Además, la ruta bioquímica de la síntesis de la melanina es bien conocida y altamente conservada entre los mamíferos. Por lo tanto, en especies como la llama, cuyo genoma completo aún no ha sido secuenciado, el estudio de genes candidatos resulta una alternativa válida para identificar la variación genética responsable de diferentes fenotipos. El objetivo de este trabajo es presentar los recientes avances en el conocimiento de los genes que controlan el color de capa en llamas y comparar nuestros resultados con los publicados para la alpaca.

**Keywords:** llamas, coat color, MC1R, ASIP, polymorphisms

Melanogenesis is the process of forming melanin, a pigment that gives color to the skin, hair and eyes. The melanin synthesis is produced in the melanosome, a specialized organelle inside the melanocyte. The two types of pigments that are

responsible for color are eumelanin (dark brown/black) and pheomelanin (red/yellow). The ratio between both pigments is controlled by the interaction between the melanocortin receptor (MC1R) and its antagonist, the agouti signaling protein (ASIP). The binding of the  $\alpha$ -melanocyte-stimulating hormone ( $\alpha$ -MSH), which is the natural ligand of the MC1R, causes the exclusive production of eumelanin, whereas Mc1r inhibition by ASIP leads to the synthesis of pheomelanin (Lu et al., 1994). Polymorphisms in the genes encoding these two proteins have been associated with coat color variation in mammals and birds (Kijas et al., 1998; Andersson et al 2003, Schmutz et al., 2003; 2003; Kerns et al., 2004).

In many species, dominant MC1R alleles (E) produce a uniform black color, while recessive alleles (e) produce red-yellow pigment. Conversely, dominant alleles of the Agouti locus produce a yellow coat whereas the recessive allele in homozygosis is associated with a black coat. In addition to mutations in the genes that control the type of pigment produced, mutations in other genes related to melanogenesis also affect the coat color. For example, genes that affect correct migration and differentiation of the melanocytes, such as KIT or MITF are responsible for unpigmented or spotted phenotypes in most species (Brooks and Bailey, 2005; Pielberg et al., 2002; Cooper et al., 2005). Llamas and alpacas are characterized by a wide variety of coat colors and patterns, although molecular basis of coat color determination is not yet fully understood in camelids. However, in recent years some advances have been made in the knowledge of the genes that control the pigmentation in these species. Molecular genetics studies in alpacas have identified mutations in the MC1R and ASIP genes that are associated to coat color (Powell et al., 2008; Feeley and Munyard 2009; Chandramohan et al., 2011; Feeley et al., 2011; Chandramohan et al., 2013) Much more recently, the first work characterizing these genes and their relation to the color in llamas has been published (Daverio et al., 2016).

The goal of this work is to present the progress made so far in llama coat color genetics, and discuss it with regard to the alpaca findings. To understand how color variation occurs, it is necessary to first understand how the eumelanic and phaeomelanic coats are produced. For these, the coding region of MC1R and ASIP were sequenced in animals with different phenotypes: 1) Eumelanic (animals that have black coat with their variants or dark brown coat) 2) pheomelanic (animals with reddish brown coat) 3) mixed (pheomelanic coat with black face and trims) 4) white (non-albino), representing absence of pigmentation in the coat. Moreover, samples of the llama wild ancestor, the guanaco (*Lama guanicoe*), were also sequenced. Screening for genetic variation in 84 llamas showed 13 Single nucleotide polymorphism (SNPs) in the MC1R gene, 10 of which were non-synonymous. Two of them, c.205C > A and c.638G>A, are novel SNPs that had not been previously described in alpacas. The combination of three polymorphisms, c.259G> A, c.376G> A and c.383T> C, defined three main haplotypes (or alleles) for MC1R: MC1R\*1 (c.259A/c.376A/c.383T), MC1R\*2(c.259G/c.376G/c.383C) and MC1R\*3 (c.259G/c.376G/c.383T). The guanaco sequences showed no variation,

and were identical to MC1R3. Allele frequencies were similar in reddish and black animals. However, MC1R\*1 showed a significant association with pigmented coat ( $P < 0.0001$ ). This allele was not found in any of the 29 white llamas analyzed. In this last phenotypic group, MC1R\*2 was found at frequency significantly higher than in the other groups. Moreover, the wild genotype (MC1R\*3/MC1R\*3) was observed in ten individuals, four “black face” and six white animals, suggesting the influence of other genes in the production of those phenotypes.

MC1R mutations responsible for coat color in llamas appear to be different from those reported in alpacas. Feeley and Munyard (2009) found that alpacas with haplotype combination c.82A/c.126T/c.901C (E/E or E/e) in MC1R are able to produce eumelanin whereas animals which have the combination c.82G/c.126C/c.901T (e/e), only express pheomelanin. We found that SNP c.901C > T, c.82A > G and c.126T > C appeared in very low frequency, being almost fixed for the ‘eumelanic’ combination proposed by those authors in the alpaca. Different results were found by Chandramoan et al. (2011) for alpaca: animals homozygous for A82/A259/A376/C901 expressed black phenotypes whereas white animals were homozygous for the combination of 82/G259/G376/T901. In llamas, the c.259A/c.376A/c.383T haplotype is associated to a pigmented coat, but not exclusively eumelanic and c.259G/c.376G/c.383C (MC1R\*2) is found in high frequency in white animals. Moreover, almost half of the white llamas were homozygous MC1R\*2/ MC1R\*2, a genotype not observed in any of the llamas with pigmented coat. In principle, we did not expect any particular genotype at the MC1R in white animals since white is caused in other species by mutations located in other genes, which are dominant and epistatic to MC1R. The c.383T>C substitution in the MC1R\*2 produces a p.M128T change in the protein, in a site highly conserved in mammals. Functional studies in humans confirmed that this variant, found in malignant melanoma, shows marked loss of function and reduced agonist binding affinity. Nevertheless, for the moment we have no direct experimental evidence of the impact of this substitution on the llama protein. As MC1R alleles were not specifically associated with pheomelanic/eumelanic phenotypes in llamas, the complete coding region of ASIP was also analyzed in the same animals. Two polymorphisms were found within exon 4, a 57 bp deletion (c.325\_381del) and a nonsynonymous SNP (c.292C > T). Both polymorphisms are predicted to have a deleterious effect on the protein (Feeley et al., 2011)

All llamas with a pheomelanic coat carried at least one copy of the non-deleted (wild type) ASIP allele. Instead, 17 from 19 black llamas were homozygous for the deletion, homozygous for the T variant of c.292C > T or heterozygous for the combination of both. This is consistent with a recessive inheritance mode for black color proposed for llamas (Frank et al, 2006). The same two variants have been previously associated to black color in alpacas (Feeley et al., 2011; Chandramohan et al., 2013). Moreover, a third substitution c.353G>A associated with this phenotype has been described but has not been yet identified in llamas. Although we have now molecular evidence of how eumelanic and pheomelanic phenotypes can



be produced, some questions remain to be answered. Segregation analysis has shown that the white phenotype is dominant to pigmented phenotypes in llamas and alpacas (Frank et al, 2006). So, why are MC1R\*2/MC1R\*2 llamas white? Why they are not pheomelanic? Why llamas with wild genotype MC1R \*3/MC1R\*3 can also have a white coat? Non albino-white phenotypes are caused in most species by mutations in MITF and KIT genes. On these bases, these genes were afterward sequenced. To this objective, we obtained total RNA from skin biopsies and the complete cDNA sequence was determined in 23 llama samples. Neither polymorphisms in the coding region nor splicing variants associated with coat color were found. Therefore, to further understand how white phenotype in llama is produced studies on the expression of KIT, MITF and some other genes involved in melanogenesis are currently underway within our group.

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Animal fibres from South American camelids and other fibre or wool bearing species provide important products for use by the human population. The contemporary context includes the competition with petrocarbon-based artificial fibres and concern about excessive persistence of these in the natural environment. Animal fibres present highly valuable characteristics for sustainable production and processing as they are both natural and renewable. On the other hand, their use is recognised to depend on availability of appropriate quality and quantity, the production of which is underpinned by a range of sciences and processes which support development to meet market requirements. This collection of papers combines international experience from South and North America, China and Europe. The focus lies on domestic South American camelids (alpacas, llamas) and also includes research on sheep and goats. It considers latest advances in sustainable development under climate change, breeding and genetics, reproduction and pathology, nutrition, meat and fibre production and fibre metrology.

Publication of this book is supported by the Animal Fibre Working Group of the European Federation of Animal Science (EAAP). 'Advances in Fibre Production Science in South American Camelids and other Fibre Animals' addresses issues of importance to scientists and animal breeders, textile processors and manufacturers, specialised governmental policy makers and students studying veterinary, animal and applied biological sciences.



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