

Genética: es el campo de la biología que busca comprender la herencia biológica que se transmite de generación en generación. Genética proviene de la palabra γένος (gen) que en griego significa “**descendencia**”.

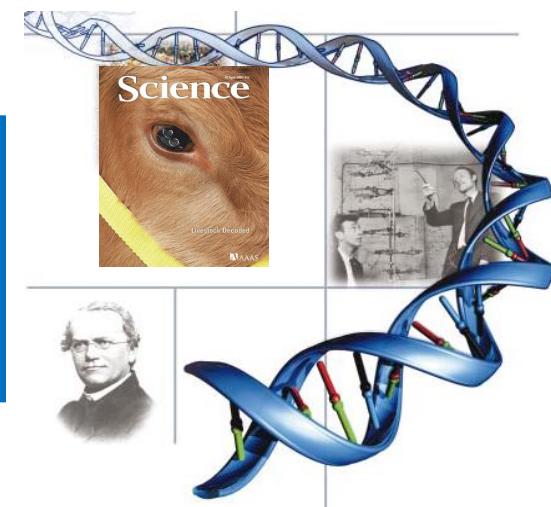
El **objetivo** de estudio de la genética son los **genes**, formados por segmentos de **ADN**.

Genómica: estudio de los genes y sus funciones.

El **objetivo** es entender la estructura del genoma, incluyendo el mapeo de los genes, el secuenciado del ADN y su funcionamiento interactuando con el ambiente.

Por lo tanto, hoy existen niveles diferentes de “resolución” para un mismo **objetivo**: **los genes y su funcionamiento**

...“*El simposio tiene el objetivo general de mostrar, en distintas especies, cómo nuevas tecnologías nos hacen cambiar la mirada sobre el mejoramiento animal*”...





ALAG
MENDOZA,
ARGENTINA 2019

La arquitectura
del genoma:
su expresión en
los fenotipos
y las poblaciones

Un cambio de la mirada para la cría animal aprovechando la información genómica

- .- Mejoramiento genético de ovinos en la Patagonia: actualidad y desafíos ante nuevos escenarios climáticos y comerciales . Vozzi, P A
- .- El cerdo criollo pampa Rocha de Uruguay como recurso zoogenético local (una mirada desde la genética). Llambí M.S
- .- Avances en el conocimiento del genoma de la alpaca. Gutiérrez Reynoso, G y Abel Ponce de Leon
- .- Clonación equina, herramienta práctica para preservar genética. Kaiser, G y col.
- .- Estrategias de investigación multidisciplinarias usando información genómica para la elección de animales superiores. Poli, M A y col.



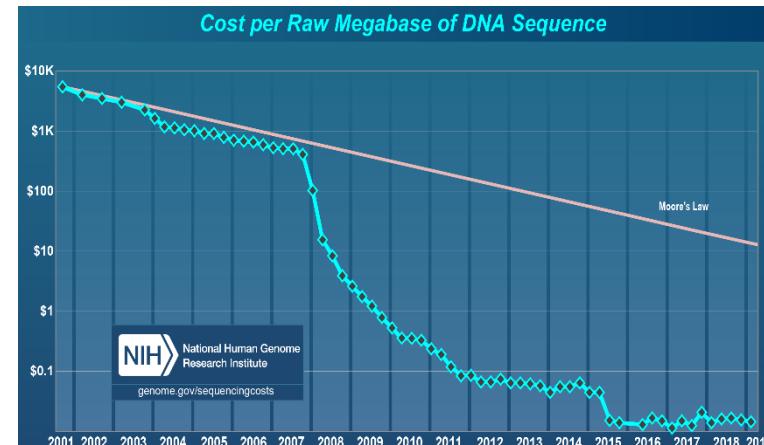
Estrategias de investigación multidisciplinarias usando información genómica para la elección de animales superiores.

Poli, M A



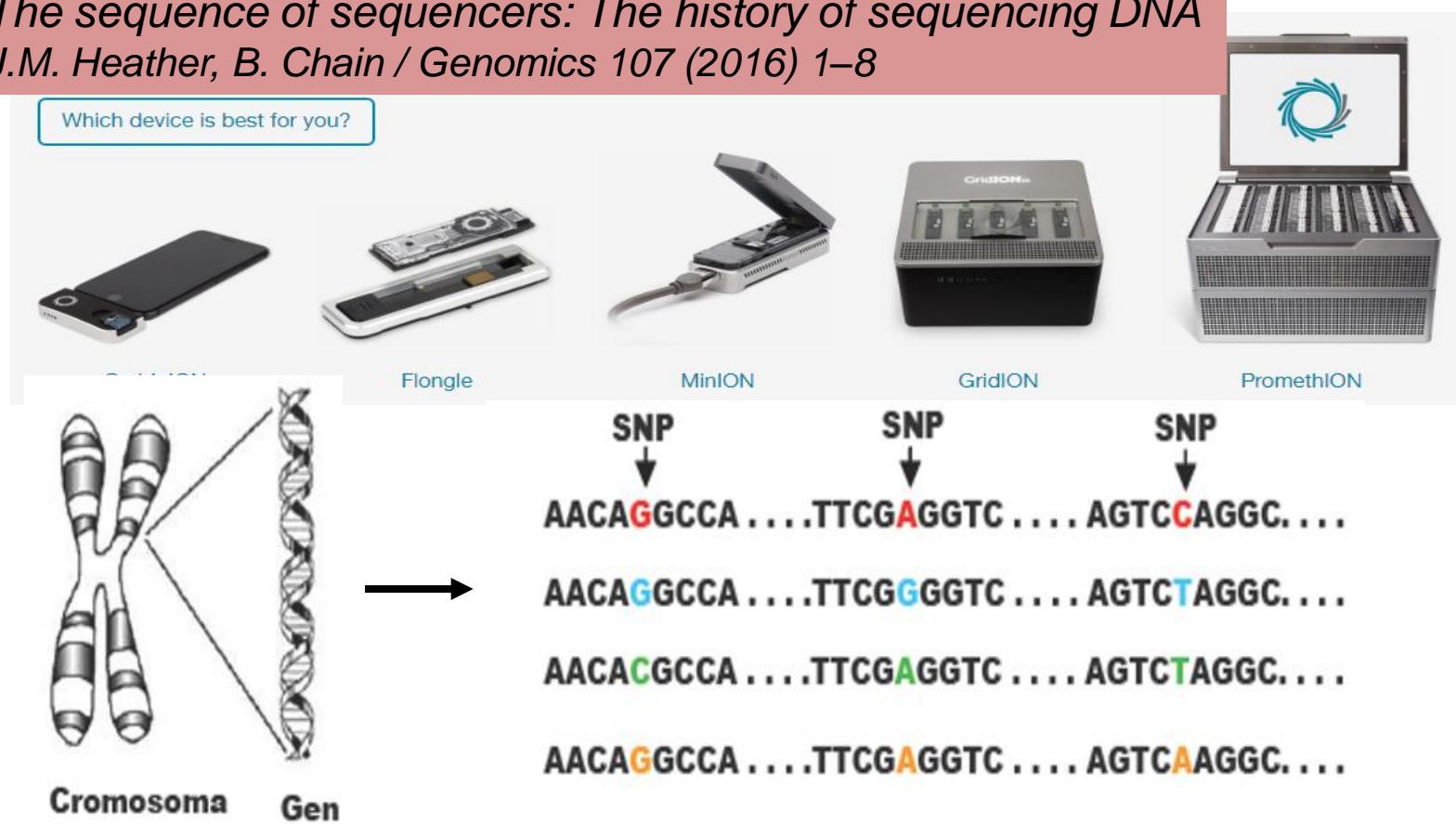
NGS platforms

- Roche/454 FLX: 2004
- Illumina Solexa Genome Analyzer: 2006
- Applied Biosystems SOLiD™ System: 2007
- Helicos Heliscope™ : 2008
- Pacific Biosciences SMRT: 2010



The sequence of sequencers: The history of sequencing DNA
J.M. Heather, B. Chain / Genomics 107 (2016) 1–8

Which device is best for you?

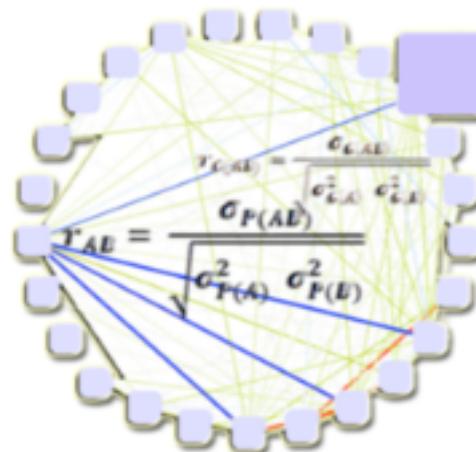


CorrDB Animal Trait Correlation Database

Genetic and phenotypic trait correlation studies have been conducted for 70+ years since Hazel proposed the methods. A genetic correlation is the proportion of shared variance between two traits that is due to genetic causes; a phenotypic correlation is the degree to which two traits co-vary among individuals in a population. In the genomics era, while gene expression, genetic association, and network analysis provide unprecedented means to decode the genetic basis of complex phenotypes, it is important to recognize the possible effects genetic progress in one trait can have on other traits. This database is designed to collect all published livestock genetic/phenotypic trait correlation data, aimed at facilitating genetic network analysis or systems biology studies.

As of the current Release, a total of **18,049** correlations data on **629** traits, and **3,083** heritability data on **912** traits in **5** livestock animal species have been curated into the CorrDB. [Note of Apr 29, 2018] In order to maintain a high standard for data integrity and for better quality controls, a total of **3,628** historic correlation data were taken offline and are currently being re-entered into the database with data integrity check mechanisms newly built into the curator tools.

- Phenotypic correlations
- Genetic correlations
- Environmental correlations
- Residual correlations
- Trait heritabilities
- Navigate by browsing the traits



Data summary (as of Aug 22, 2019)				
Species	Correlations		Heritability	
	Count	Traits	Count	Traits
Cattle	10,000	290	1,300	332
Chicken	1,221	72	263	95
Horse	209	27	118	53
Pig	5,469	194	1,310	363
Sheep	1,150	46	92	69
sum	18,049	629	3,083	912

Last month visitor activities

- Web visits: 2,687 (hits)
- Downloads: 97.641(MB)



New Data: If you found correlation data that has not been included in the current release, please either (1) register for a curator account to directly input them (see "Database Curators / Editors" below), or (2) inform our Bioinformatics Team so that we can include your data for curation.



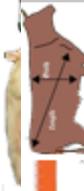
Vertebrate Trait (VT) Ontology

Enhancing the ability to standardize phenotype nomenclature



Livestock Product Trait (LPT) Ontology

Enhancing the ability to standardize phenotype nomenclature



Clinical Measurement Ontology (CMO)

Enhancing the ability to standardize phenotype nomenclature



The Clinical Measurement Ontology (CMO) is designed to be used to standardize morphological and physiological measurement records generated from clinical and model organism research and health programs.



Consortium:



The CMO was developed at the Rat Genome Database (rgd.mcw.edu) to encompass qualitative or quantitative values that result from the assessment of a morphological or physiological state or property of single or groups of individuals/samples. Terms relevant to livestock were contributed by Iowa State University incorporating terms from the Animal Trait Ontology (ATO). This project is aimed at enhancing the ability to standardize phenotype nomenclature across species. This will undoubtedly help the animal genome community by facilitating the transfer of genomics information from some well-studied species.

- Department of Animal Science, Iowa State University, Ames, IA, USA
- Mouse Genome Informatics, The Jackson Laboratory, Bar Harbor, ME, USA
- Human and Molecular Genetics Center, Medical College of Wisconsin, Milwaukee, WI, USA
- Department of Physiology, Medical College of Wisconsin, Milwaukee, WI, USA
- Department of Surgery, Medical College of Wisconsin, Milwaukee, WI, USA



Access the data:

(Version: 2.47 with 3,000 terms, updated on Jan 07, 2019)

[Data archive and details](#) (hosted by BioPortal@NCBO)

[CMO browser](#) (at MCW, home of the CMO project)

[Browse the glossary](#) (hosted at AnimalGenome.ORG)

[Tree view on AmiGO](#) (hosted at AnimalGenome.ORG)

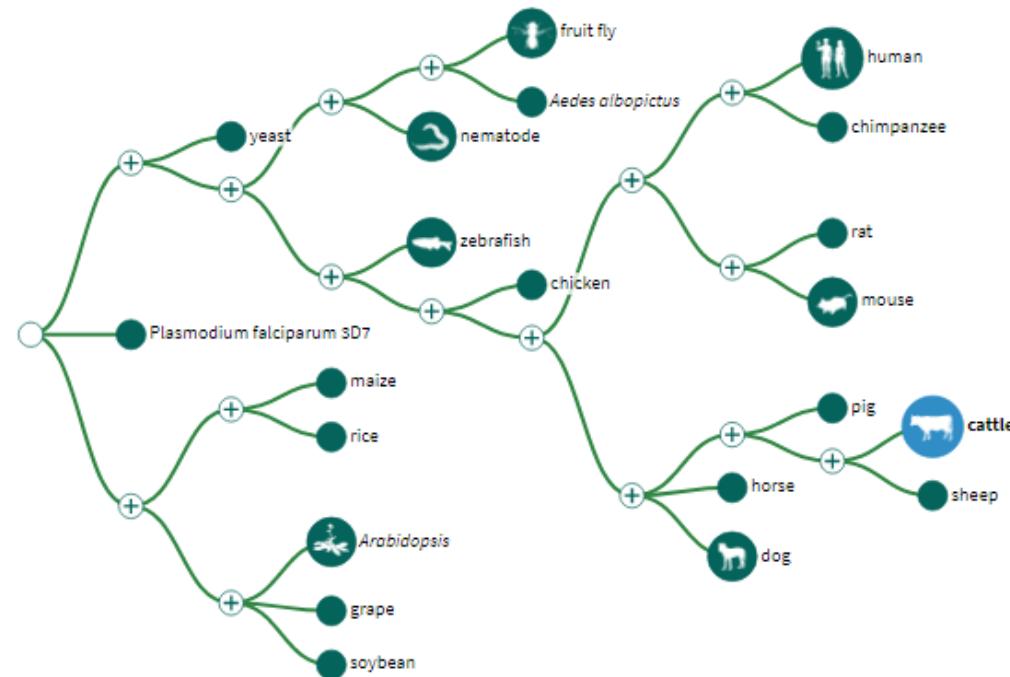
[VCmap tool](#)

Genome Data Viewer

GDV is a genome browser supporting the exploration and analysis of more than 740 eukaryotic RefSeq genome assemblies. ⓘ

Select organism

Bos taurus (cattle)



Bos taurus (cattle) genome



Search in genome

Location, gene or phenotype



Examples: RHO, chr22:56224000-56233000, DNA repair

Assembly

ARS-UCD1.2

[Browse genome](#)

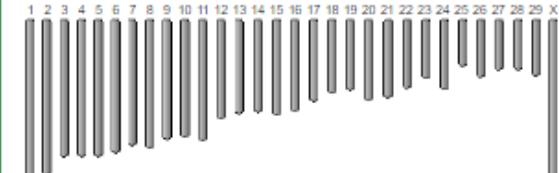
[BLAST genome](#)

Assembly details

Name	ARS-UCD1.2
RefSeq accession	GCF_002263795.1
GenBank accession	GCA_002263795.2
Download via FTP	RefSeq , GenBank
Submitter	USDA ARS
Level	Chromosome
Category	Representative genome

Annotation details

Annotation Release 106
Release date 2018-05-11



genecards.org/Search/Symbol?queryString=SDE2

Su GeneCardsSuite GeneCards MalaCards LifeMap Discovery PathCards TGex VarElect GeneAnalytics GeneALaCart GenesLikeMe

Free for academic non-profit institutions. Other users need a [Commercial license](#) WEIZMANN INSTITUTE OF SCIENCE

GeneCards® HUMAN GENE DATABASE

Symbols ▾ SDE2 Advanced

Home User Guide Analysis Tools ▾ News And Views About ▾ My Genes Log In / Sign Up

Showing 1 of 1 Results for SDE2 Search Time: 2 ms

Export ▾ Show: 25 ▾

	Symbol	Description	Category	GIFs	GC id	Score ▾
1	SDE2	SDE2 Telomere Maintenance Homolog	Protein Coding	34	GC01M225982	16.06

section	Paralogs	Pathways	Products	Proteins	Publications	Sources	Summaries	Transcripts	Variants
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External links for SDE2 Gene

HGNC: 26643 Entrez Gene: 163859 Ensembl: ENSG00000143751 UniProtKB: Q6IQ49

Cattle

Previous HGNC Symbols for SDE2 Gene

C1orf55

Previous GeneCards Identifiers for SDE2 Gene

GC01M226171

Search aliases for SDE2 gene in PubMed and other databases

Summaries for SDE2 Gene



GeneCards Summary for SDE2 Gene

SDE2 (SDE2 Telomere Maintenance Homolog) is a Protein Coding gene. An important paralog of this gene is SF3A3.

Pig

UniProtKB/Swiss-Prot for SDE2 Gene

Involved in both DNA replication and cell cycle control (PubMed:27906959). Unprocessed SDE2 interacts with PCNA via its PIP-box. The interaction with PCNA prevents monoubiquitination of the latter thereby inhibiting translesion DNA synthesis. The binding of SDE2 to PCNA also leads to processing of SDE2 by an unidentified deubiquitinating enzyme, cleaving off the N-terminal ubiquitin-like domain. The resulting mature SDE2 is degraded by the DCX(DTL) complex in a cell cycle- and DNA damage dependent manner (PubMed:27906959). Binding of SDE2 to PCNA is necessary to counteract damage due to ultraviolet light induced replication stress. The complete degradation of SDE2 is necessary to allow S-phase progression (PubMed:27906959). [SDE2_HUMAN,Q6IQ49](#)

Additional gene information for SDE2 Gene

HGNC(26643) Entrez Gene(163859) Ensembl(ENSG00000143751) UniProtKB(Q6IQ49)

Search for SDE2 at DataMed

Search for SDE2 at HumanCyc

No data available for Entrez Gene Summary , CIViC summary , Tocris Summary , Gene Wiki entry , PharmGKB "VIP" Summary , fRNAdb sequence ontologies and piRNA Summary for SDE2 Gene

1000 Bull Genomes Project to Map Simple and Complex Genetic Traits in Cattle: Applications and Outcomes

Annual Review of Animal Biosciences

Vol. 7:89-102 (Volume publication date February 2019)

First published as a Review in Advance on December 3, 2018

<https://doi.org/10.1146/annurev-animal-020518-115024>

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²Agriculture Victoria Research, Avondale, Bendigo, Victoria 3550, Australia

Figures Tables



[Download All Figures as Powerpoint](#)

[<< Previous Article](#)

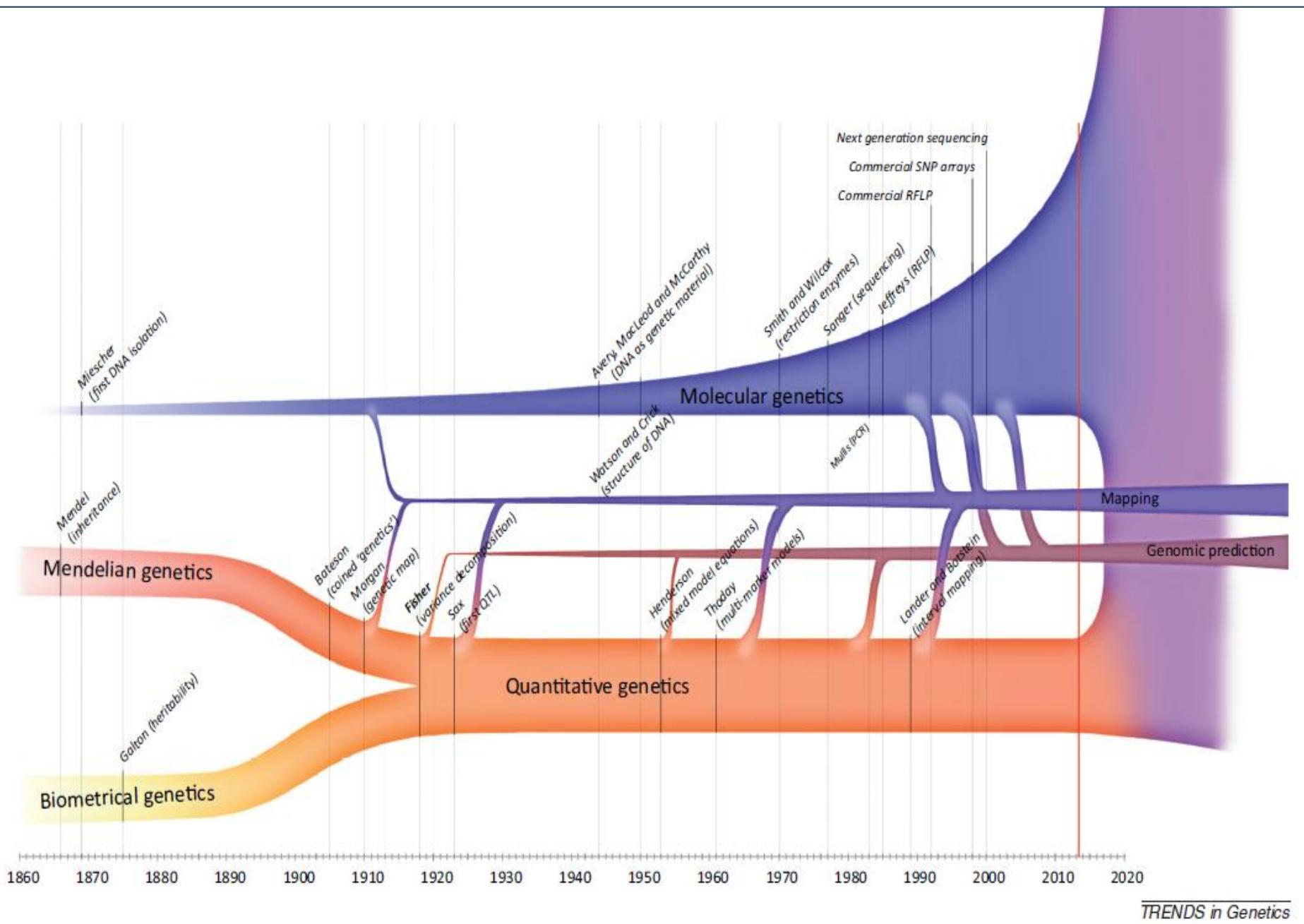
[Next Article >>](#)



Coordinated international action to accelerate genome-to-phenome with FAANG, the Functional Annotation of Animal Genomes project

Andersson *et al.* *Genome Biology* (2015) 16:57
DOI 10.1186/s13059-015-0622-4





Sheep more resistant to the Parasite Gastrointestinal Infestation



EEA Mercedes y EEA Concepción del Uruguay - **Corriedale** - Dec. 2009 - cont.



EEA Balcarce – **Texel** - Dec. 2018 - cont.



EEA Anguil - **Pampinta** - 2006-2009 ; 2010-2013



Artificially challenge L3 lambs 3-6 months old

.- Day 0, 28, 35 y 42

- 1.- Body weight
- 2.- FEC
- 3.- Hematocrit
- 4.- FAMACHA

Others:

- 5.- Birth body weight and every 45 days until one year old
- 6.- Ewe body weight/lamb body weight to weaning
- 7.- Fleece weight
- 8.- Wool quality (OFDA)

Diámetro medio (mic)	Desviación st (mic)	Coeficiente var (%)	Curvatura de ondulación (grados/mm)	Factor de confort (%)	Rinde al lavado (%)	Largo de mecha (mm)	Res.Trac. (N/ktex)	Punta (%)	Medio (%)	Base (%)
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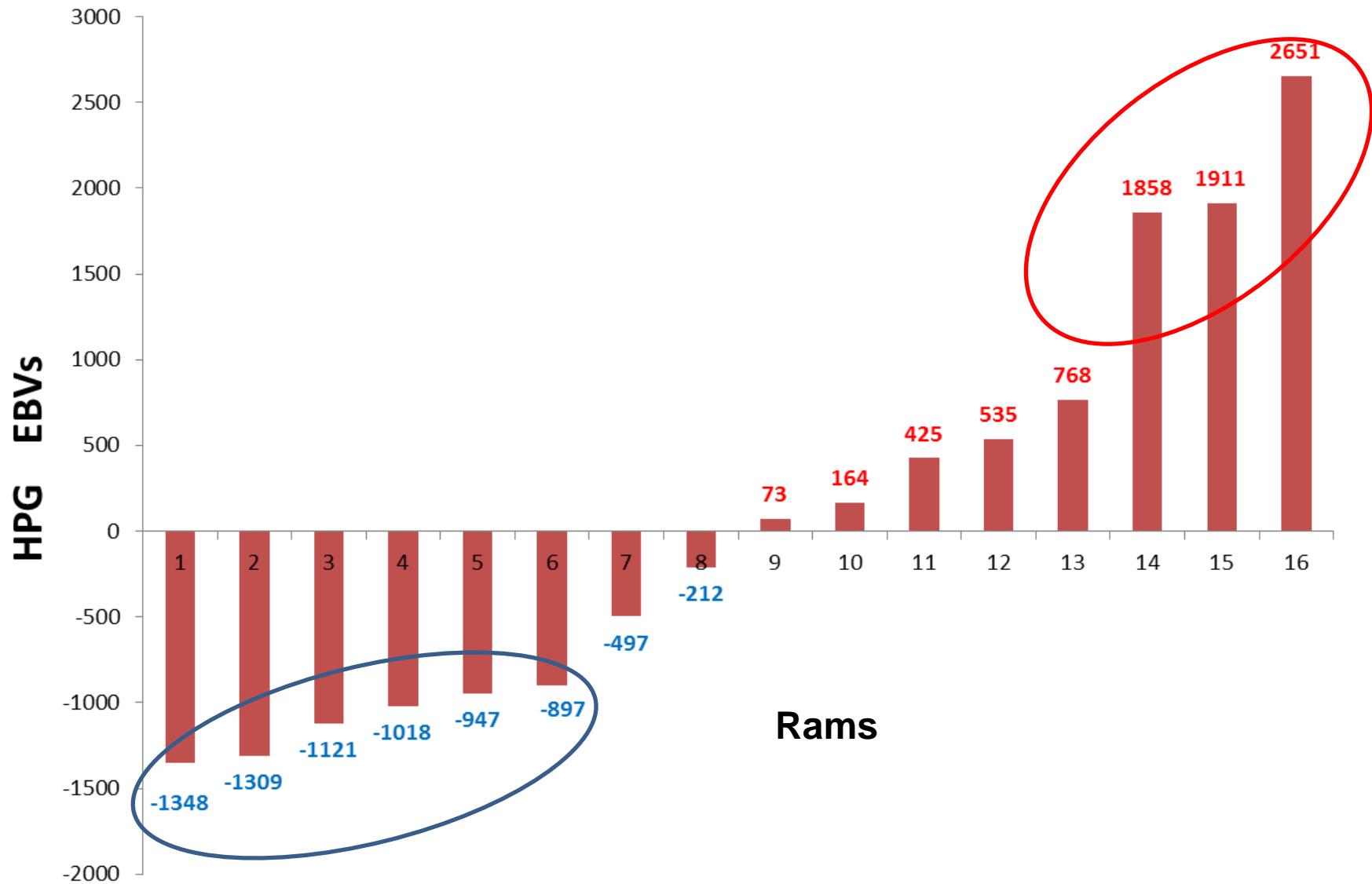
- 9.- Reproductive traits
- 10.- IgG; IgE . . .
- 11.- Ewe FEC

Heredabilities (h^2) and genetic and phenotypic correlations.

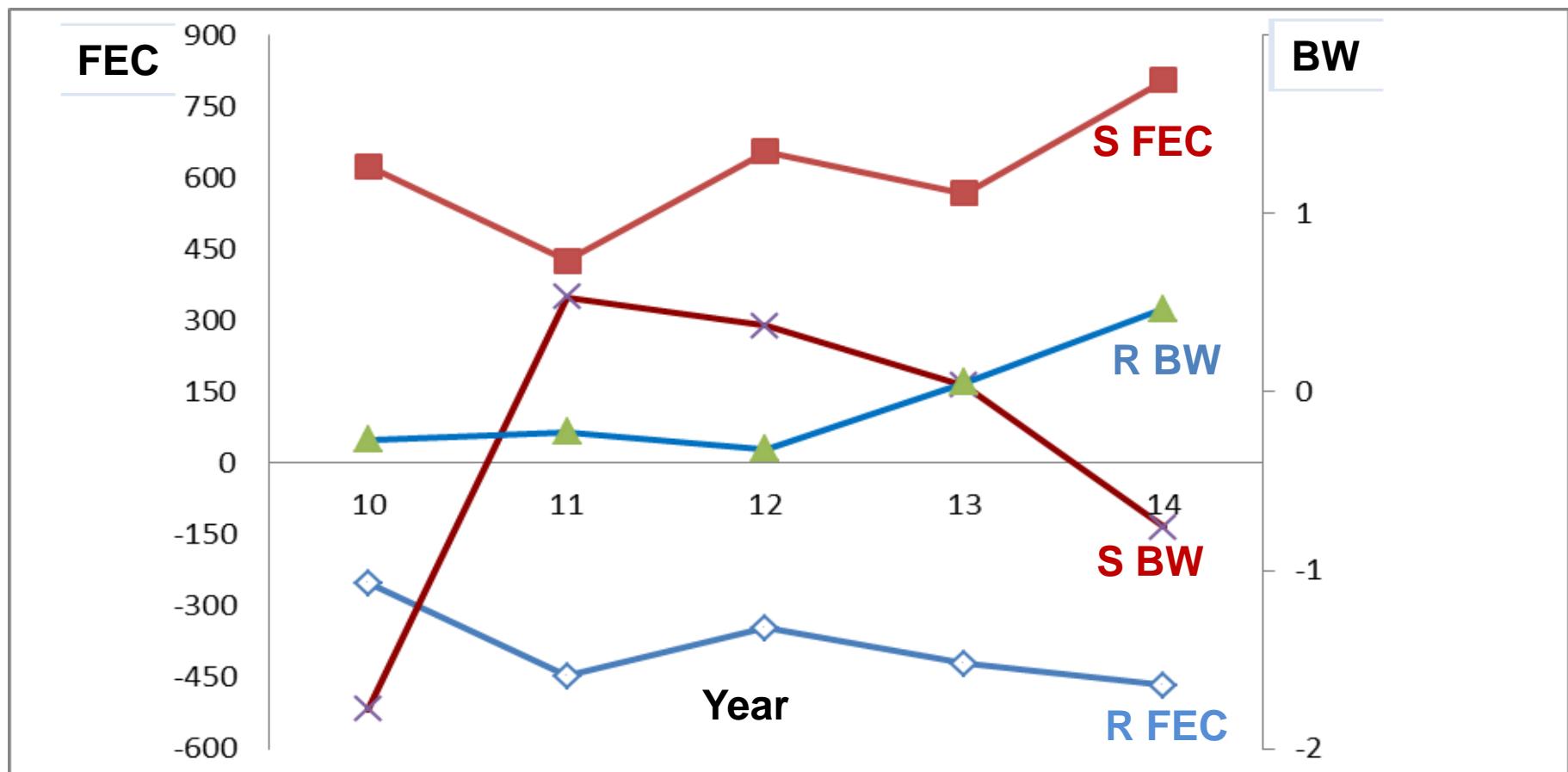
Trait	BW	FEC	Hematocrit	FAMACHA [©]
BW	0.41 (0.11)	0.09 (0.25)	0.00 (0.19)	-0.43 (0.16)
FEC	-0.16 (0.05)	0.20 (0.08)	-0.38 (0.20)	0.50 (0.19)
Hematocrit	0.24 (0.04)	-0.40 (0.03)	0.42 (0.08)	-0.41 (0.14)
FAMACHA [©]	-0.26 (0.04)	0.22 (0.03)	-0.36 (0.03)	0.29 (0.06)

^a Heritabilities on the diagonal, genetic correlations above the diagonal and phenotypic correlations below the diagonal; between parentheses their standard errors.

Estimated Breeding Values (acc > 0.80)



BW and FEC by year



A .- Quantitative genetics classical approaches (variance components; h^2 ; genetics and phenotypic correlations, EBVs, etc.)

B .- Molecular tools

Genome Wide Association Study (GWAS)

- Genotyping field samples with 50K SNP chip to assess whole genome variation
- Detection of Selection Signature within sheep & goat genome for parasite resistance
- Whole genome association with phenotype

Candidate Gene Study

- Targeted re-sequencing of candidate genes involved in immune pathways, pathogen recognition receptors, adaptive immune response, etc.
- Detection of functional allelic variants
- Association of allelic variants with phenotype

OPEN  ACCESS Freely available online



Candidate Gene Approach for Parasite Resistance in Sheep – Variation in Immune Pathway Genes and Association with Fecal Egg Count

Kathiravan Periasamy^{1*}, Rudolf Pichler¹, Mario Poli², Silvina Cristel³, Bibiana Cetrá⁴, Daniel Medus⁵, Muladno Basar⁶, Thiruvenkadan A. K.⁷, Saravanan Ramasamy⁷, Masroor Babbar Ellahi⁸, Faruque Mohammed⁹, Atanaska Teneva¹⁰, Mohammed Shamsuddin¹, Mario Garcia Podesta¹, Adama Diallo¹

Candidate gene – Preliminary Results (*transformed FEC*)

Chr	SNP	Benjamini-Hochberg - value	Bonferroni p-value	gene
2	IFIH1_400_AT	0,03827		Interferon induced with helicase C domain 1
3	CLEC8A_532_CT	0,005027	0,01005	C-type lectin domain family 8, member A (LOX1)
3	CLEC12A_567_CT	0,01219	0,03823	C-type lectin domain family 12, member A (MICL)
3	CLEC12A_440_CT	0,01219	0,04876	C-type lectin domain family 12, member A (MICL)
3	ZBTB39_51_GA	0,03581		Zinc finger and BTB domain containing 39
3	NLRC4_918_AG	0,03887		NLR family, CARD domain containing 4
3	IL2RB_180_TC	0,04805		Interleukin 2 receptor, beta
3	CLEC1B_777_GC	0,04841		C-type lectin domain family 1, member B
11	MAP2K3_832_GC	0,03827		Mitogen Activated Protein Kinase 3
11	MAP2K3_945_CT	0,03827		Mitogen Activated Protein Kinase 3
11	MAP2K3_123_AG	0,03887		Mitogen Activated Protein Kinase 3
12	MASP2_104_CT	0,02853		Mannan binding lectin serine peptidase 2
12	MASP2_573_CT	0,03887		Mannan binding lectin serine peptidase 2
12	MASP2_194_CT	0,03887		Mannan binding lectin serine peptidase 2
15	NLRX1B_1088_AG	0,02853		NLR family member X1
20	OLADRA1_479_CT	3,20E-05	3,20E-05	MHC-Ovine Lymphocyte Antigen-DRA
24	CIITA_606_AG	0,02853		class II, major histocompatibility complex, transactivator
24	NLRC3_95_AG	0,03208		NLR family, CARD domain containing 3
24	CIITA_813_GT	0,04841		class II, major histocompatibility complex, transactivator
25	SFTPA1_964_AT	0,0333		Collectin-SP-A
25	SFTPД_596_AG	0,03887		Collectin-SP-D
25	SFTPД_331_AG	0,03887		Collectin-SP-D
25	SFTPД_505_CT	0,0422		Collectin-SP-D
25	SFTPД_709_CT	0,04661		Collectin-SP-D

Productive and Health phenotypic data - Cattle

Data Base : *Las Taperitas S.A.* .-

Breeds = 73% Holstein ; 23% Holstein x Jersey

900,000 records (Milk, P, F)

25,542 milk cows. 08/14/1992 to 07/05/ 2012

.- Leukemia n= 1000

.- 2 times - 6 months ELISA p24 + WBC

.- Real Time PCR for proviral low

.- PCR specific

.- Mastitis : SCC in 2,000 cows during 2 years

.- Internal parasite : FEC for the 1,864 cows

.- Genotype : (a) 1600 animals 39 SNPs Candidate gene SNplex®

(b) 1050 animals Bovine 50K Chip 

(c) 900 cows genotyped for BoLA DRB3.2 gene (Seq)



**Genome-wide association study for somatic cell score
in Argentinean dairy cattle**

Livestock Science 175 (2015) 1–9

Juan P Nani^a, Maria A Raschia^b, Mario A Poli^b, Luis F Calvinho^a,
Ariel F Amadio^{a,c*}

Table 6

Effect sizes (SD) per genotype class and MAF for the SNP significantly associated with the different variables calculated from SCS.

SNP	BTA	MA (F)	Variable ^a	B/B	B/A	A/A
ARS-BFGL-NGS-114608	1	B(0.20)	AM	0.65 (0.42)	0	-0.11 (0.08)
Genotype frequency				0.05	0.31	0.65
Hapmap60306-rs29023088	5	A(0.14)	AM	0.22 (0.20)	0	-0.27 (0.21)
Genotype frequency				0.01	0.26	0.73
ARS-BFGL-NGS-107504	10	P(0.07)	TOP2	0.70 (0.61)	0	0.52 (0.16)
Genotype frequency						
ARS-BFGL-NGS-109705	26	A(0.45)	MAX	1.52 (0.42)	0	-0.52 (0.27)
Genotype frequency				0	0.13	0.87
ARS-BFGL-NGS-109705			TOP3	-0.04 (0.25)	0	0.56 (0.23)
Genotype frequency			MAX	-0.17 (0.05)	0	0.53 (0.49)
Genotype frequency				0.29	0.52	0.18

As expected, no very large QTL effects were detected, showing that the QTLs affecting these variables are dispersed throughout the genome, each with a relatively small effect.

^a Variables calculated from the SCS per lactation: the arithmetic mean (AM), the maximum value (MAX) and the arithmetic mean of the top 3 values (TOP3).



Single nucleotide polymorphisms in candidate genes associated with milk yield in Argentinean Holstein and Holstein x Jersey cows

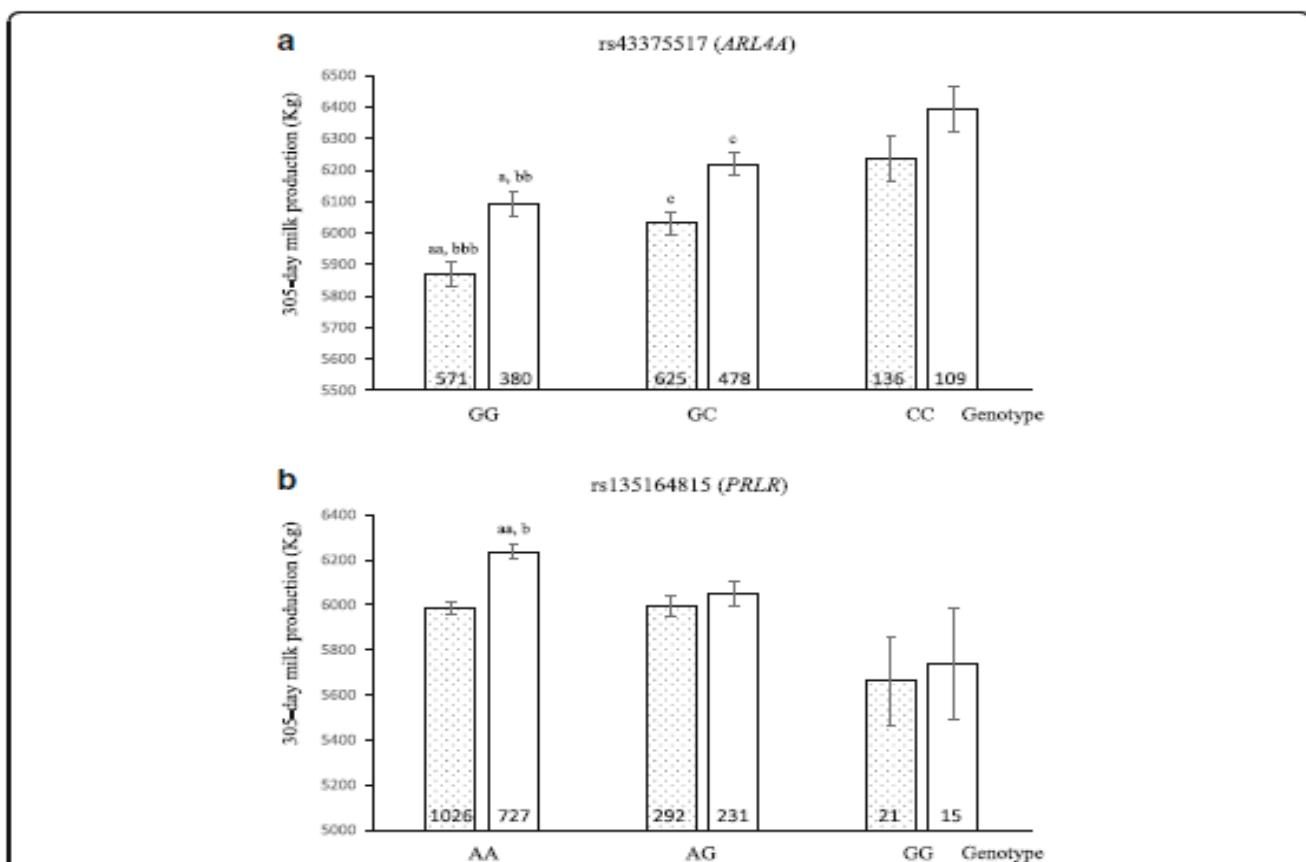
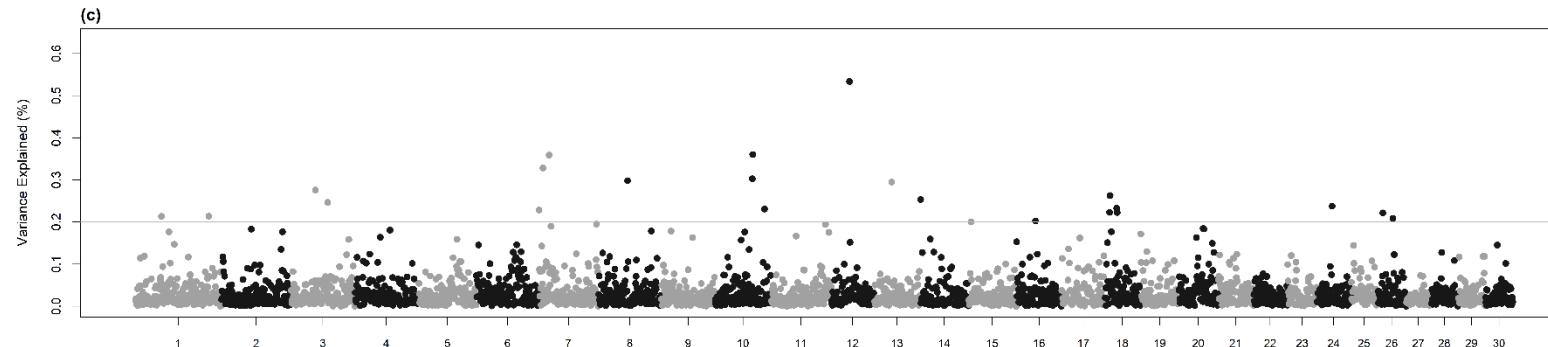
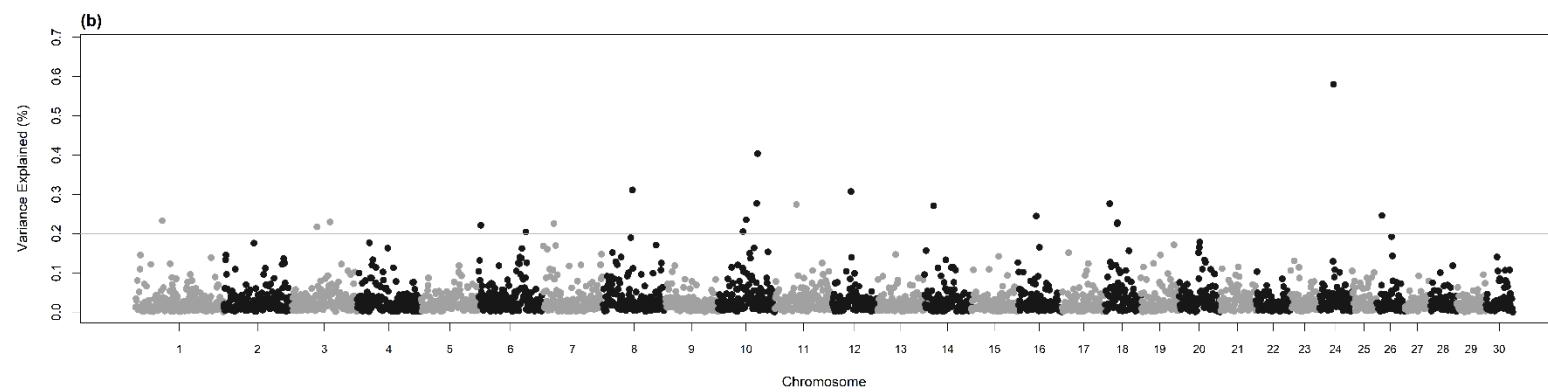
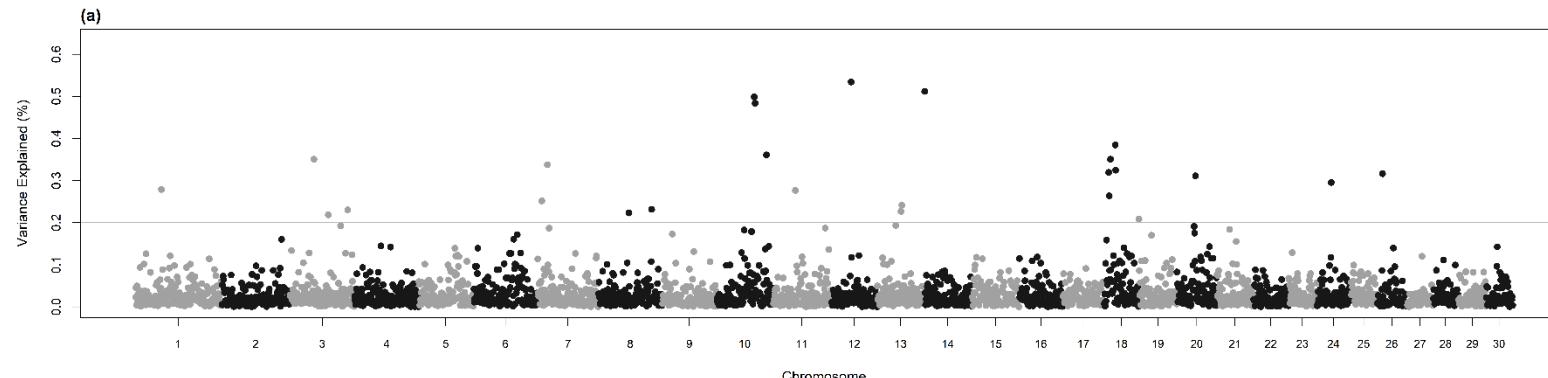


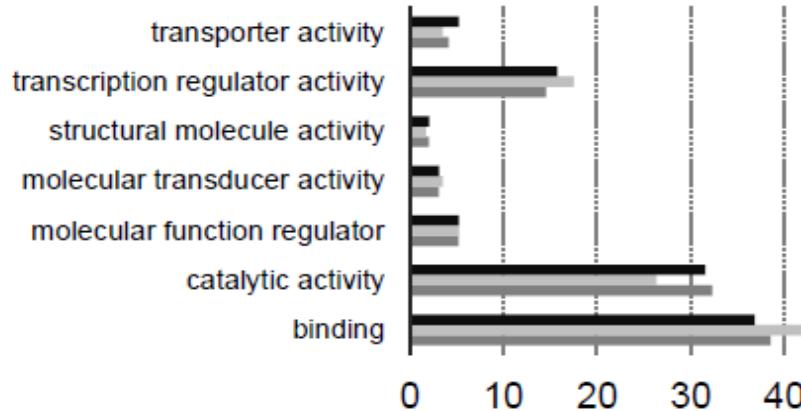
Fig. 1 Effect of different SNP genotypes on milk production. Genotypes for SNPs from *ARL4A* (a) and *PRLR* (b) genes are shown in the x-axis and 305-day cumulative milk production (Kg) in the y-axis. Dot bars and white bars denote the whole population and the Holstein subpopulation, respectively. The number of animals presenting each genotype is indicated inside each bar. Homozygote GG for rs43375517 or AA for rs135164815 vs heterozygote, ^a**p* < 0.005; ^b**p* < 0.05. Homozygote 1 vs homozygote 2, ^{bbb}**p* < 0.0005; ^{bb}**p* < 0.005; ^b**p* < 0.05. Heterozygote vs homozygote CC for rs43375517 or GG for rs135164815, ^c**p* < 0.005

Genome-wide association study for milk production and milk composition traits in Holstein and Holstein x Jersey dairy cattle.
Raschia, M et. al., (enviado)



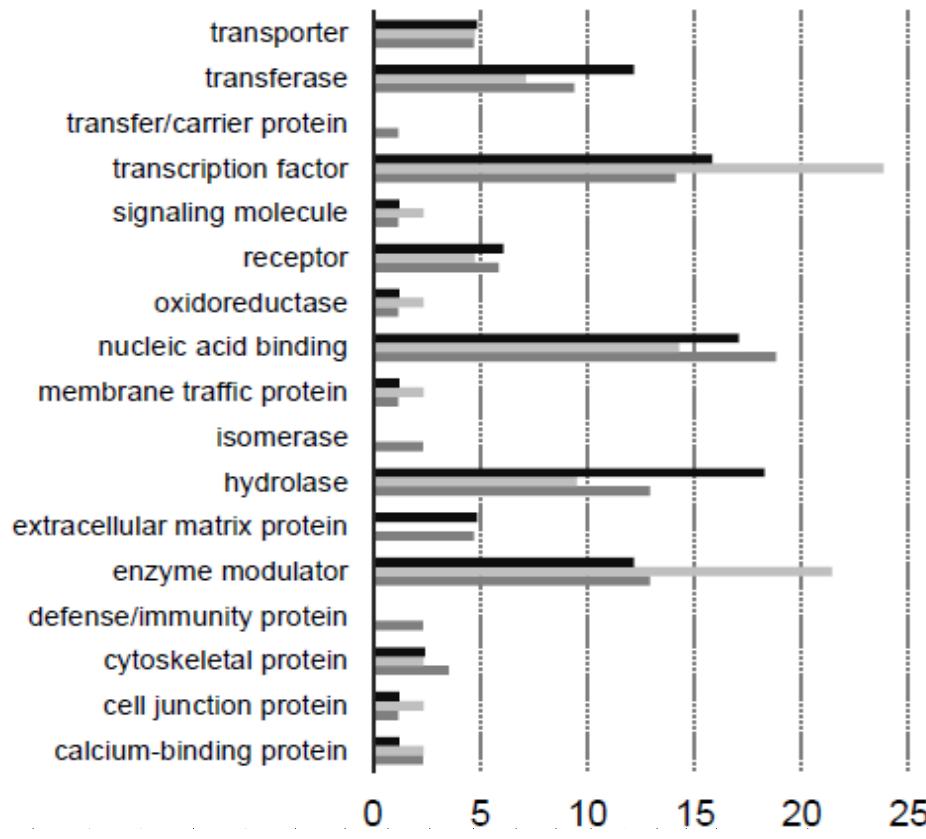
(a)

Functional category



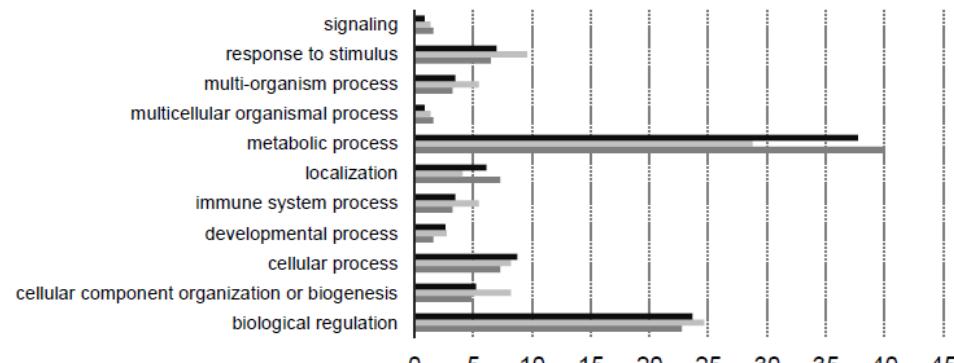
(b)

Protein class



(c)

Biological process





Genome-wide scan for commons SNPs affecting bovine leukemia virus infection level in dairy cattle

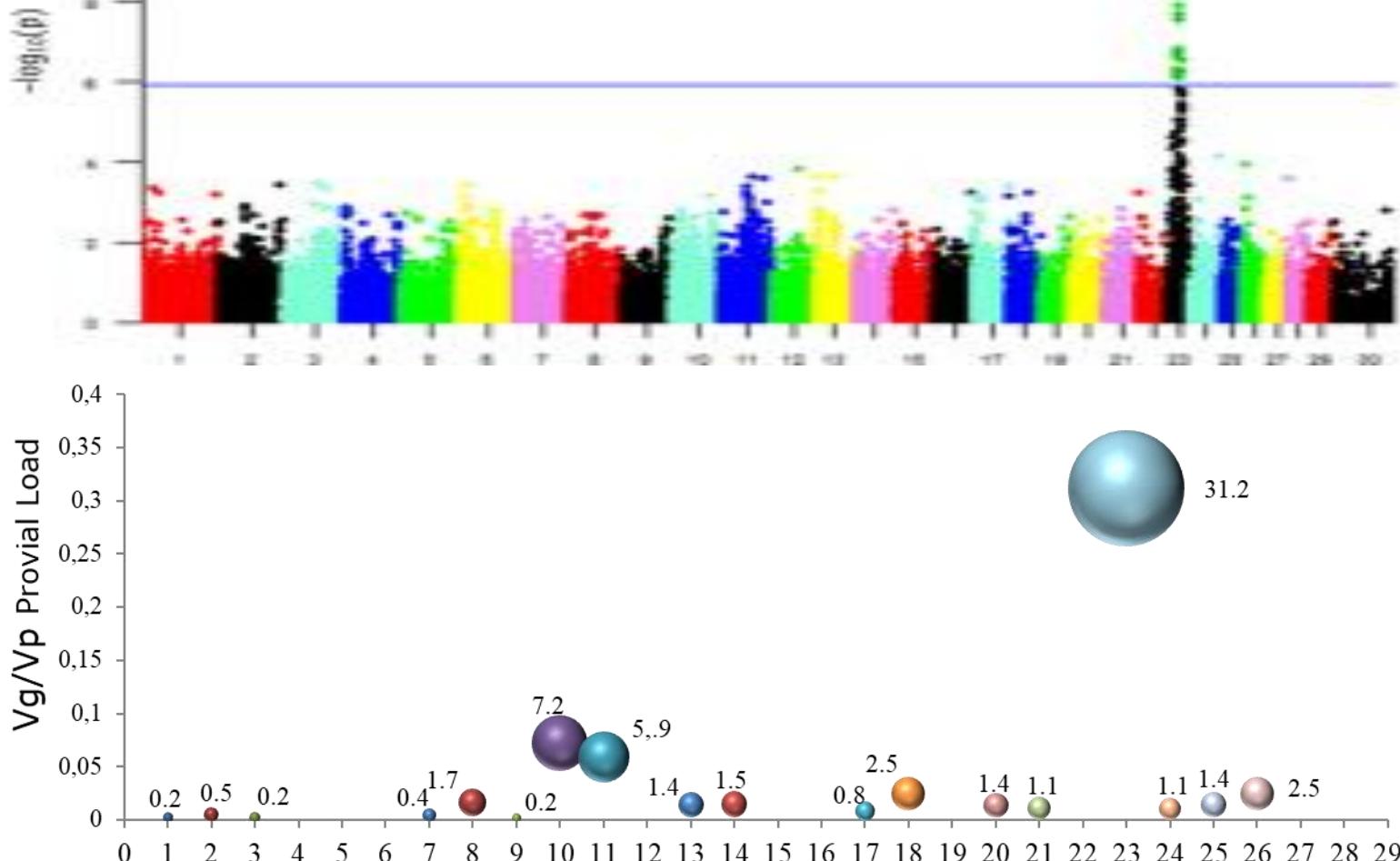
Carignano *et al.* *BMC Genomics* (2018) 19:142
<https://doi.org/10.1186/s12864-018-4523-2>

GWAS in Case-control study

The case-control phenotypes to conduct GWAS for LI in vivo considered **396 cases (HPVL)** and **373 controls (ND/LPVL)**

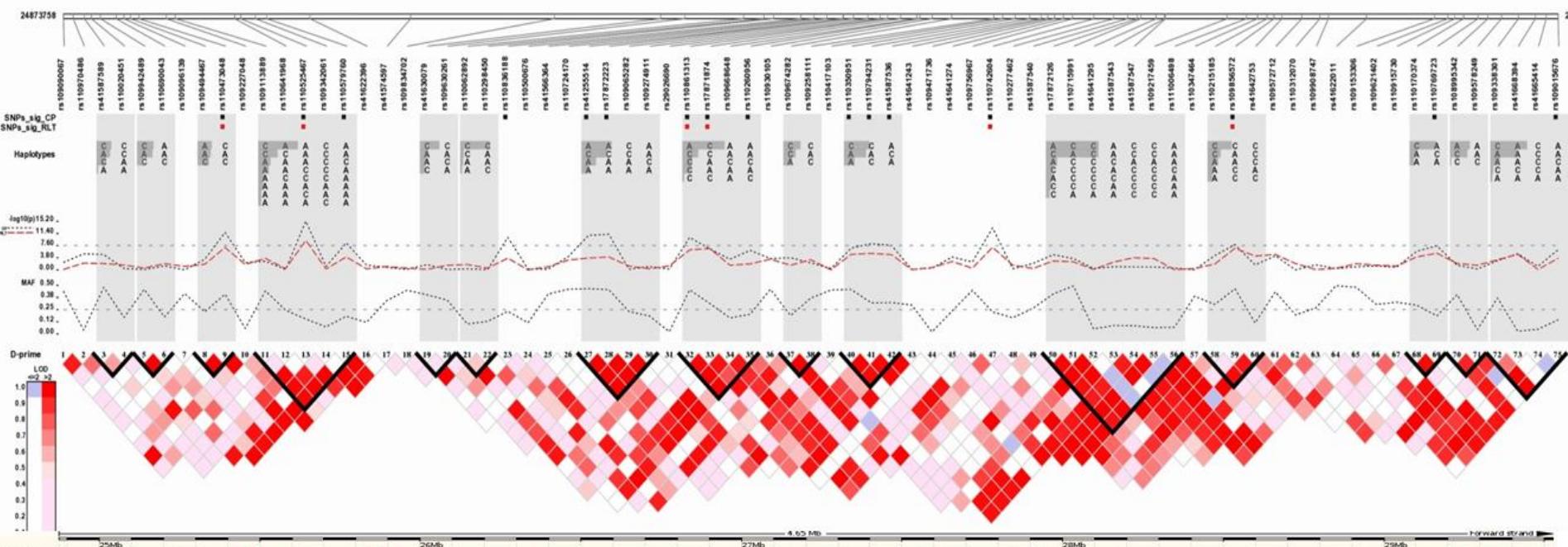
The GWAS were performed using LMMs as implemented in GCTA v1.24. Gene to SNPs mapping was performed using BedTools package. Gene functional classification based in Gene Ontology terms (GO) was performed using the PANTHER classification system website (<http://www.pantherdb.org/>).

Manhattan plot depicting GWAS results of BLV level of infection



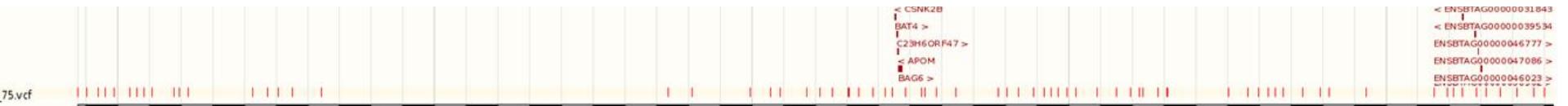
Phenotypic variance captured by the common SNPs in the genotyping panel. The heritability (h^2) for PVL was estimated at 0.63 ($SE \pm 0.14$) on the scale of liability.

Gene context and the LD map in the BTA23. SNPs associated with infection level.



25 hits ($p < 1.13 \cdot 10^{-6}$) in the MHC region with genes relate to immune esponse All SNPs exceeding the significance threshold after Bonferroni's correction ($-\log_{10}p > 5.94$) are located on BTA 23.

- transcriptomic from high and low proviral load animals
- Proviral integration places (targeted -sequencing)
- re-sequencing MHC regions in LLI and HLI



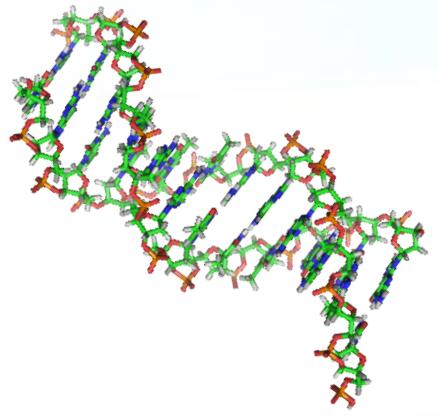
Harnessing genomic information for livestock improvement

[Nature Reviews Genetics](#)
volume 20, pages 135–156 (2019)

Michel Georges^{1,2*}, Carole Charlier^{1,2} and Ben Hayes^{1,3}

Final remarks . . .

- The livestock genomes sequencing is running.
 - The quantitative-molecular model works towards integrative and predictive biology approaches that improve the ability to move from genomic sequence to trait consequence.
 - The gene edition is an exiting technology.
-
- Nevertheless, up today in livestock, it is necessary more/new phenotypes . . .



Thank you



