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Issues and perspectives of genomic selection in limited size breeds: the case of Italian Simmental

NPP Macciotta, L Degano, D Vicario,

C. Dimauro, A. Cesarani,

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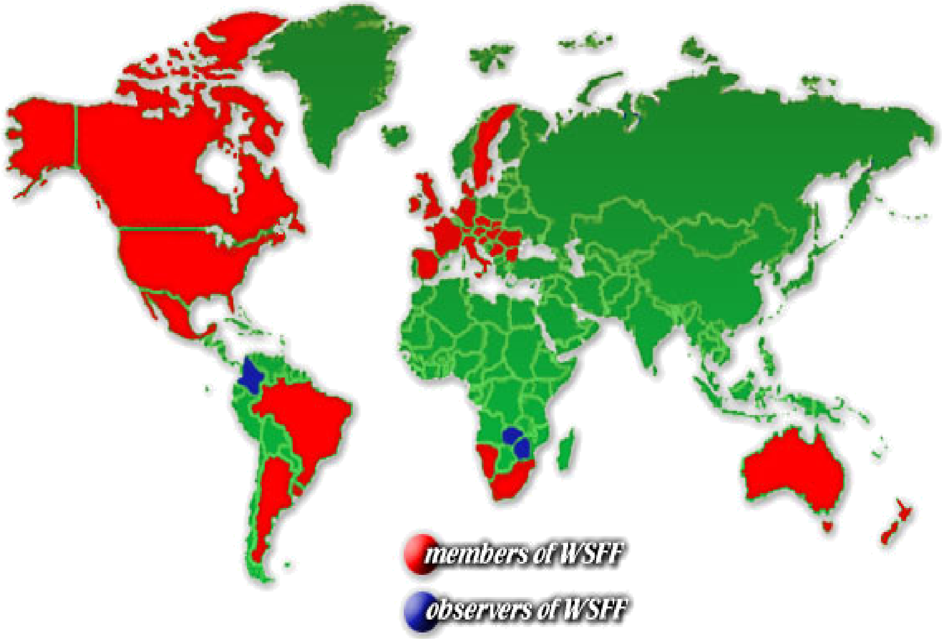
Libro Genealogico
Pezzata
Rossa
Italiana
A.N.A.P.R.I. - Udine

Ente selezionatore
A.N.A.P.R.I.
Associazione Nazionale
Allevatori Pezzata
Rossa Italiana

Simmental: population that originates from the Simme valley (Switzerland)

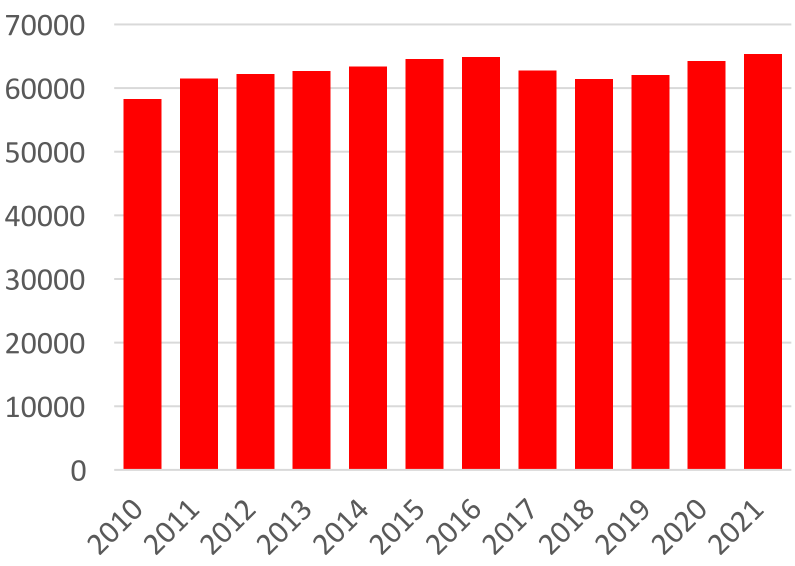


About 41 M animals, the second largest cattle breed in the world

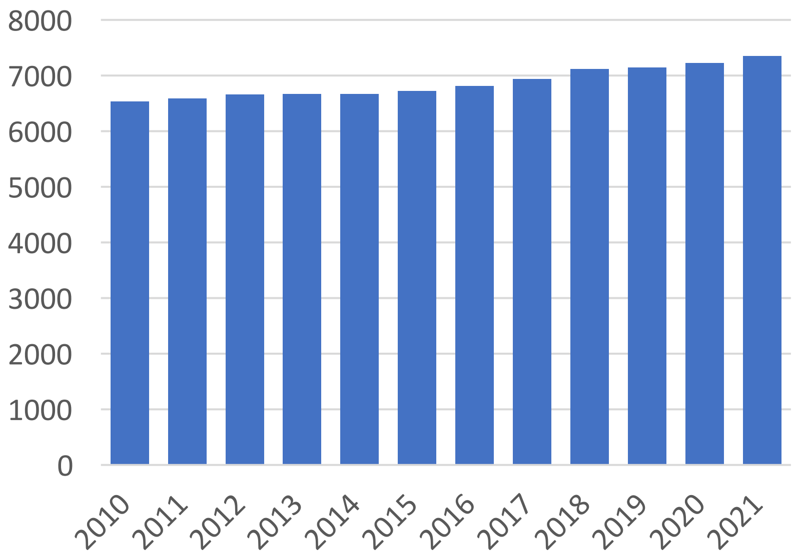
Italian Simmental

- dual-purpose, mainly farmed in small herds (< 20 heads)
- About 90k registered cattle, 57% farmed in the mountainous area of North-East Italy
- Average lactation (2022) = 7449 kg milk, 3.93 fat %, 3.42 protein %
- High fertility, low somatic cells, good beef performances
- Managed by the Italian Simmental Breeders Association

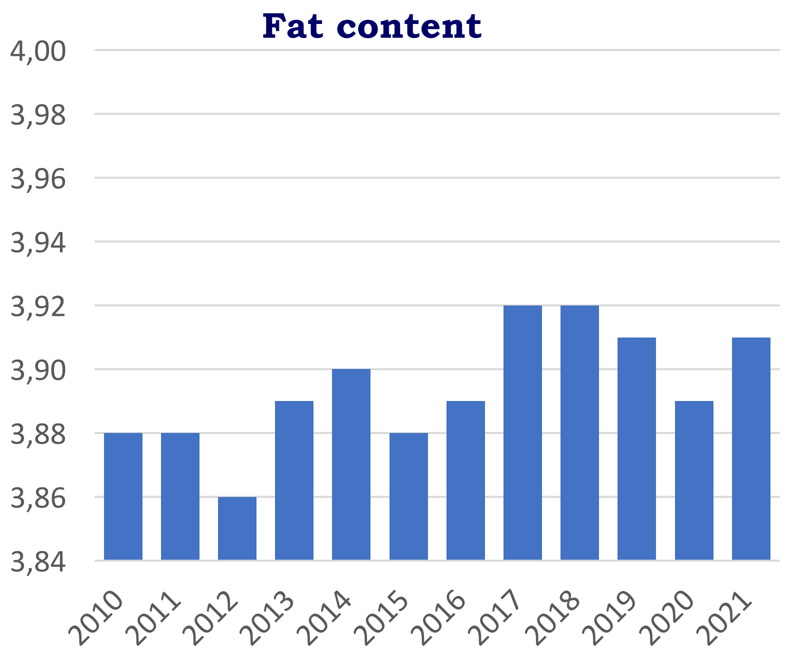
Some numbers



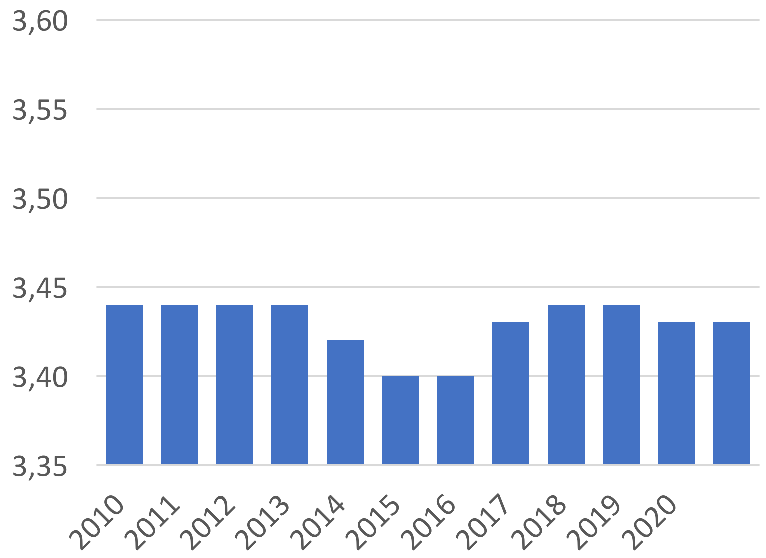
Cattle



Milk yield



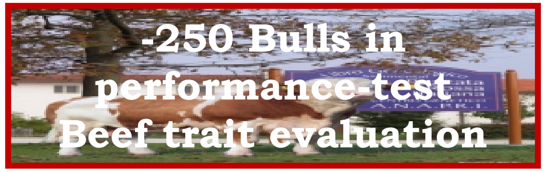
Fat content



Protein content

The breeding plan

Programmed matings:
800 Bull dams (half genotyped) (Rk 95)
20 Sires of bull 80% geomic, 20% progeny tested) (Rk 99)


-250 Bulls in performance-test
Beef trait evaluation

Genomic evaluation for production, morphology ad functional traits
ssGBLUP genetic evaluation:
- ✓ daily gain
- ✓ size
- ✓ muscularity
- ✓ Feet & legs
- ✓ RFI

Selected on the basis of IDAS PT

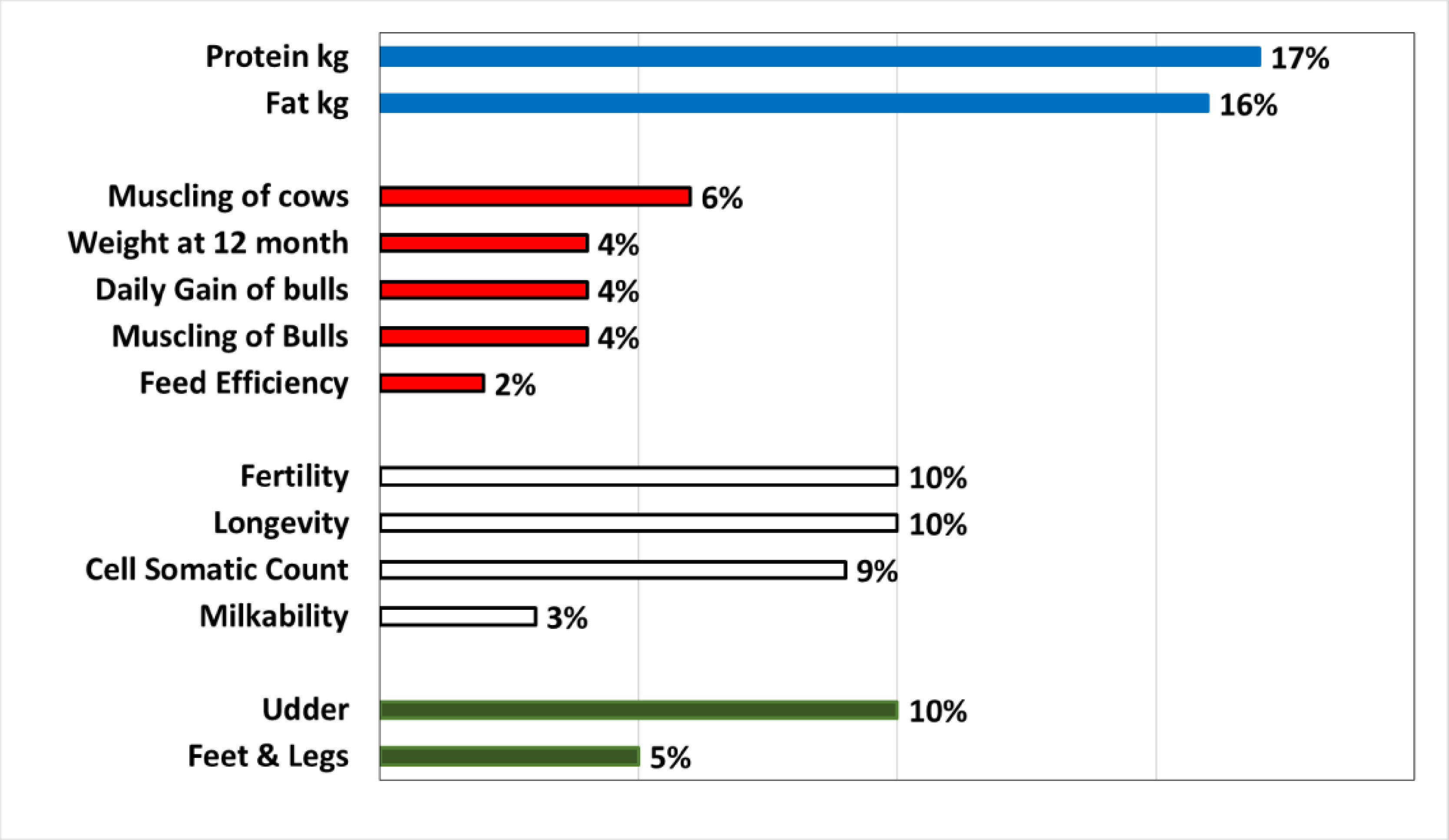
Best 25-30 YOUNG BULLS A.I.
- ✓ intensive use:35% A.I.
- ✓ max age prod. semen: 30 months
- ✓ semen doses: max 7000
- ✓ 180 day distribution period
- ✓ Semen RK<80 destroyed

Cow population subjected to DHI C.F.
≈ 65.000 cows

Evaluation ssGBLUP:
- ✓ milk, fat.%, pr.%, CCS
- ✓ beef traits
- ✓ morphology
- ✓ Fertility
- ✓ Longevity

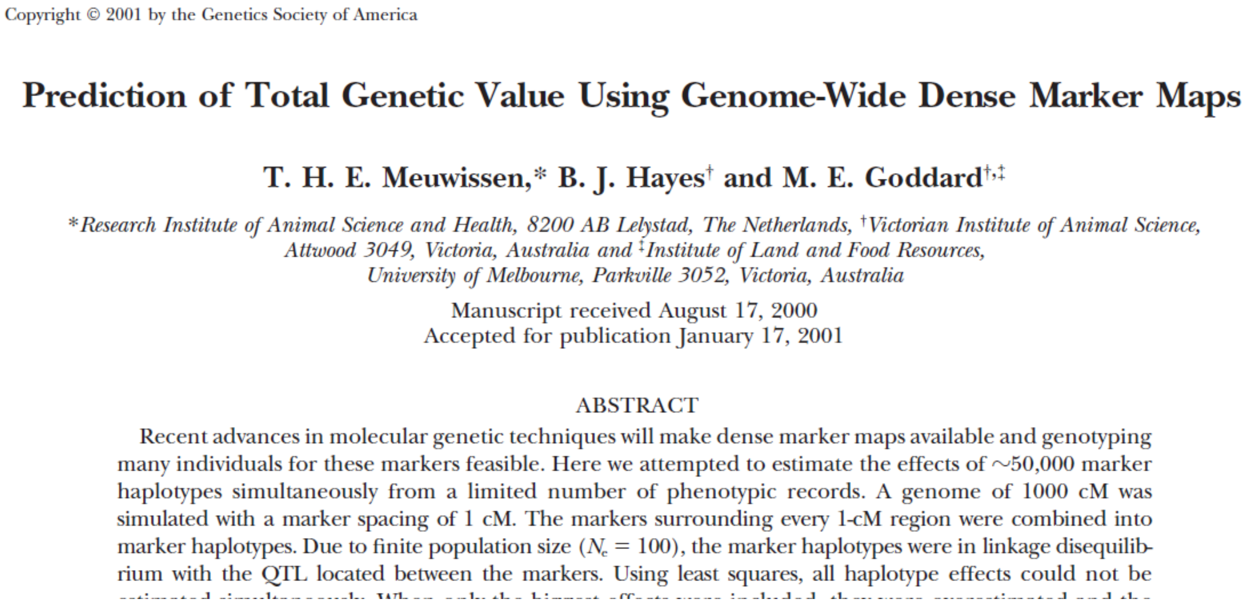
Selection based on IDAS

The selection index (IDAS)



- ✓33% milk
- ✓20% meat
- ✓15% morphology
- ✓32% fitness

The genomic revolution



Prediction of Total Genetic Value Using Genome-Wide Dense Marker Maps

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ABSTRACT

Recent advances in molecular genetic techniques will make dense marker maps available and genotyping many individuals for these markers feasible. Here we attempted to estimate the effects of ~50,000 marker haplotypes simultaneously from a limited number of phenotypic records. A genome of 1000 cM was simulated with a marker spacing of 1 cM. The markers surrounding every 1-cM region were combined into marker haplotypes. Due to finite population size ($N_e = 100$), the marker haplotypes were in linkage disequilibrium with the QTL located between the markers. Using least squares, all haplotype effects could not be

Genomic selection in Italian Simmental: some milestones

SELMOL
RICERCA E INNOVAZIONE
NELLE ATTIVITA' DI MIGLIORAMENTO GENETICO ANIMALE
MEDIANTE TECNICHE DI GENETICA MOLECOLARE
PER LA COMPETITIVITA DEL SISTEMA ZOOTECNICO NAZIONALE

- ✓**2009** **SELMOL** project:
  - ✓National bulls genotyping
  - ✓Development of a genomic evaluation procedure based on PCA
- ✓**2011/03** genotype exchange with DE/AT
- ✓**2011/11** INTERBULL GEBV validation test passed
- ✓**2011** Start of the genomic selection programme

The curse of dimensionality N<<P

✓About 480 national top bulls genotyped in the SELMOL project

$$\mathbf{M} = \begin{bmatrix} 0 & -1 & \ldots\ldots\ldots & 1 \\ 1 & 0 & \ldots\ldots\ldots & 1 \\ : & : & : & : \\ -1 & 0 & \ldots\ldots\ldots & 0 \end{bmatrix}_{(Nx\infty)}$$

✓Compression of SNP genotype information using dimension reduction techniques

✓Multivariate techniques as PCA or PLSR suggested for GS (Solberg et al., 2009; Macciotta et al., 2010)

The curse of dimensionality

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Prediction of genomic breeding values for dairy traits in Italian Brown and Simmental bulls using a principal component approach

M. A. Pintus,* G. Gaspa,* E. L. Nicolazzi,† D. Vicario,‡ A. Rossoni,§ P. Ajmone-Marsan,† A. Nardone,# C. Dimauro,* and N. P. P. Macciotta*[1]
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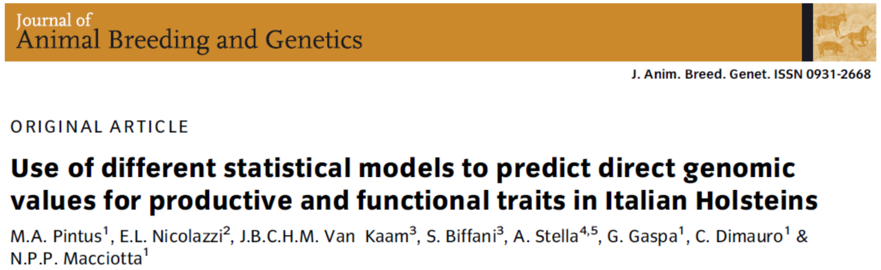


Journal of
Animal Breeding and Genetics

J. Anim. Breed. Genet. ISSN 0931-2668

ORIGINAL ARTICLE

Use of different statistical models to predict direct genomic values for productive and functional traits in Italian Holsteins

M.A. Pintus[1], E.L. Nicolazzi[2], J.B.C.H.M. Van Kaam[3], S. Biffani[3], A. Stella[4,5], G. Gaspa[1], C. Dimauro[1] & N.P.P. Macciotta[1]

Breed	SNP	PC	Exp. variance
Holstein	40,658	2,564	80%
Brown	37,254	2,257	80%
Simmental	40,179	2,476	70%

SNP (-1,0,1)

↓

PCA

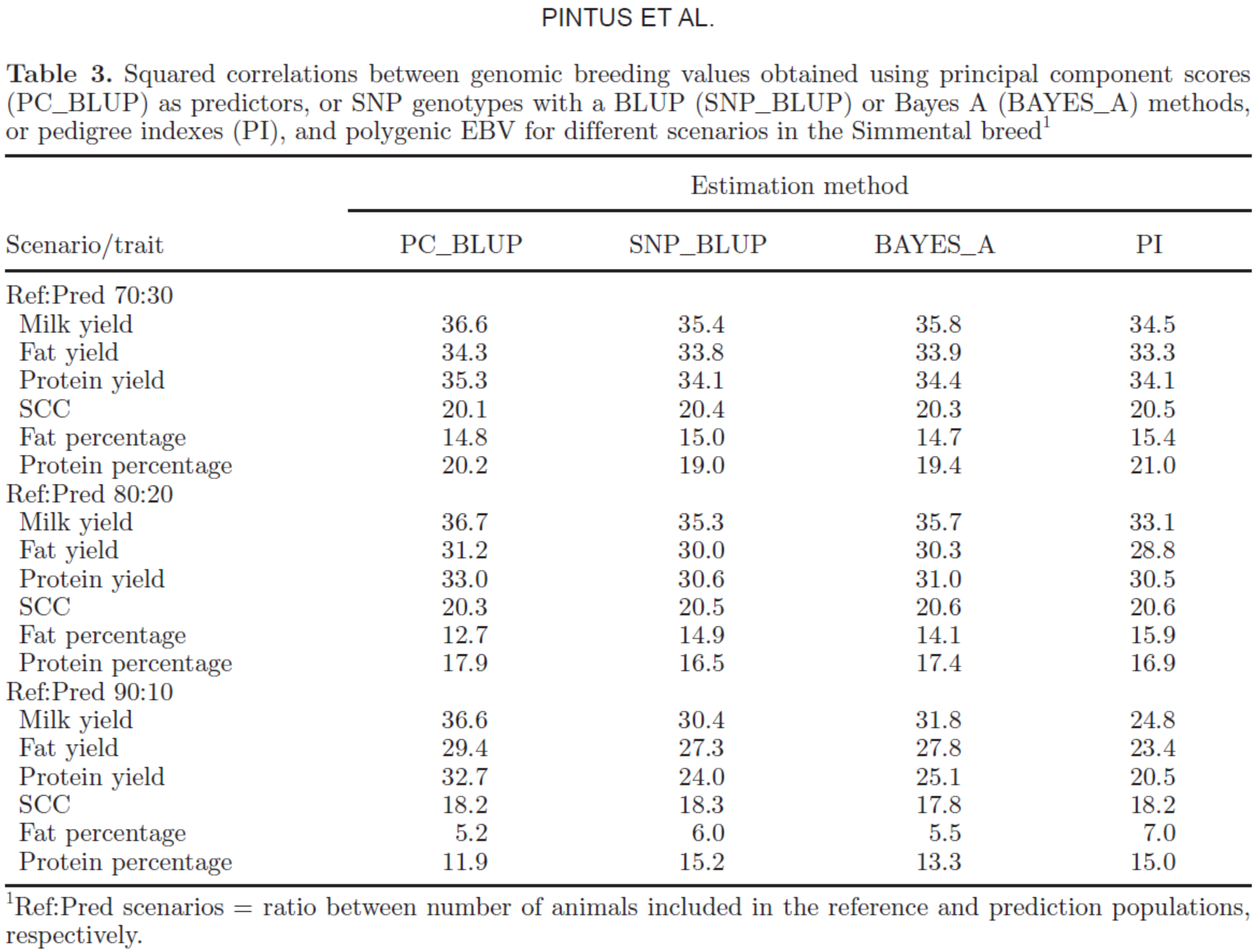
↓

Estimation of PC score effects

↓

DGV calculations

The curse of dimensionality



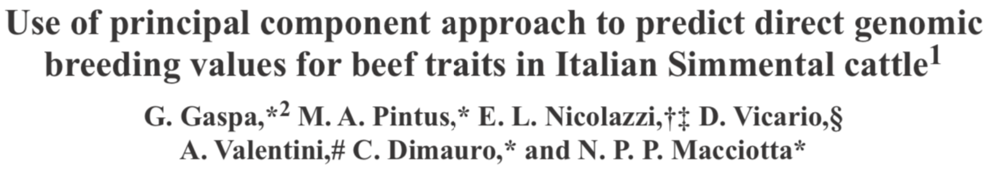
PINTUS ET AL.

Table 3. Squared correlations between genomic breeding values obtained using principal component scores (PC_BLUP) as predictors, or SNP genotypes with a BLUP (SNP_BLUP) or Bayes A (BAYES_A) methods, or pedigree indexes (PI), and polygenic EBV for different scenarios in the Simmental breed[1]

	Estimation method			
Scenario/trait	PC_BLUP	SNP_BLUP	BAYES_A	PI
Ref:Pred 70:30				
Milk yield	36.6	35.4	35.8	34.5
Fat yield	34.3	33.8	33.9	33.3
Protein yield	35.3	34.1	34.4	34.1
SCC	20.1	20.4	20.3	20.5
Fat percentage	14.8	15.0	14.7	15.4
Protein percentage	20.2	19.0	19.4	21.0
Ref:Pred 80:20				
Milk yield	36.7	35.3	35.7	33.1
Fat yield	31.2	30.0	30.3	28.8
Protein yield	33.0	30.6	31.0	30.5
SCC	20.3	20.5	20.6	20.6
Fat percentage	12.7	14.9	14.1	15.9
Protein percentage	17.9	16.5	17.4	16.9
Ref:Pred 90:10				
Milk yield	36.6	30.4	31.8	24.8
Fat yield	29.4	27.3	27.8	23.4
Protein yield	32.7	24.0	25.1	20.5
SCC	18.2	18.3	17.8	18.2
Fat percentage	5.2	6.0	5.5	7.0
Protein percentage	11.9	15.2	13.3	15.0

[1] Ref:Pred scenarios = ratio between number of animals included in the reference and prediction populations, respectively.

Beef traits



Use of principal component approach to predict direct genomic breeding values for beef traits in Italian Simmental cattle[1]

G. Gaspa,*[2] M. A. Pintus,* E. L. Nicolazzi,†‡ D. Vicario,§
A. Valentini,# C. Dimauro,* and N. P. P. Macciotta*

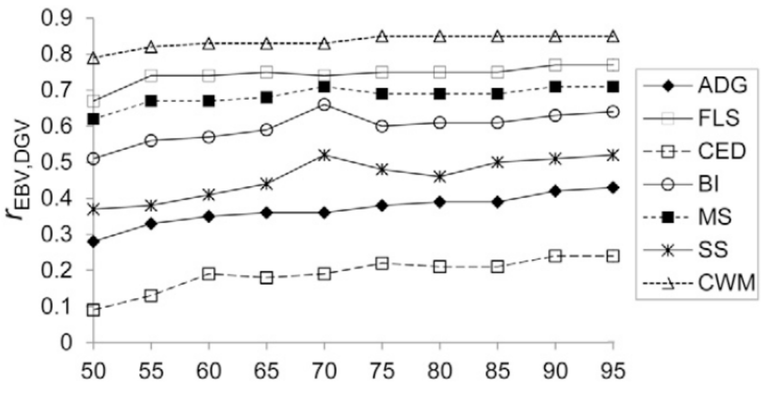


Figure 2. Pattern of correlation ($r_{EBV,DGV}$) between EBV and direct genomic breeding values (DGV), as function of percent of variance explained by the principal components (PC) for 7 meat traits using PC-BLUP (FLS = feet and leg score, CED = calving ease direct effect, BI = beef index, MS = muscularity score, SS = size score, CWM = cow muscularity).

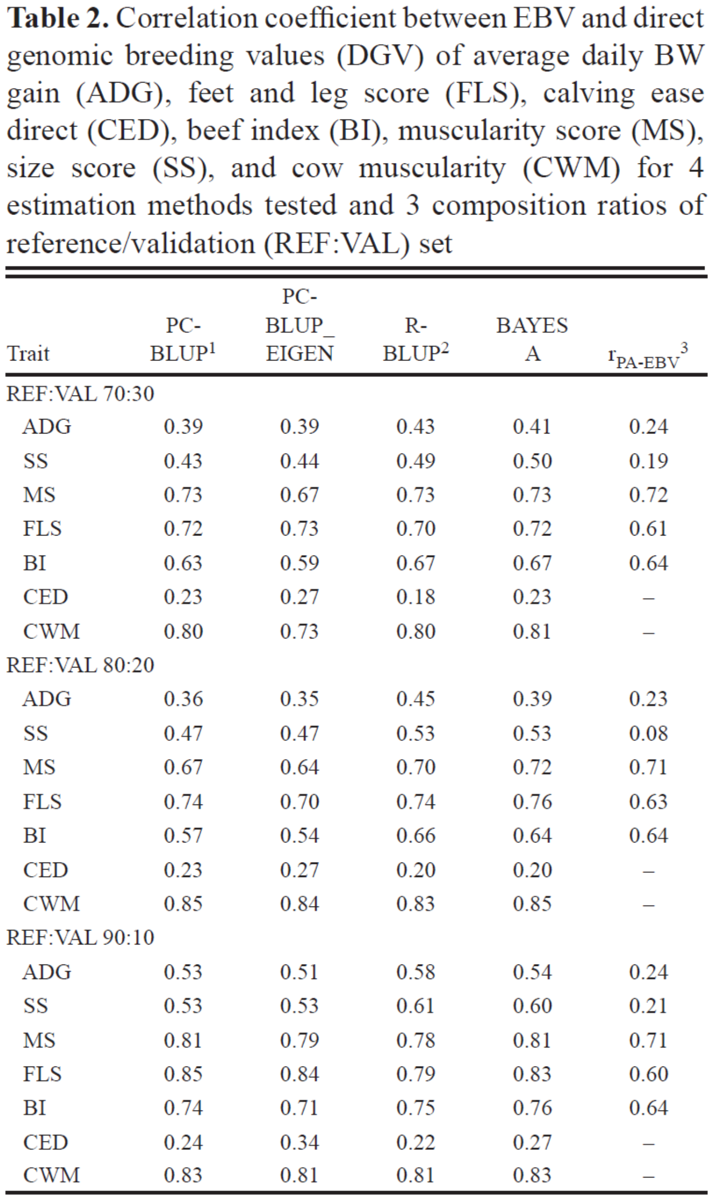
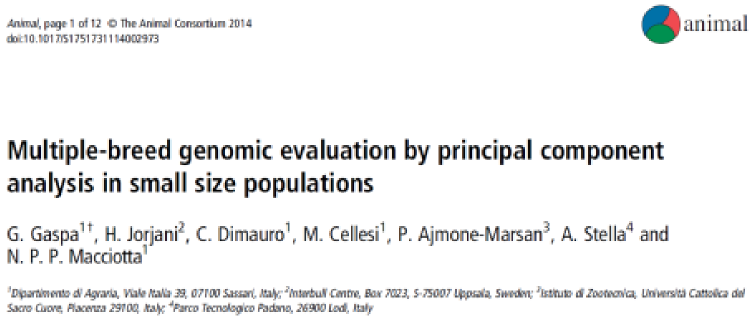


Table 2. Correlation coefficient between EBV and direct genomic breeding values (DGV) of average daily BW gain (ADG), feet and leg score (FLS), calving ease direct (CED), beef index (BI), muscularity score (MS), size score (SS), and cow muscularity (CWM) for 4 estimation methods tested and 3 composition ratios of reference/validation (REF:VAL) set

Trait	PC-BLUP[1]	PC-BLUP_EIGEN	R-BLUP[2]	BAYES A	$r_{PA\text{-}EBV}$[3]
REF:VAL 70:30					
ADG	0.39	0.39	0.43	0.41	0.24
SS	0.43	0.44	0.49	0.50	0.19
MS	0.73	0.67	0.73	0.73	0.72
FLS	0.72	0.73	0.70	0.72	0.61
BI	0.63	0.59	0.67	0.67	0.64
CED	0.23	0.27	0.18	0.23	–
CWM	0.80	0.73	0.80	0.81	–
REF:VAL 80:20					
ADG	0.36	0.35	0.45	0.39	0.23
SS	0.47	0.47	0.53	0.53	0.08
MS	0.67	0.64	0.70	0.72	0.71
FLS	0.74	0.70	0.74	0.76	0.63
BI	0.57	0.54	0.66	0.64	0.64
CED	0.23	0.27	0.20	0.20	–
CWM	0.85	0.84	0.83	0.85	–
REF:VAL 90:10					
ADG	0.53	0.51	0.58	0.54	0.24
SS	0.53	0.53	0.61	0.60	0.21
MS	0.81	0.79	0.78	0.81	0.71
FLS	0.85	0.84	0.79	0.83	0.60
BI	0.74	0.71	0.75	0.76	0.64
CED	0.24	0.34	0.22	0.27	–
CWM	0.83	0.81	0.81	0.83	–

Multi-breed approach



Animal, page 1 of 12 © The Animal Consortium 2014
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animal

Multiple-breed genomic evaluation by principal component analysis in small size populations

G. Gaspa[1†], H. Jorjani[2], C. Dimauro[1], M. Cellesi[1], P. Ajmone-Marsan[3], A. Stella[4] and
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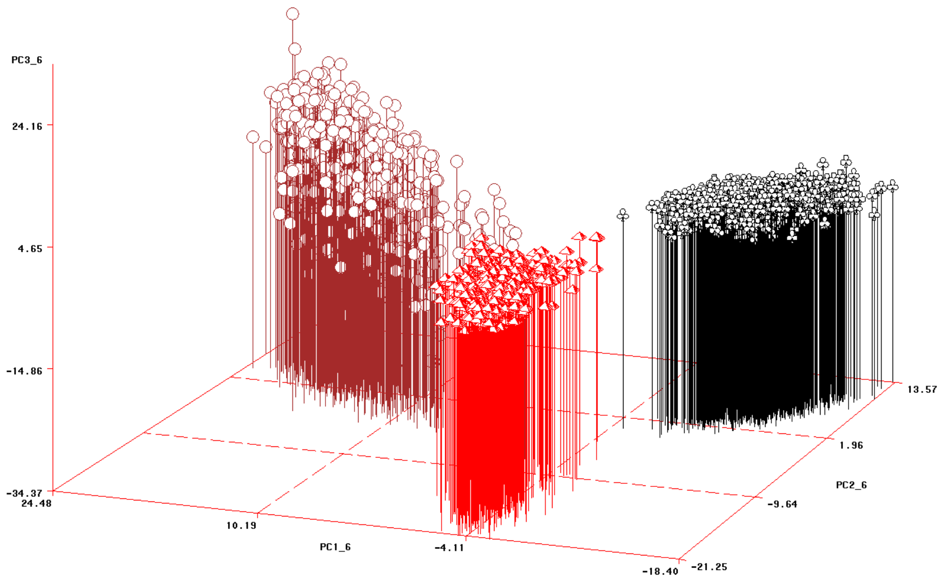

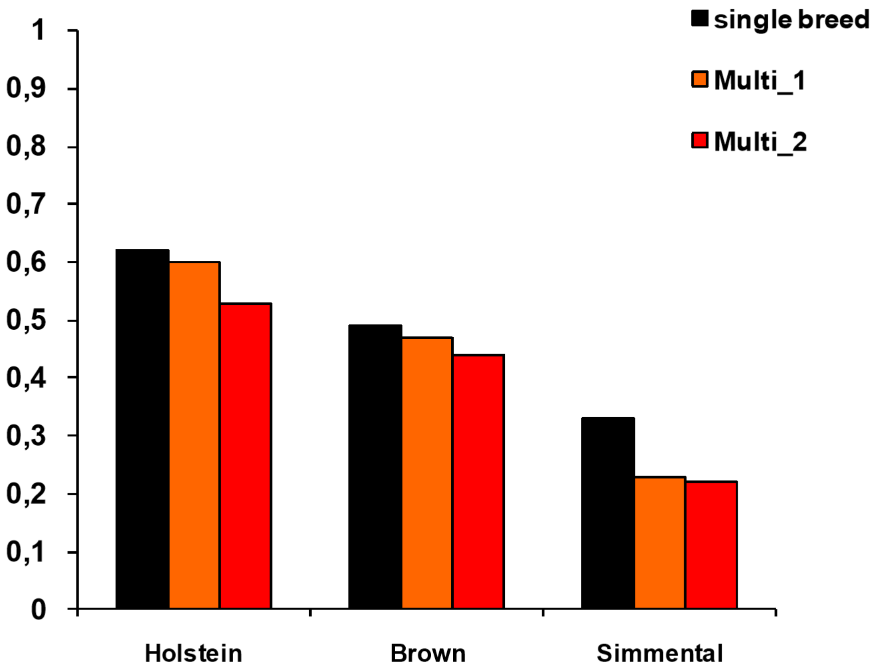

Genomic selection in Italian Simmental: some milestones - *bis*

- ✓**2011** INNOVAGEN
- ✓**2012** GENE2FARM
- ✓**2012** Genotype exchange with Czech republic
- ✓**2014** Genotype exchange with Switzerland
- ✓**2014** ONESTEP project start
- ✓**2015** change from PCA to SNP-BLUP
- ✓**2016** adoption of ssGBLUP
- ✓**2018** DUALBREEDING

ssGBLUP

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Genomic information allows for more accurate breeding values for milkability in dual-purpose Italian Simmental cattle

A. Cesarani,[1*] A. Garcia,[1] J. Hidalgo,[1] L. Degano,[2] D. Vicario,[2] N. P. P. Macciotta,[3] and D. Lourenco[1]
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[3]Department of Agricultural Sciences, University of Sassari, 07100 Sassari, Italy

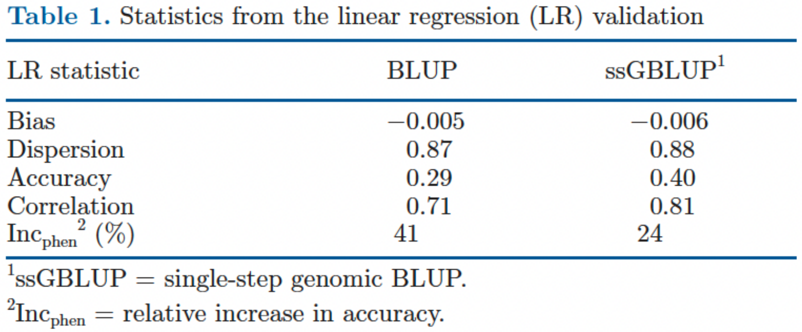
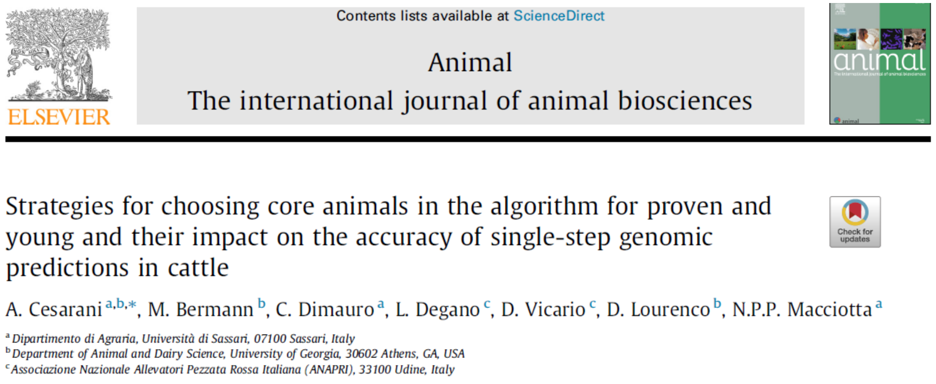


Table 1. Statistics from the linear regression (LR) validation

LR statistic	BLUP	ssGBLUP[1]
Bias	−0.005	−0.006
Dispersion	0.87	0.88
Accuracy	0.29	0.40
Correlation	0.71	0.81
$\text{Inc}_{\text{phen}}^{2}$ (%)	41	24

[1]ssGBLUP = single-step genomic BLUP.
[2]Inc_{phen} = relative increase in accuracy.



Contents lists available at ScienceDirect

Animal
The international journal of animal biosciences

Strategies for choosing core animals in the algorithm for proven and young and their impact on the accuracy of single-step genomic predictions in cattle

A. Cesarani[a,b,*], M. Bermann[b], C. Dimauro[a], L. Degano[c], D. Vicario[c], D. Lourenco[b], N.P.P. Macciotta[a]
[a] Dipartimento di Agraria, Università di Sassari, 07100 Sassari, Italy
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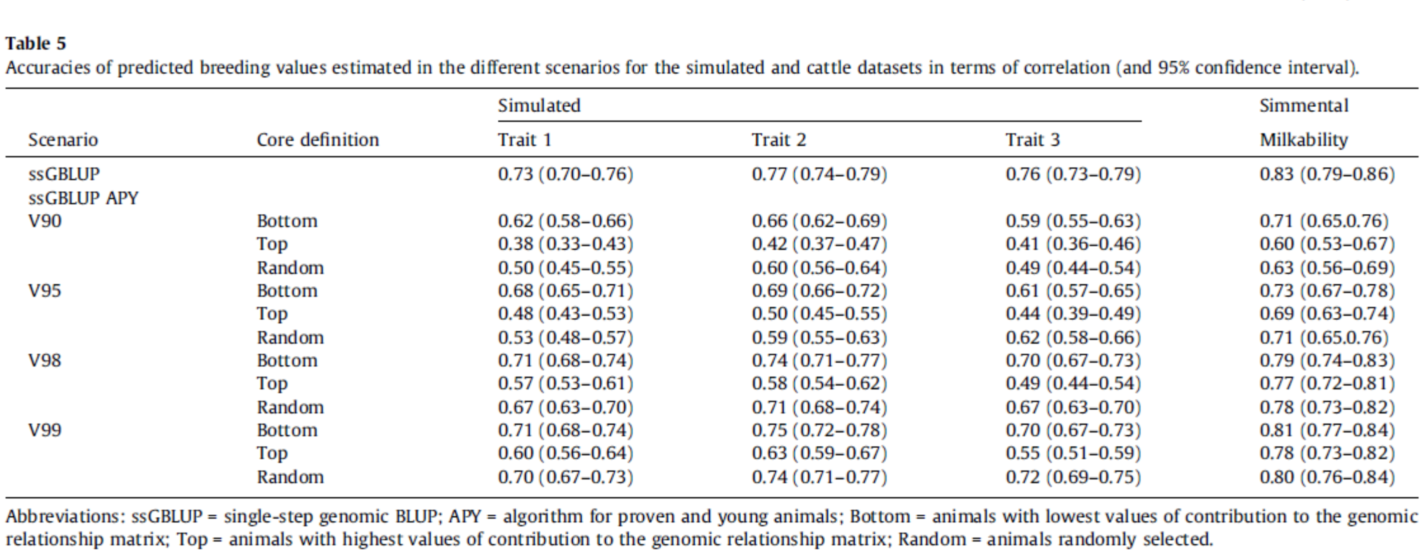
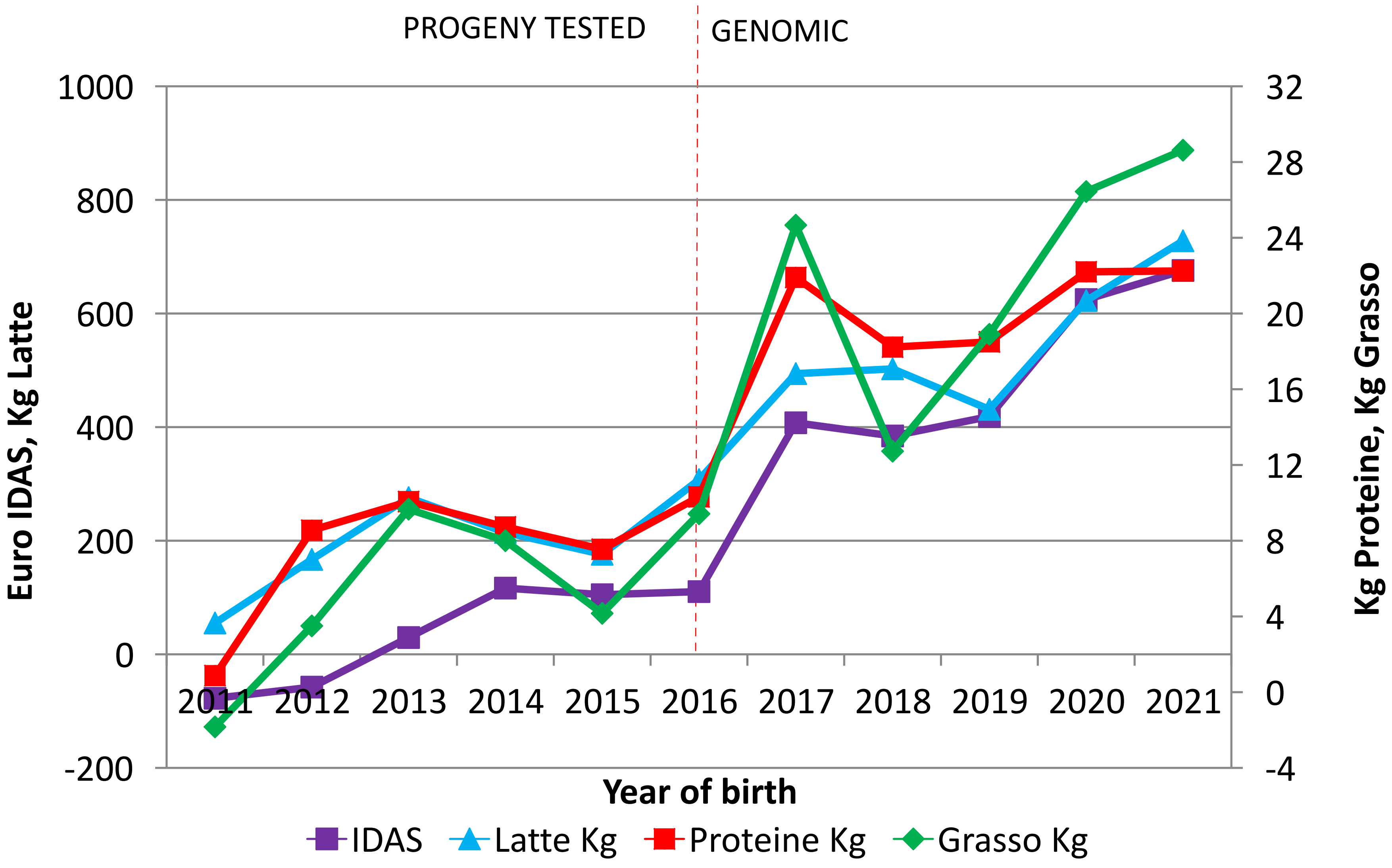


Table 5
Accuracies of predicted breeding values estimated in the different scenarios for the simulated and cattle datasets in terms of correlation (and 95% confidence interval).

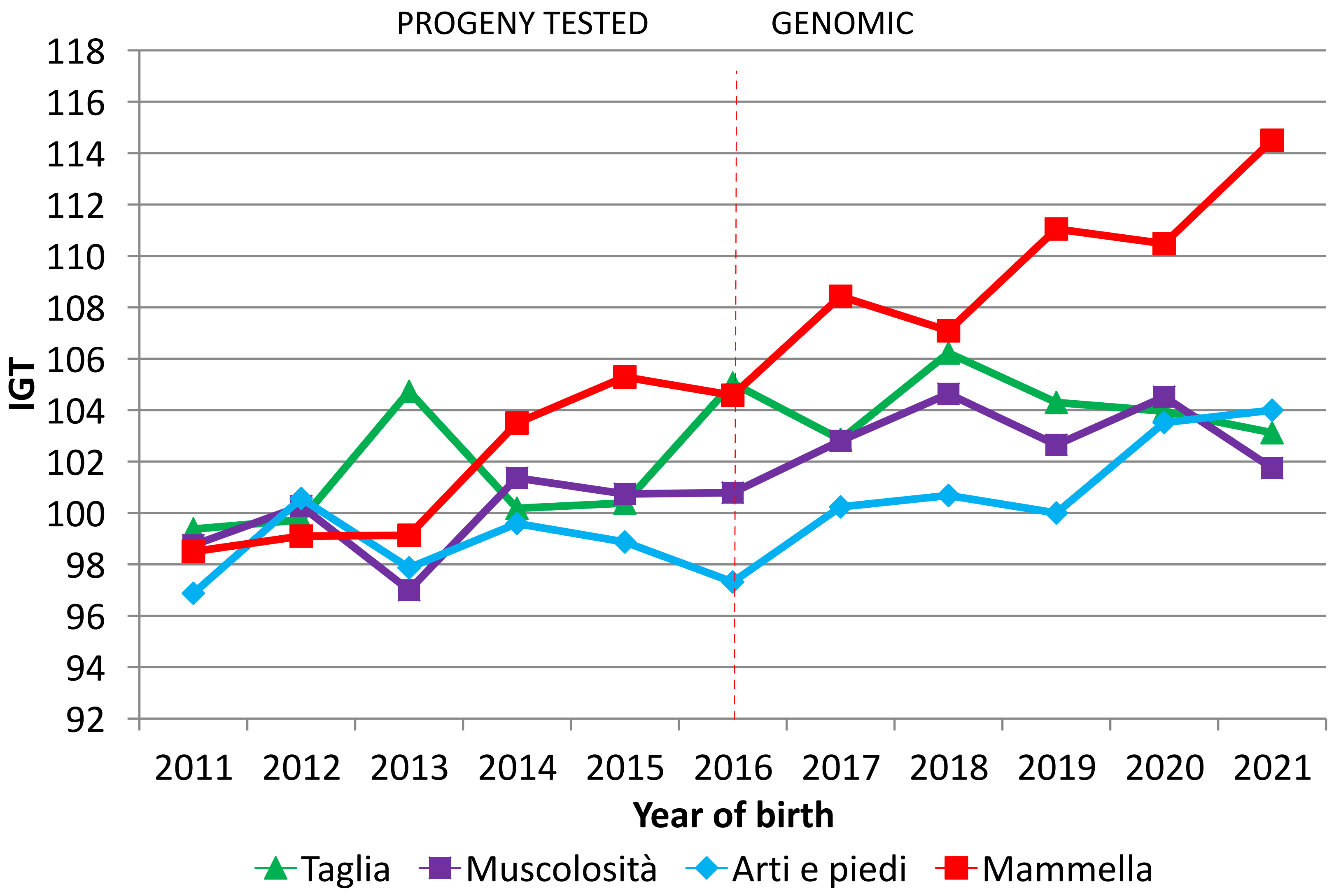
Scenario	Core definition	Simulated			Simmental
		Trait 1	Trait 2	Trait 3	Milkability
ssGBLUP		0.73 (0.70–0.76)	0.77 (0.74–0.79)	0.76 (0.73–0.79)	0.83 (0.79–0.86)
ssGBLUP APY					
V90	Bottom	0.62 (0.58–0.66)	0.66 (0.62–0.69)	0.59 (0.55–0.63)	0.71 (0.65.0.76)
	Top	0.38 (0.33–0.43)	0.42 (0.37–0.47)	0.41 (0.36–0.46)	0.60 (0.53–0.67)
	Random	0.50 (0.45–0.55)	0.60 (0.56–0.64)	0.49 (0.44–0.54)	0.63 (0.56–0.69)
V95	Bottom	0.68 (0.65–0.71)	0.69 (0.66–0.72)	0.61 (0.57–0.65)	0.73 (0.67–0.78)
	Top	0.48 (0.43–0.53)	0.50 (0.45–0.55)	0.44 (0.39–0.49)	0.69 (0.63–0.74)
	Random	0.53 (0.48–0.57)	0.59 (0.55–0.63)	0.62 (0.58–0.66)	0.71 (0.65.0.76)
V98	Bottom	0.71 (0.68–0.74)	0.74 (0.71–0.77)	0.70 (0.67–0.73)	0.79 (0.74–0.83)
	Top	0.57 (0.53–0.61)	0.58 (0.54–0.62)	0.49 (0.44–0.54)	0.77 (0.72–0.81)
	Random	0.67 (0.63–0.70)	0.71 (0.68–0.74)	0.67 (0.63–0.70)	0.78 (0.73–0.82)
V99	Bottom	0.71 (0.68–0.74)	0.75 (0.72–0.78)	0.70 (0.67–0.73)	0.81 (0.77–0.84)
	Top	0.60 (0.56–0.64)	0.63 (0.59–0.67)	0.55 (0.51–0.59)	0.78 (0.73–0.82)
	Random	0.70 (0.67–0.73)	0.74 (0.71–0.77)	0.72 (0.69–0.75)	0.80 (0.76–0.84)

Abbreviations: ssGBLUP = single-step genomic BLUP; APY = algorithm for proven and young animals; Bottom = animals with lowest values of contribution to the genomic relationship matrix; Top = animals with highest values of contribution to the genomic relationship matrix; Random = animals randomly selected.

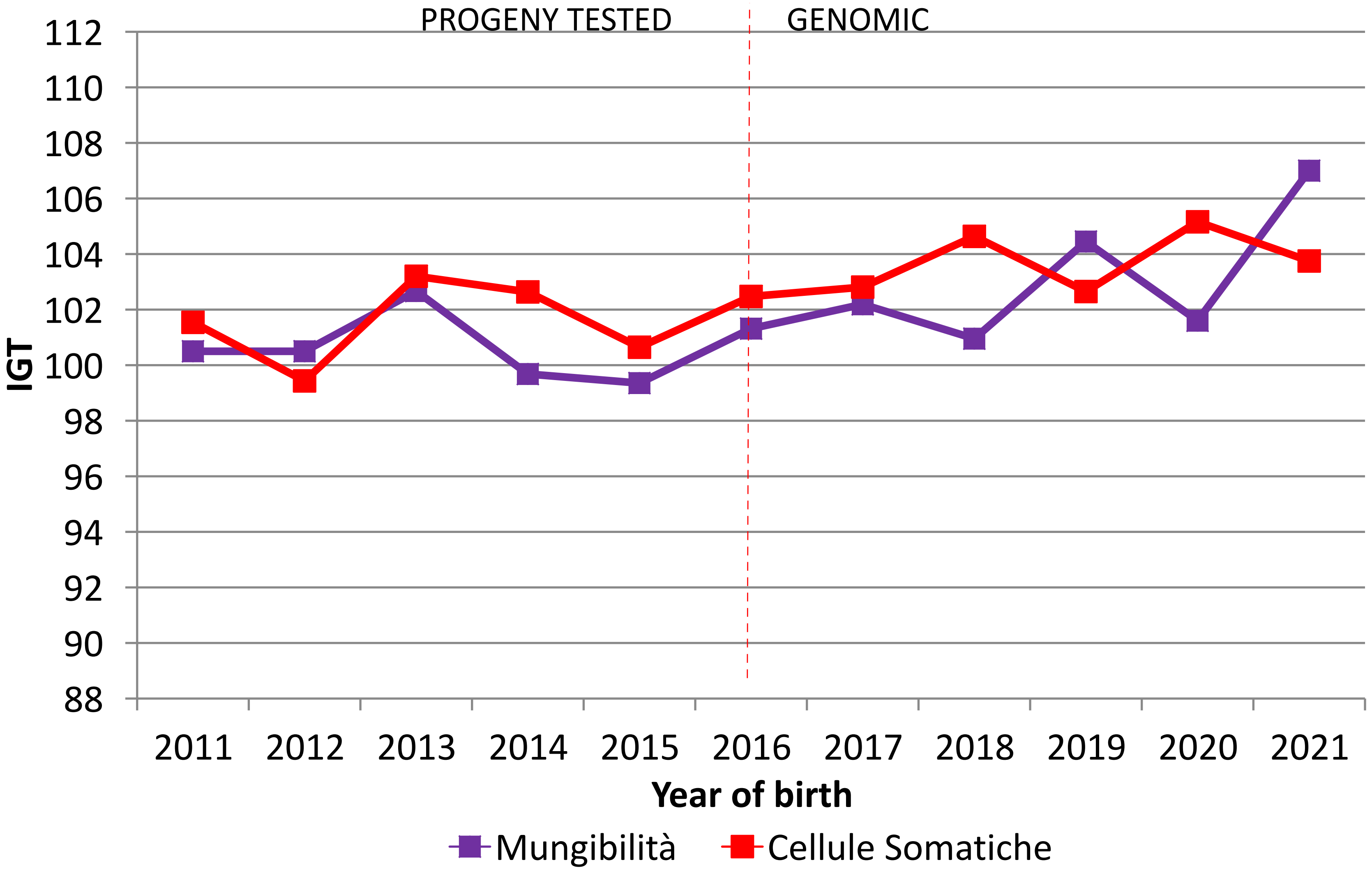
Genetic trend national bulls



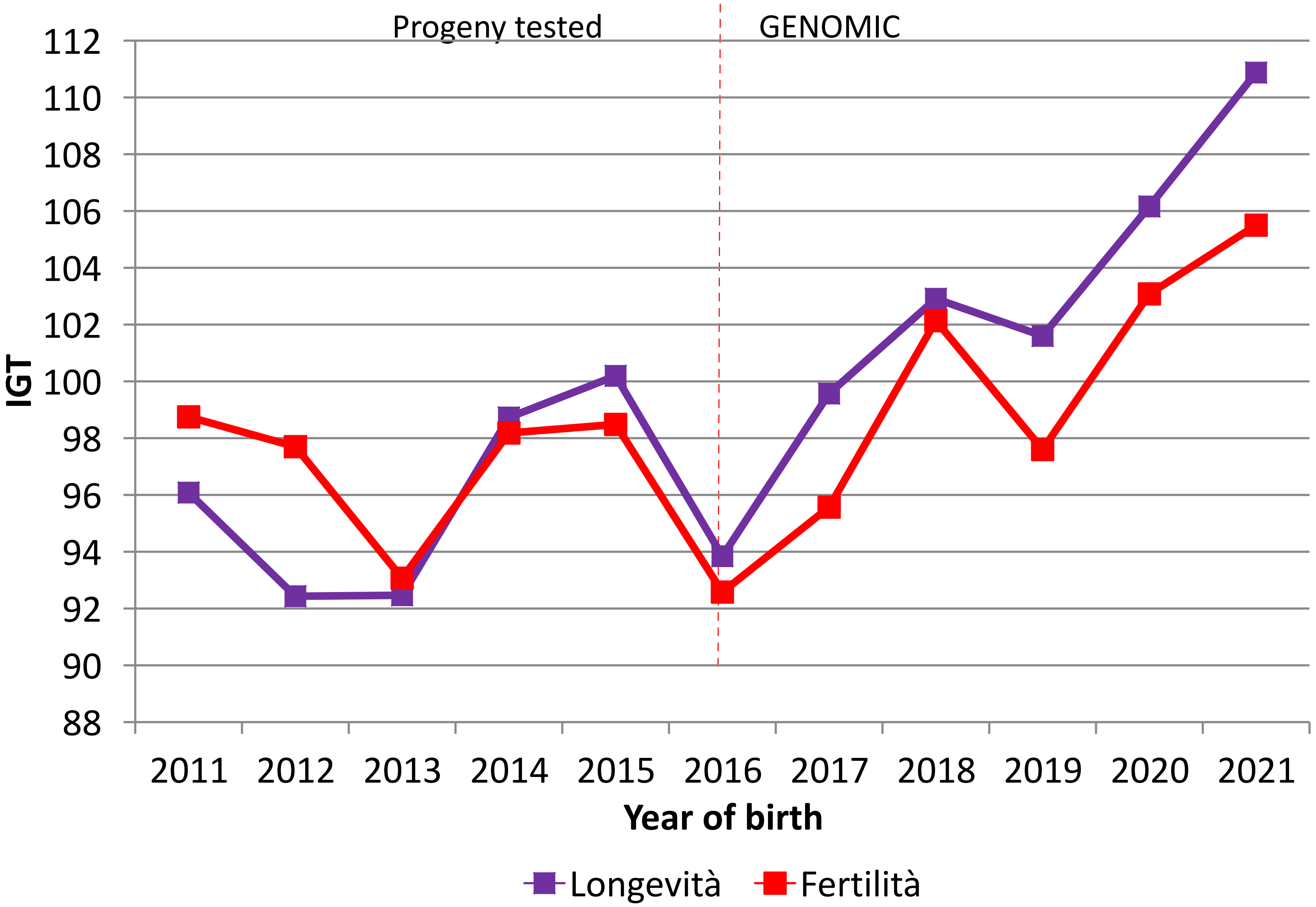
Genetic trend national bulls



Genetic trend national bulls

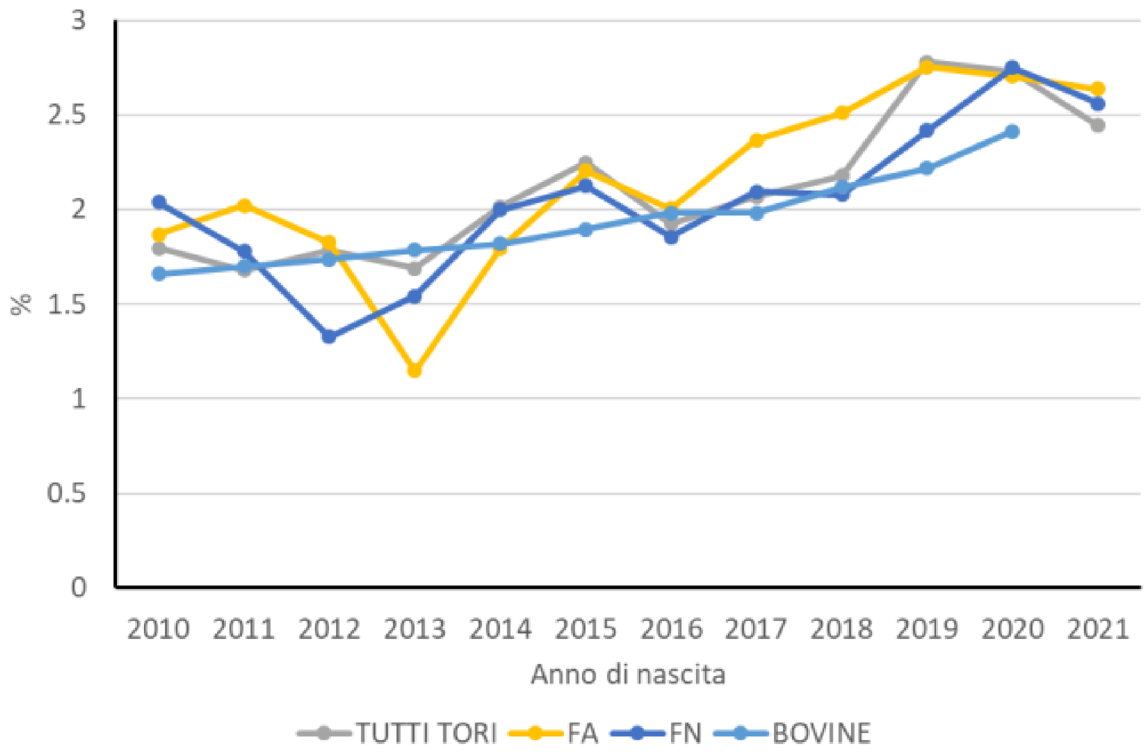


Genetic trend national bulls

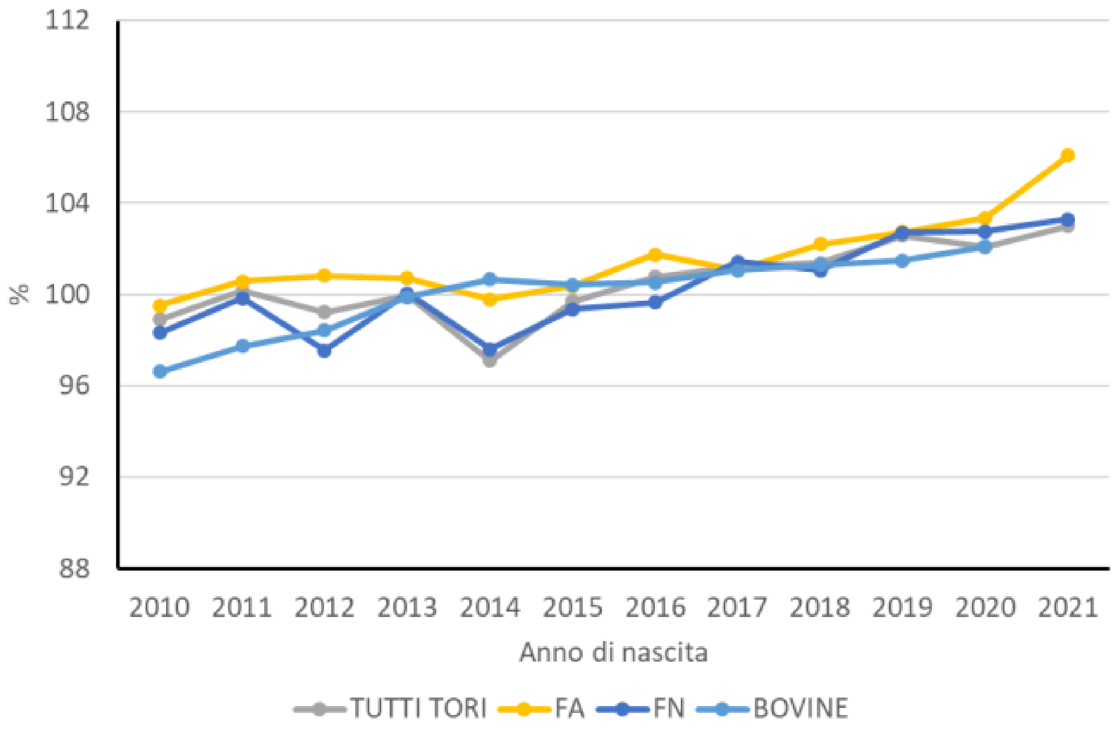


INBREEDING TREND

Pedigree

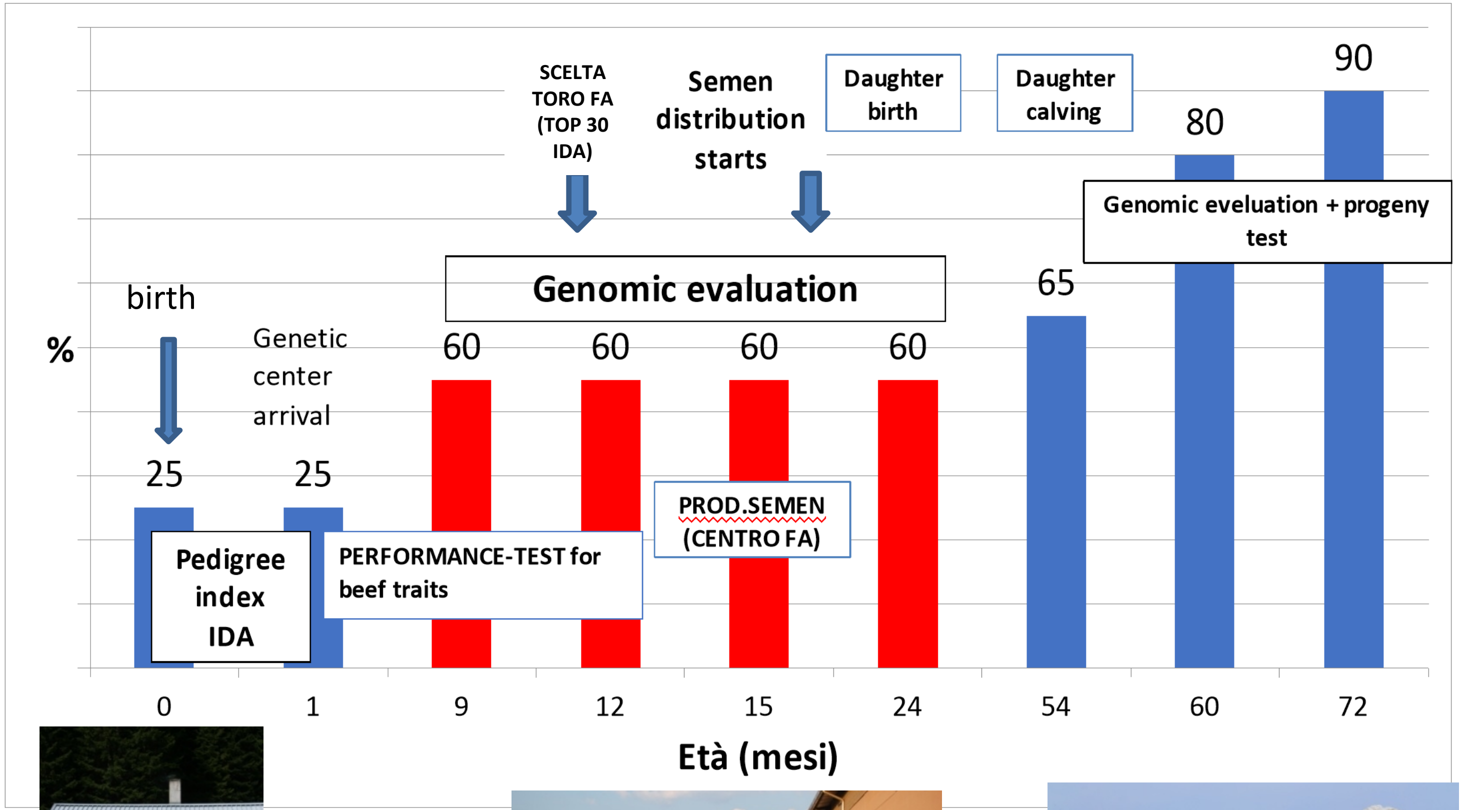


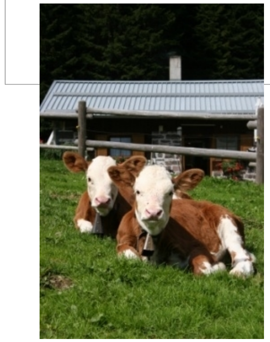
Genomic standardized



dualbreeding

Variation of EBV reliability during a bull career





ROH & Genomic inbreeding

ANIMAL GENETICS Immunogenetics, Molecular Genetics and Functional Genomics

doi: 10.1111/age.12259

Analysis of runs of homozygosity and their relationship with inbreeding in five cattle breeds farmed in Italy

Gabriele Marras[*,§], Giustino Gaspa[*], Silvia Sorbolini[*], Corrado Dimauro[*], Paolo Ajmone-Marsan[†], Alessio Valentini[‡], John L. Williams[§] and Nicolò P. P. Macciotta[*]

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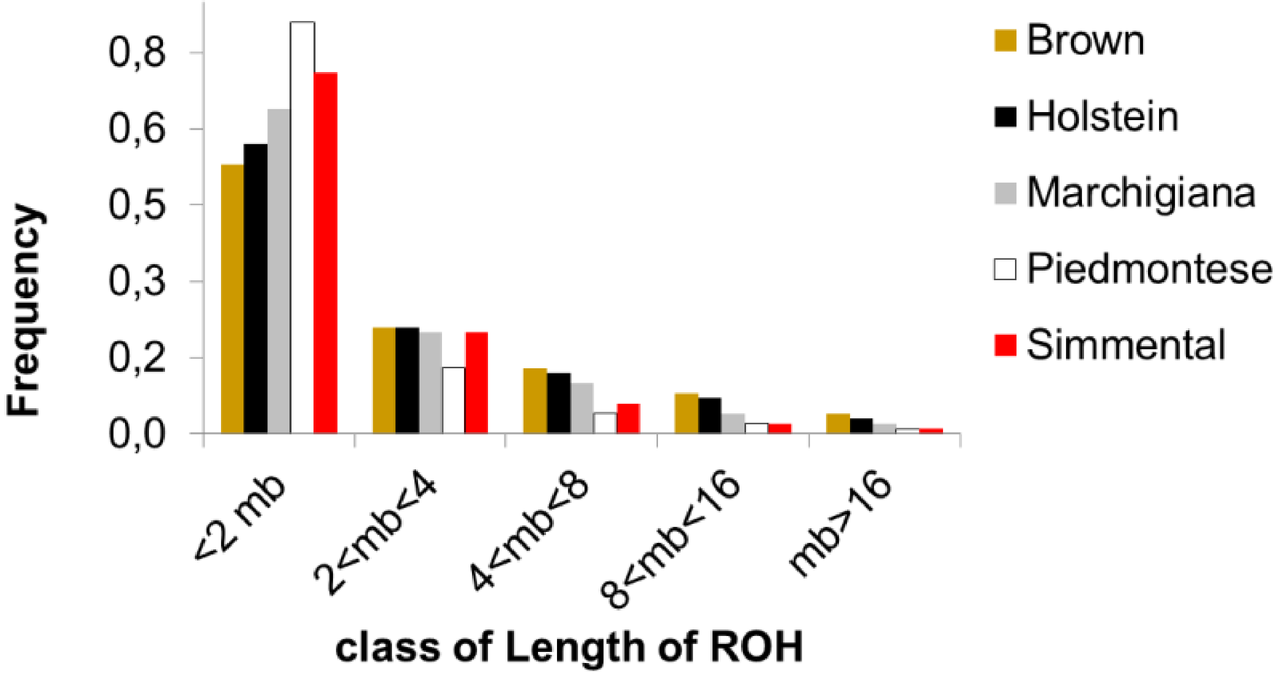
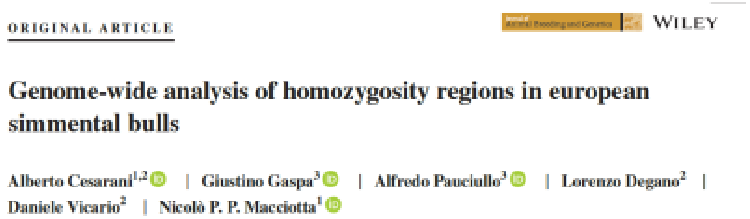
Breed	F_{PED} ± *sd*	F_{ROH} ± *sd*	$F_{ROH>2}$ ± *sd*	$F_{ROH>4}$ ± *sd*	$F_{ROH>8}$ ± *sd*	$F_{ROH>16}$ ± *sd*
Brown	0.026±0.021	0.145±0.038	0.119±0.038	0.097±0.036	0.068±0.032	0.034±0.023
Holstein	0.044±0.022	0.116±0.030	0.092±0.030	0.073±0.029	0.051±0.026	0.026±0.021
Simmental	0.008±0.013	0.083±0.022	0.048±0.022	0.028±0.021	0.015±0.018	0.007±0.014



ROH & Biodiversity



ORIGINAL ARTICLE

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Genome-wide analysis of homozygosity regions in european simmental bulls

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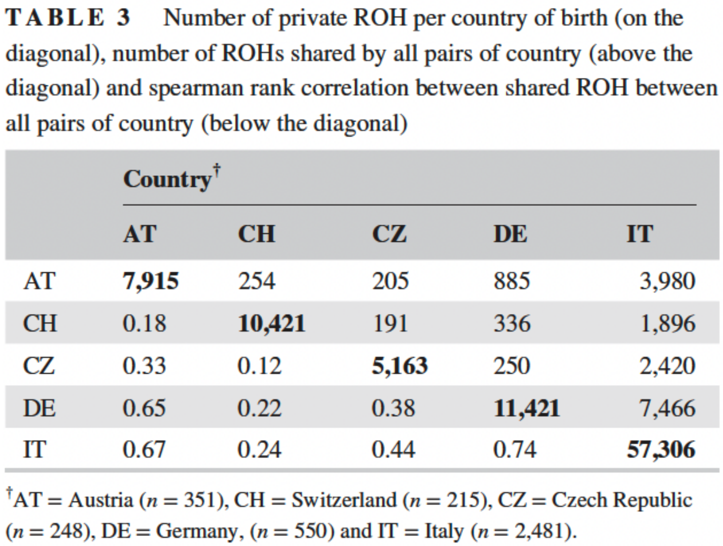
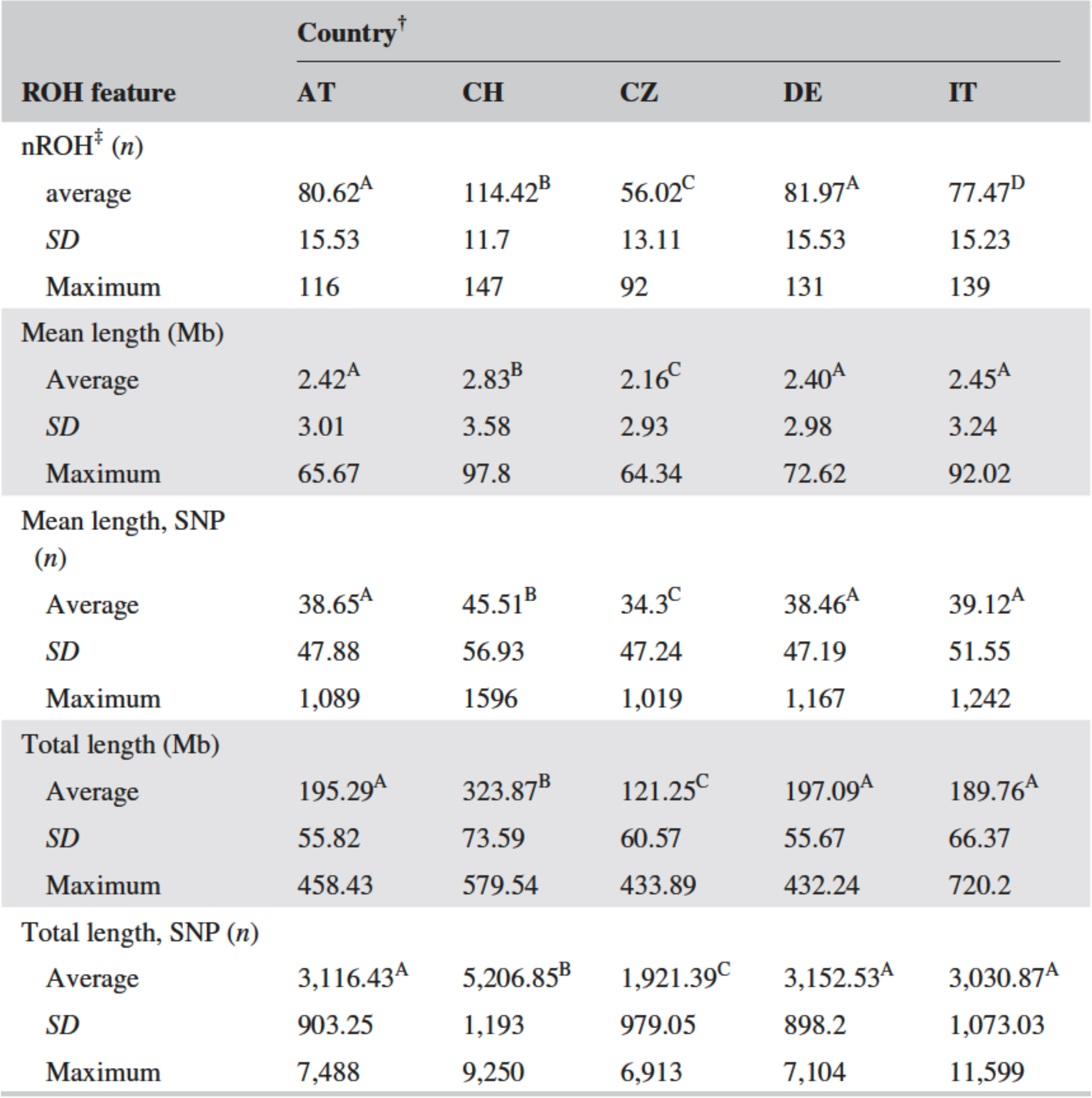


TABLE 3 Number of private ROH per country of birth (on the diagonal), number of ROHs shared by all pairs of country (above the diagonal) and spearman rank correlation between shared ROH between all pairs of country (below the diagonal)

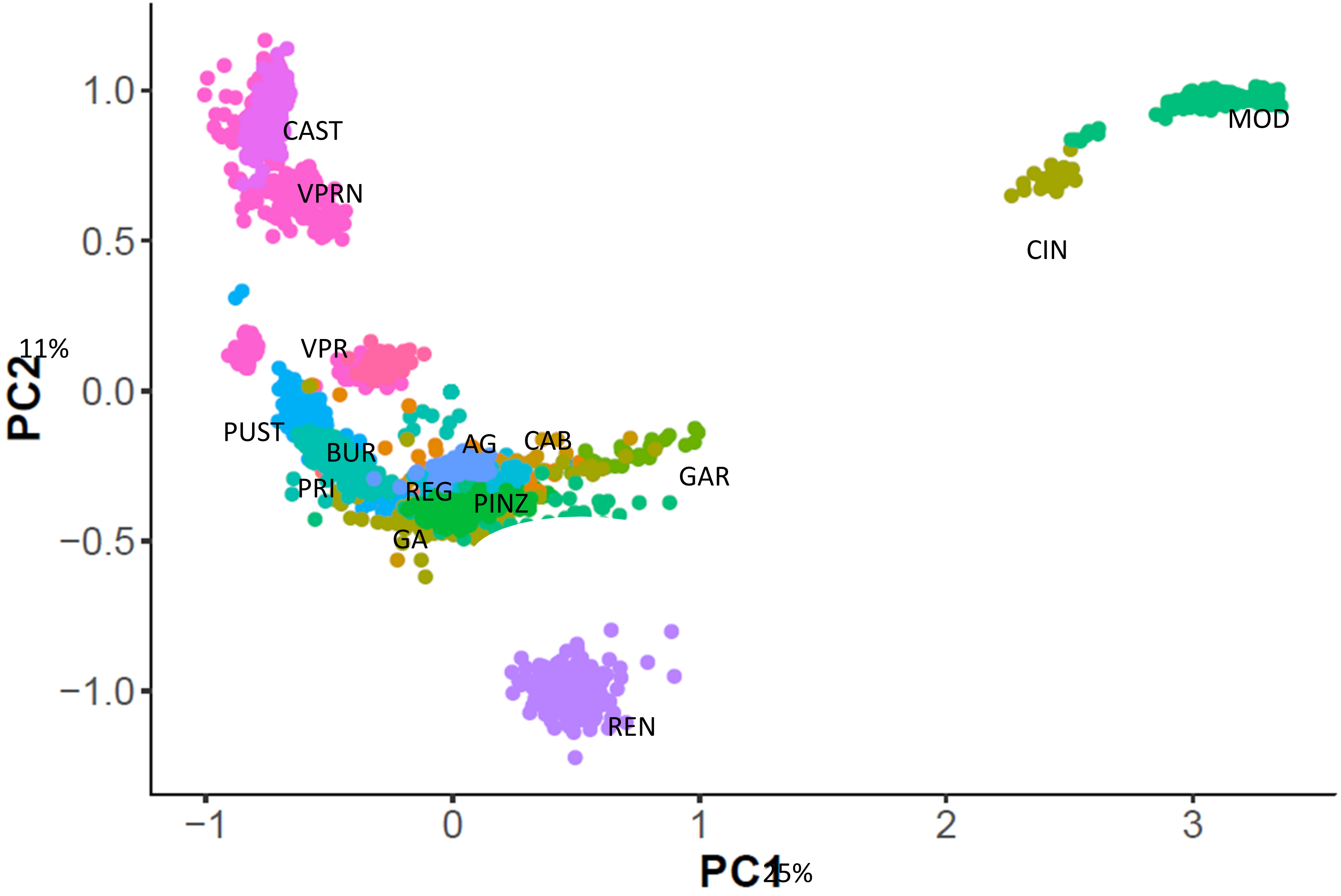
	Country[†]				
	AT	CH	CZ	DE	IT
AT	**7,915**	254	205	885	3,980
CH	0.18	**10,421**	191	336	1,896
CZ	0.33	0.12	**5,163**	250	2,420
DE	0.65	0.22	0.38	**11,421**	7,466
IT	0.67	0.24	0.44	0.74	**57,306**

[†] AT = Austria (n = 351), CH = Switzerland (n = 215), CZ = Czech Republic (n = 248), DE = Germany, (n = 550) and IT = Italy (n = 2,481).



	Country[†]				
ROH feature	**AT**	**CH**	**CZ**	**DE**	**IT**
nROH[‡] (n)					
average	80.62^{A}	114.42^{B}	56.02^{C}	81.97^{A}	77.47^{D}
SD	15.53	11.7	13.11	15.53	15.23
Maximum	116	147	92	131	139
Mean length (Mb)					
Average	2.42^{A}	2.83^{B}	2.16^{C}	2.40^{A}	2.45^{A}
SD	3.01	3.58	2.93	2.98	3.24
Maximum	65.67	97.8	64.34	72.62	92.02
Mean length, SNP (n)					
Average	38.65^{A}	45.51^{B}	34.3^{C}	38.46^{A}	39.12^{A}
SD	47.88	56.93	47.24	47.19	51.55
Maximum	1,089	1596	1,019	1,167	1,242
Total length (Mb)					
Average	195.29^{A}	323.87^{B}	121.25^{C}	197.09^{A}	189.76^{A}
SD	55.82	73.59	60.57	55.67	66.37
Maximum	458.43	579.54	433.89	432.24	720.2
Total length, SNP (n)					
Average	$3{,}116.43^{A}$	$5{,}206.85^{B}$	$1{,}921.39^{C}$	$3{,}152.53^{A}$	$3{,}030.87^{A}$
SD	903.25	1,193	979.05	898.2	1,073.03
Maximum	7,488	9,250	6,913	7,104	11,599

PCA of genomic relationship matrix of italian dual-purpose cattle breeds



Residual feed intake

	Mean ± PSD	CI 95%
Heritability	0.293±0.147	0.052 - 0.580

Method	r	b0	b1
BLUP	0.72	-0.01	1.14
ssGBLUP	0.74	-0.01	1.17

Genomic selection in Italia
Simmental - lights

- ✓Impact in the genetic trend
- ✓Development of a in house GS method
- ✓Strong collaboration with other European Simmental populations
- ✓Training of young scientists

MASSIMILIANO

gIDA +1341 kg , n.1 in top ranking in Italia, n.43 in Germania.

Esport verso DE – AT circa 19.000

Genomic selection in Italian Simmental – future challenges

✓Competition of foreign genomic bulls semen

✓Non randomized use of young bulls

✓Inbreeding management

✓Smaller breeds?

Thank you

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Multi-breed approach