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Animal and Food Genomics Group

Mining the genome of Italian animal genetic resources provided hints on peculiar genetic features: Examples of successful stories

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ERFP 2024 meeting - Florence, 29 August

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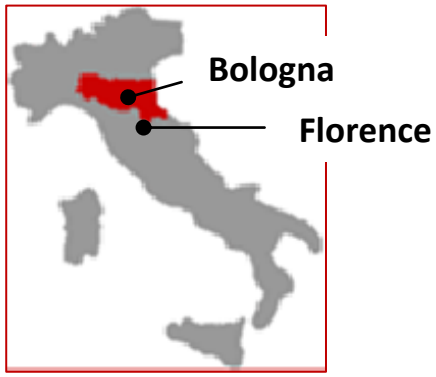
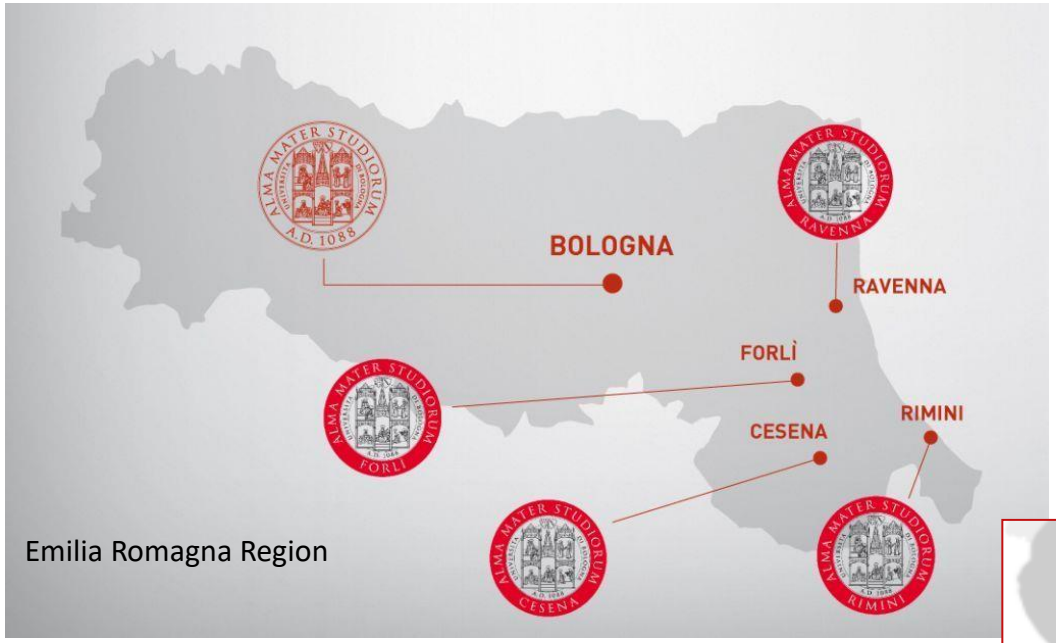


University Students: 1383
Frammenti dell'Arca di Giovanni da Legnano



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Characterisation of Italian animal genetic resources

Species

1. Cattle 
2. Pig 
3. Sheep 
4. Goat
5. Rabbit 
6. Horse
7. Donkey 
8. Turkey
9. Honeybees
10. ….

Selected case studies

Characterisation of Italian animal genetic resources

	Species	Types of information/datasets
1.	Cattle	Phenotypes; Candidate genes; SNP Chip data; NUMTs; WGS
2.	Pig	Phenotypes; Candidate genes; SNP Chip data; NUMTs; WGS
3.	Sheep	Phenotypes; Candidate genes; SNP Chip data
4.	Goat	Phenotypes; Candidate genes
5.	Rabbit	Phenotypes; Candidate genes; SNP Chip data; NUMTs; WGS
6.	Horse	Phenotypes; Candidate genes
7.	Donkey	Phenotypes; Candidate genes; WGS
8.	Turkey	NUMTs
9.	Honeybees	Phenotypes; mtDNA; Candidate genes; SNP Chip data; WGS
10.	….	

Pigmentation genetics

1 Cattle (I)

Co-funded by the European Union

MINISTERO DELL'AGRICOLTURA DELLA SOVRANITÀ ALIMENTARE E DELLE FORESTE

Reggiana

SNP chip data (n. cattle)	4075
Morphological characterisation (n. cattle)	2969
Whole Genome Sequencing (n. sires)	95
Gene markers (n. data points)	>60000

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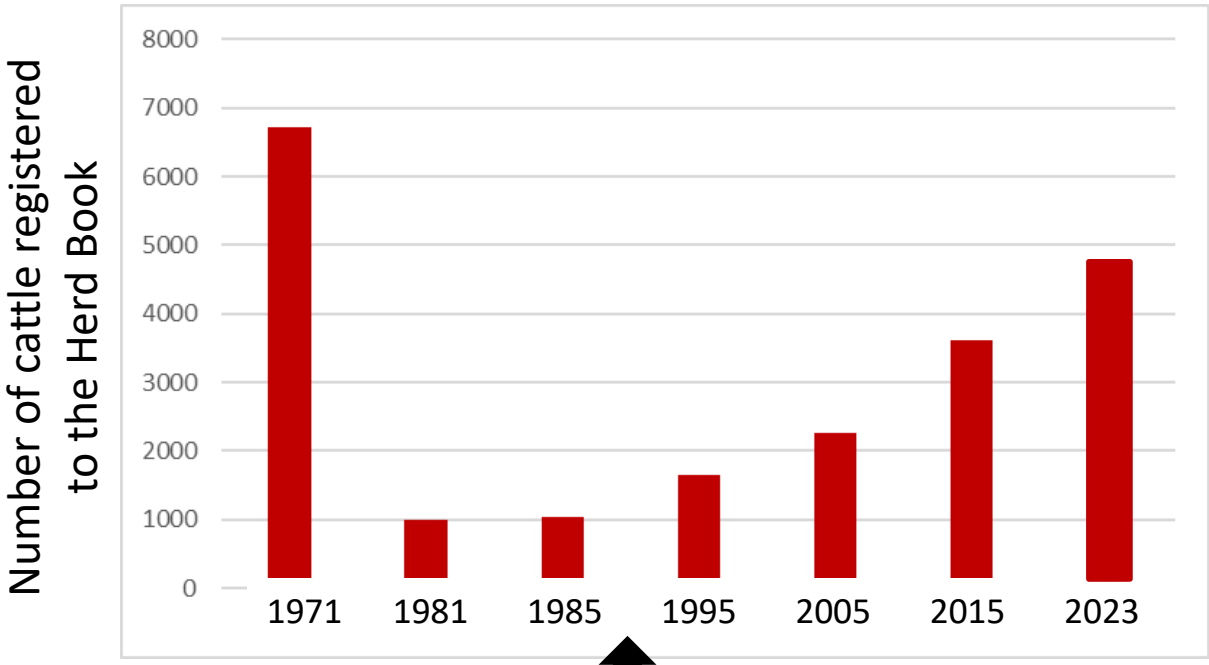
1 Cattle (I)

Reggiana

Parmigiano-Reggiano cheese area



1991

1 Cattle (I)

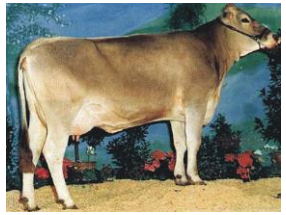
Reggiana

***MC1R e* allele**

Holstein

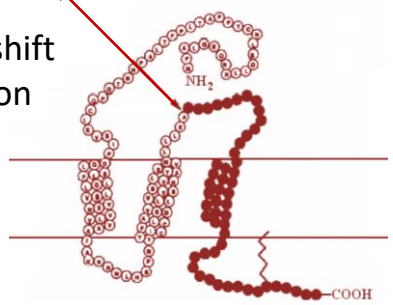
E^d

Brown



E^+

Frameshift mutation



1 Cattle (I)

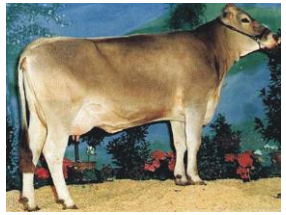
Reggiana

***MC1R e* allele**

Holstein

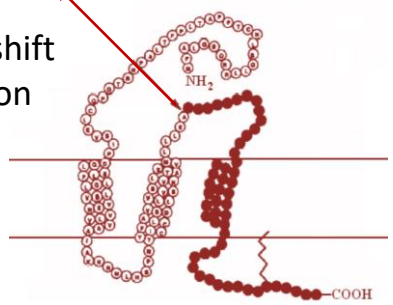
E^d

Brown

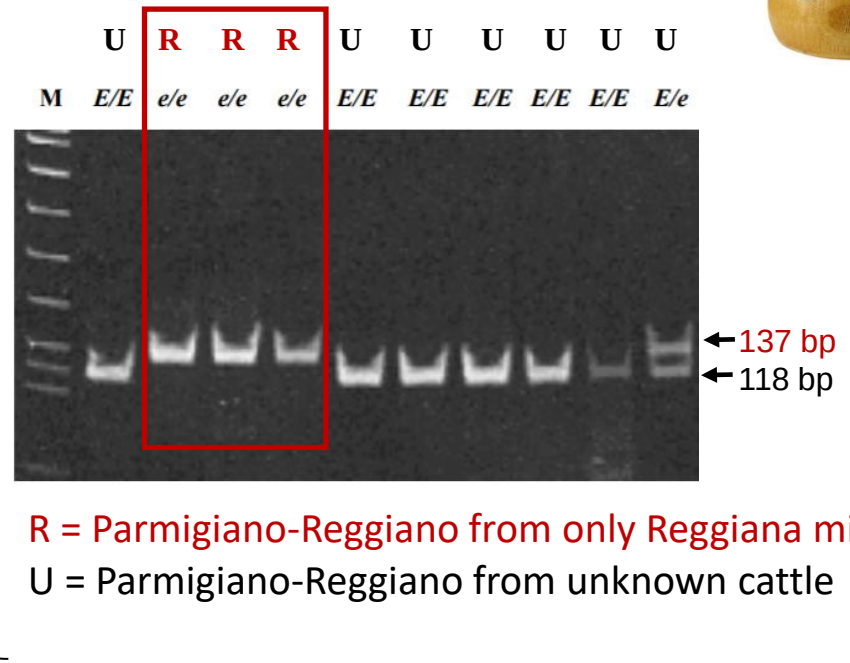


E^+

Frameshift mutation



MC1R alleles detected from cheese DNA



R = Parmigiano-Reggiano from only Reggiana milk

U = Parmigiano-Reggiano from unknown cattle

1 Cattle (I)

Reggiana

Herd Book: Fixation of the *MC1R e* allele

Breed product authentication

MC1R alleles detected from cheese DNA

R = Parmigiano-Reggiano from only Reggiana milk

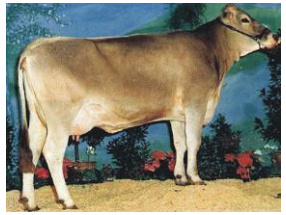
U = Parmigiano-Reggiano from unknown cattle

***MC1R e* allele**

Holstein

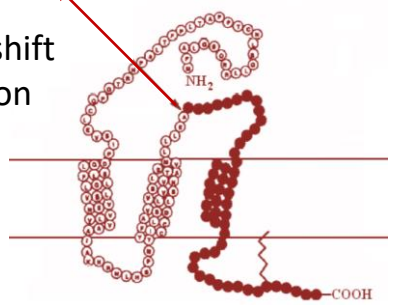
E^d

Brown



E^+

Frameshift mutation



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1 Cattle (III)

Phenotypic heterogeneity in Reggiana population

Coat colour

(a)

Dark red | Normal red | Light red

(c)

Piebaldism

Muzzle colour

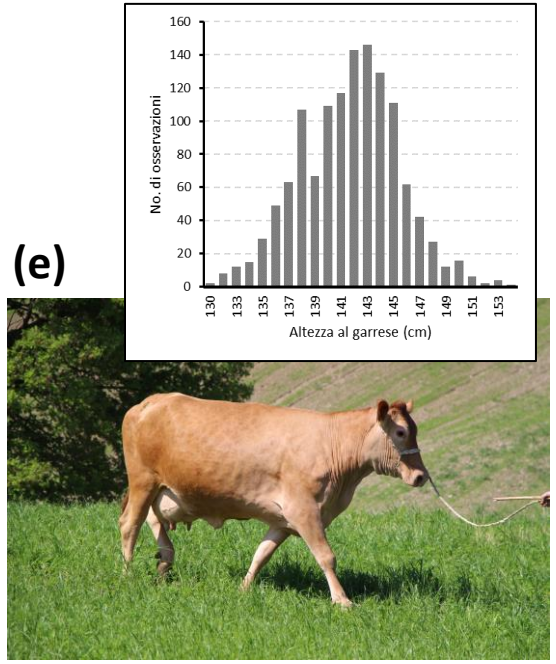
(b)

Pink muzzle | Gray muzzle | Black muzzle

(d)

Supernumerary teats

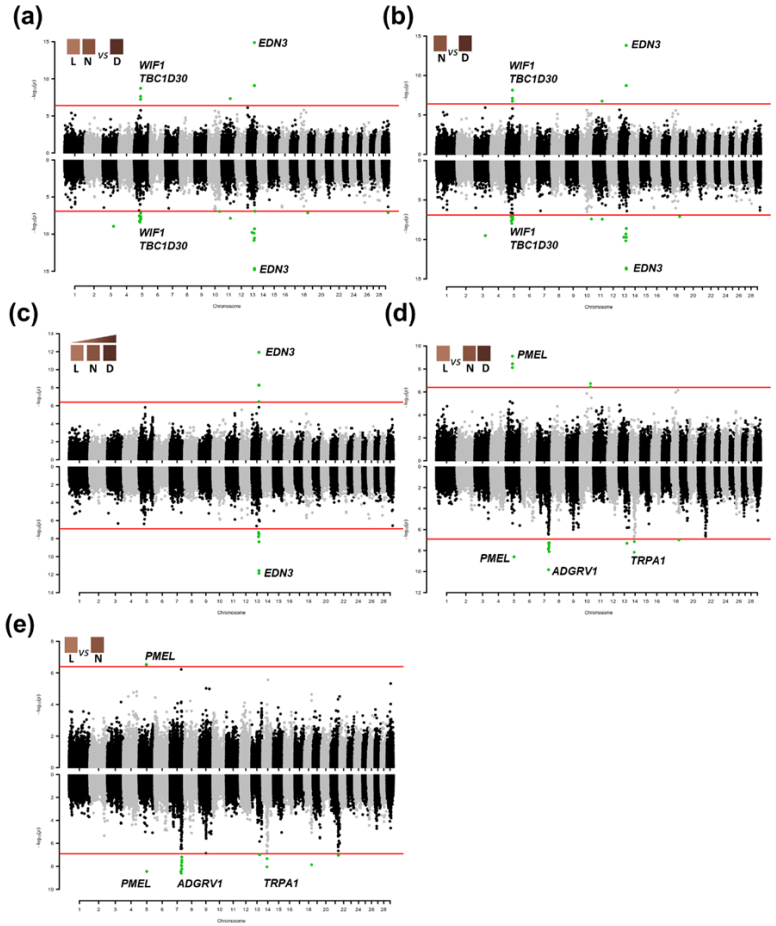
(e)



Stature

1 Cattle (III)

Phenotypic heterogeneity in Reggiana population



Summary of results

- *PMEL* (pale coat colour)
- *EDN3* (dark coat colour)
- *KIT* (piebaldism)
- *MC1R* (muzzle colour)
- *MCC*, *TBX3*, *TBX5*, *TLR4* (supernumerary teats)
- *NCAPG/LCORL* (stature)

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2 Pigs (I)


Mora Romagnola
Cinta Senese
Casertana
Sarda
Apulo Calabrese
Nero Siciliano

2 Pigs (I)

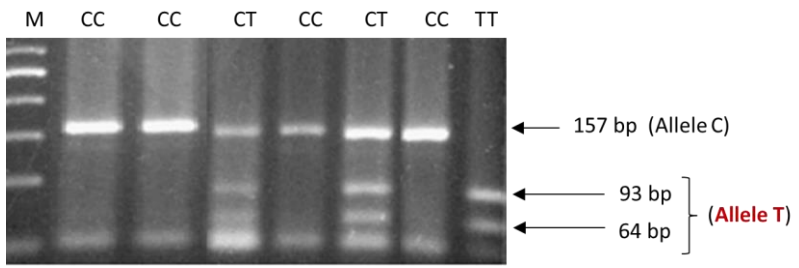
2 Pigs (I)

Cinta Senese

KIT

CCTCGCAGCAGGAGCAGTATCTACAGGAATATTTTGGAGCTTCAT
AATGAACATTGCTGACTCCCCTGTGCTTCCACTGCAG**GCTCGGCT**
ACCCGTGAAGTGGATGGCACC[C/T]GAGAGCATTTTCAACTGTG
TCTACACATTTGAAAGCGATGTCTGGTCCTATGGGATTTTTCTGT
GGGAGCTCTTCTCTTTAGGTAAAATGCACCTTGCCAAAGGCACCT
CAGTTAGACTCTGGGCATCTTCTTTAAGATGTTCCCATTGTCCTG
CTGGCTGCCTGTGACACTGATTGCAAACCCTTGTGCTCAG**GGGAG**
CAGCCCCTACCCCGGAATGCCAGTGATTCTAAATTCTACAAGATG
ATCAAGGAGGGTTTCCGAATGCTCAGCCCTGAG

Genotypes



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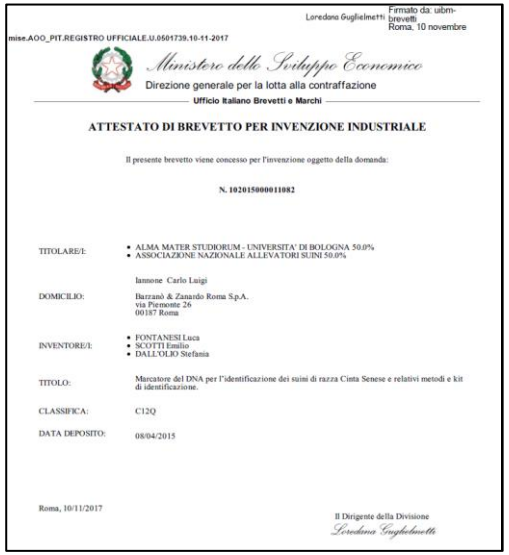
2 Pigs (I)

Breeds	No. of pigs	Genotypes (No. of pigs)			Allele frequencies		HWE	δ
		CC	CT	TT	C	T		
Cinta Senese	110	0	9	101	0.041	0.959	0.535	-
Italian Large White	105	105	0	0	1.000	0.000	-	0.959
Italian Landrace	52	52	0	0	1.000	0.000	-	0.959
Italian Duroc	86	79	6	1	0.953	0.047	0.145	0.912
Pietrain	32	32	0	0	1.000	0.000	-	0.959
Hampshire	16	0	4	12	0.125	0.875	0.449	0.084
Mora Romagnola	50	50	0	0	1.000	0.000	-	0.959
Casertana	47	43	4	0	0.957	0.043	0.673	0.916
Apulo-Calabrese	50	50	0	0	1.000	0.000	-	0.959
Nero Siciliano	42	37	5	0	0.940	0.060	0.574	0.899
Meishan	12	12	0	0	1.000	0.000	-	0.959
Wild boar	29	29	0	0	1.000	0.000	-	0.959

2 Pigs (I)

Patent owned by:



Ministero dello Sviluppo Economico
Direzione generale per la lotta alla contraffazione
Ufficio Italiano Brevetti e Marchi

ATTESTATO DI BREVETTO PER INVENZIONE INDUSTRIALE

Il presente brevetto viene concesso per l'invenzione oggetto della domanda:

N. 102015000011082

TITOLARE/I: • ALMA MATER STUDIORUM - UNIVERSITA' DI BOLOGNA 50.0%
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Barzanò & Zanardo Roma S.p.A.
via Piemonte 26
00187 Roma

INVENTORE/I: • FONTANESI Luca
• SCOTTI Emilio
• DALL'OLIO Stefania

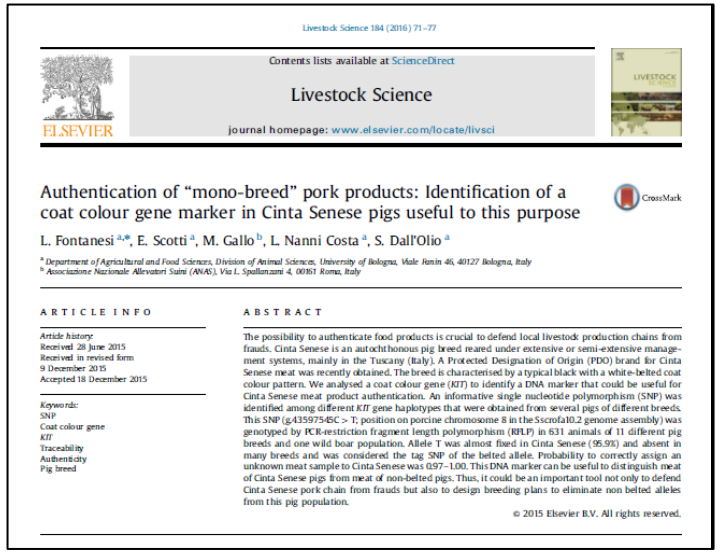
TITOLO: Marcatore del DNA per l'identificazione dei suini di razza Cinta Senese e relativi metodi e kit di identificazione.

CLASSIFICA: C12Q

DATA DEPOSITO: 08/04/2015

Roma, 10/11/2017

Il Dirigente della Divisione



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Authentication of "mono-breed" pork products: Identification of a coat colour gene marker in Cinta Senese pigs useful to this purpose

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ABSTRACT

The possibility to authenticate food products is crucial to defend local livestock production chains from frauds. Cinta Senese is an autochthonous pig breed reared under extensive or semi-extensive management systems, mainly in the Tuscany (Italy). A Protected Designation of Origin (PDO) brand for Cinta Senese meat was recently obtained. The breed is characterised by a typical black with a white-belted coat colour pattern. We analysed a coat colour gene (*KIT*) to identify a DNA marker that could be useful for Cinta Senese meat product authentication. An informative single nucleotide polymorphism (SNP) was identified among different *KIT* gene haplotypes that were obtained from several pigs of different breeds. This SNP (g.43597545C > T; position on porcine chromosome 8 in the Sscrofa10.2 genome assembly) was genotyped by PCR-restriction fragment length polymorphism (RFLP) in 631 animals of 11 different pig breeds and one wild boar population. Allele T was almost fixed in Cinta Senese (95.9%) and absent in many breeds and was considered the tag SNP of the belted allele. Probability to correctly assign an unknown meat sample to Cinta Senese was 0.97–1.00. This DNA marker can be useful to distinguish meat of Cinta Senese pigs from meat of non-belted pigs. Thus, it could be an important tool not only to defend Cinta Senese pork chain from frauds but also to design breeding plans to eliminate non belted alleles from this pig population.

Cinta Senese Herd Book:
Fixation of the *KIT T* allele

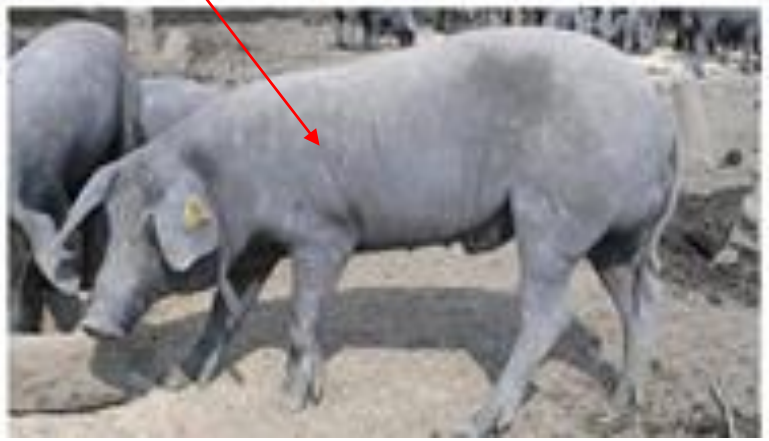
→ **Product authentication**

2 Pigs (II)

2 Pigs (II)

Phenotypic heterogeneity in Casertana population

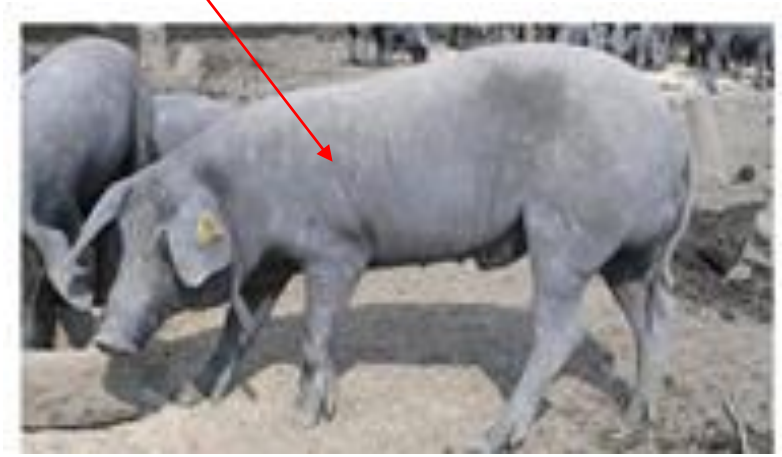
Hairless



Hairy

2 Pigs (II) Phenotypic heterogeneity in Casertana population

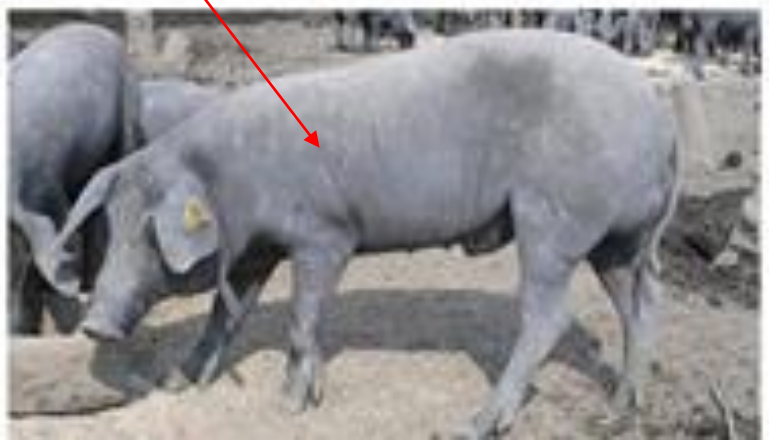
Hairless



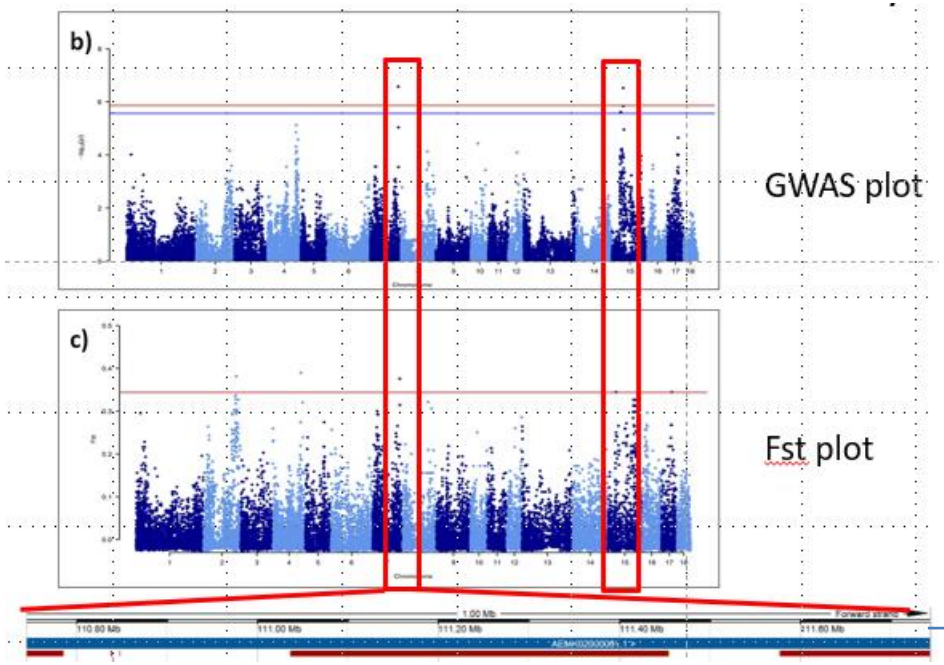
Hairy

2 Pigs (II) Phenotypic heterogeneity in Casertana population

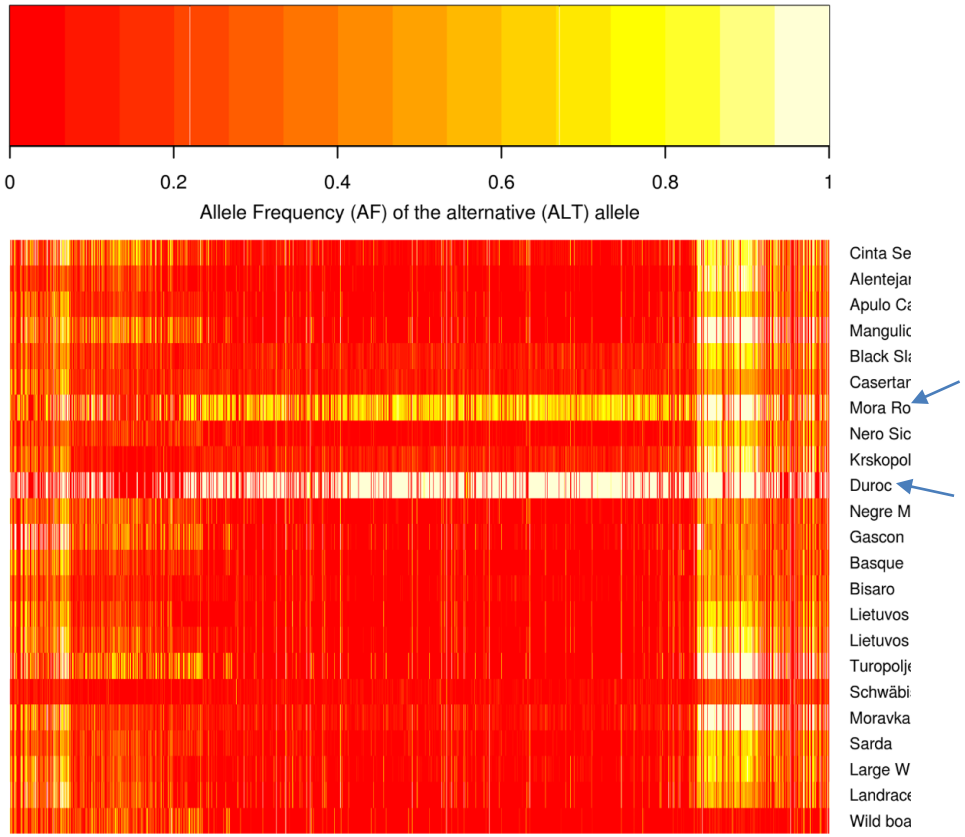
Hairless



Hairy



2 Pigs (III) Whole genome sequencing (WGS) data



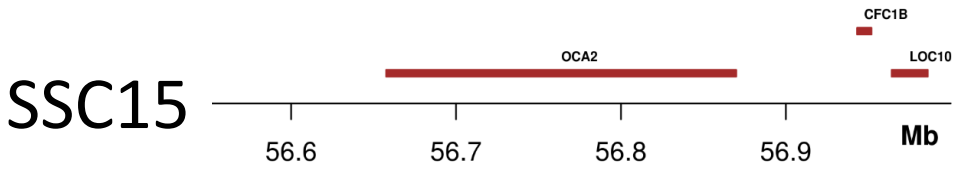
Red breeds

vs

all other breeds

Mora Romagnola

Italian Duroc

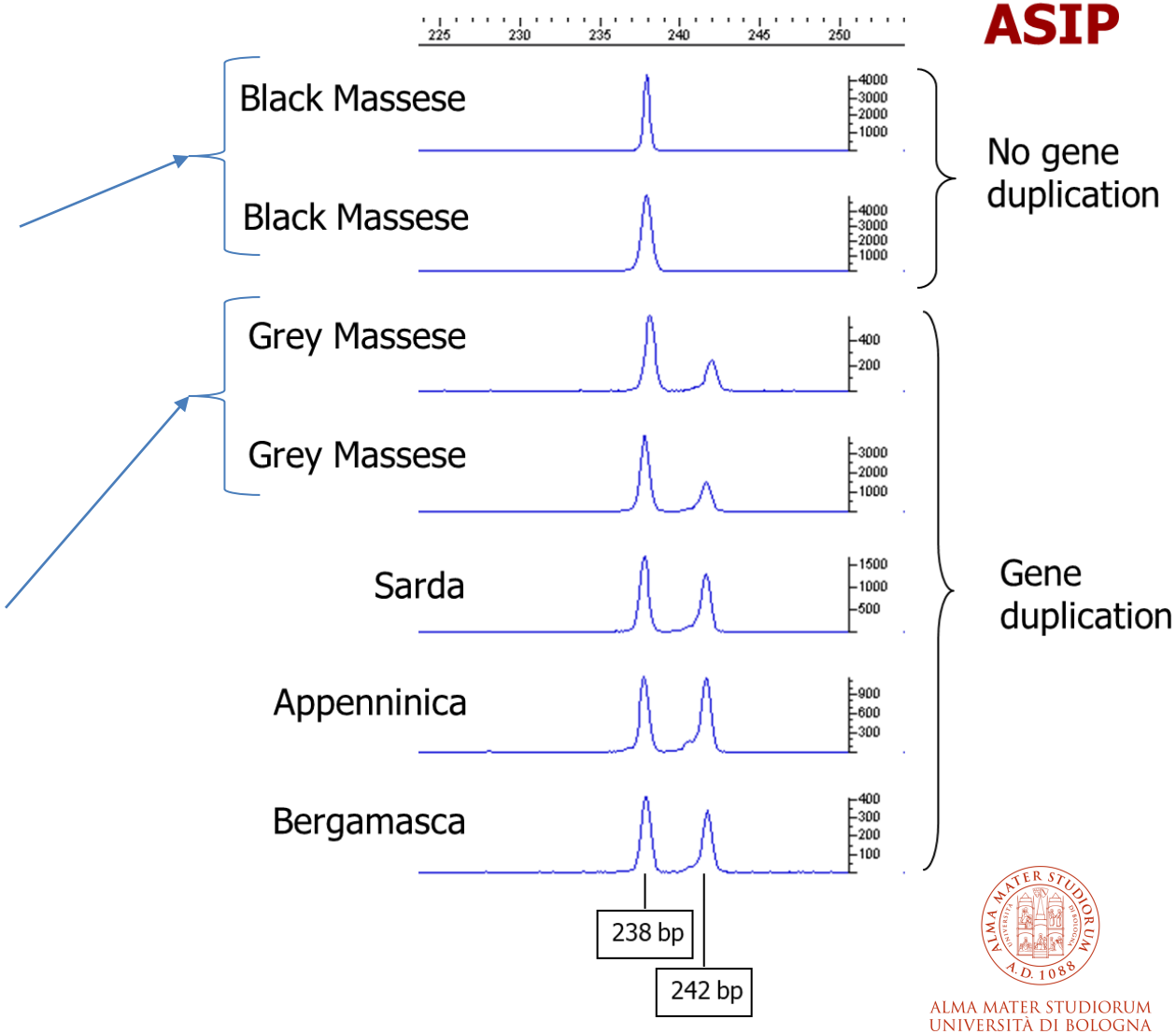


3 Sheep

Phenotypic heterogeneity in Massese population

3 Sheep

Phenotypic heterogeneity in Massese population



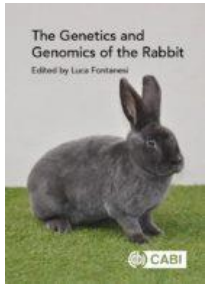
5 Rabbit

HEAVY

MEDIUM

LIGHTWEIGHT

SPECIAL HAIR



"CUN-FU-2"
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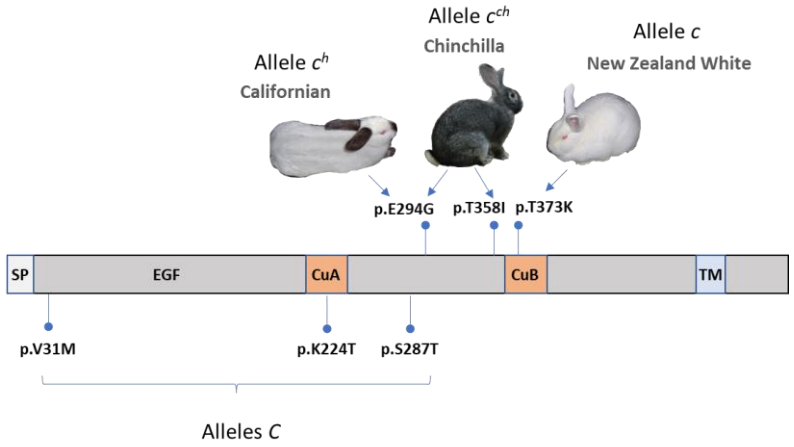
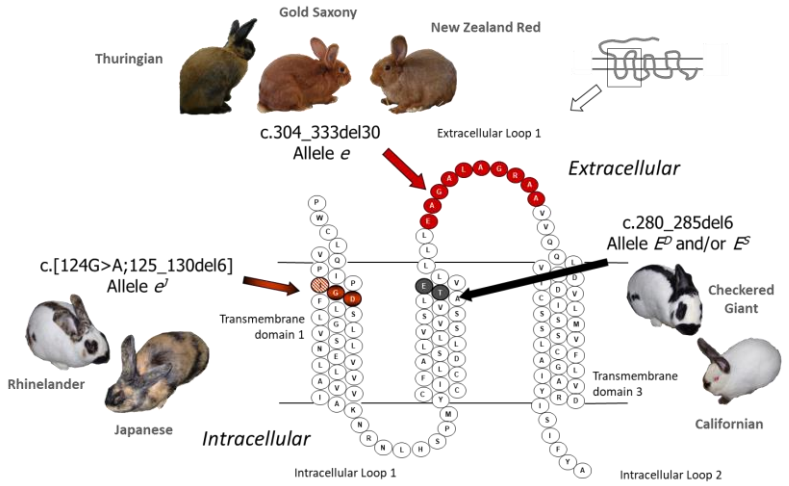
5 Rabbit

Albino locus (*TYR*)

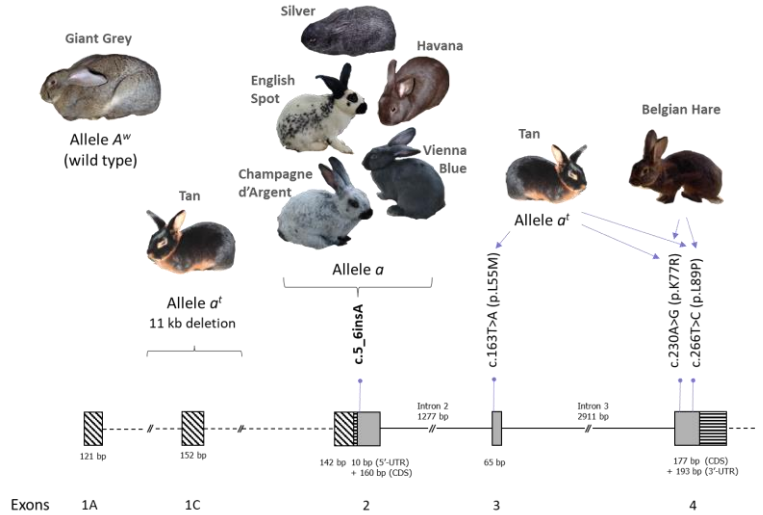


5 Rabbit

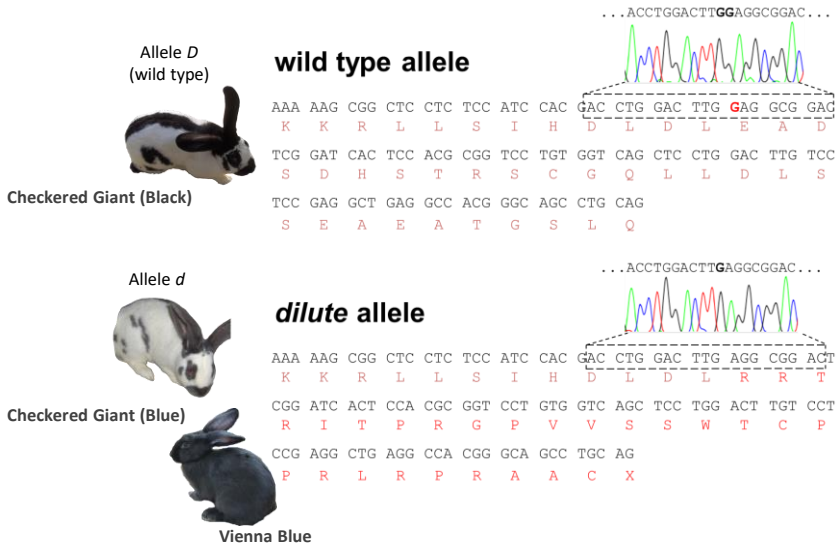
Extension locus (*MC1R*)



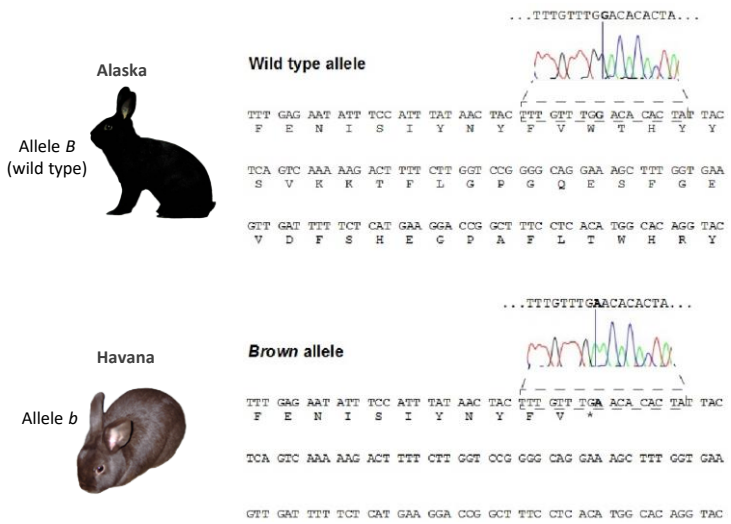
Agouti locus (*ASIP*)



Dilute locus (*MLPH*)

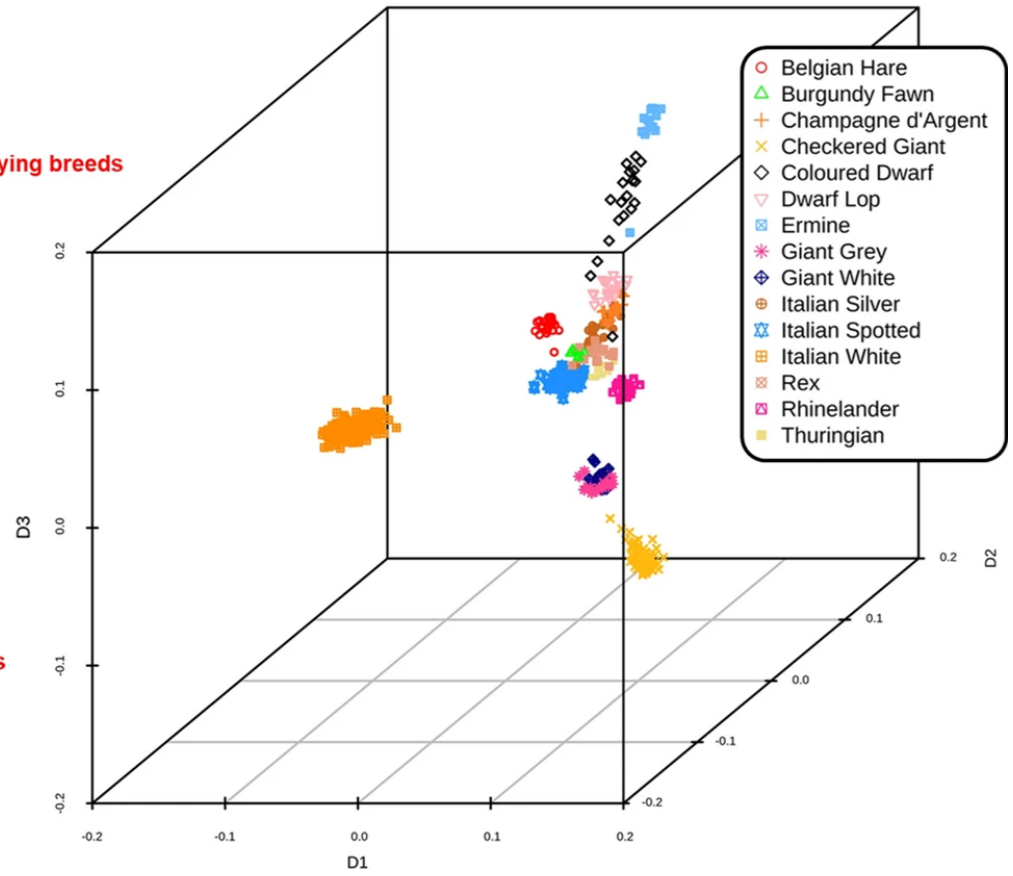


Brown locus (*TYRP1*)



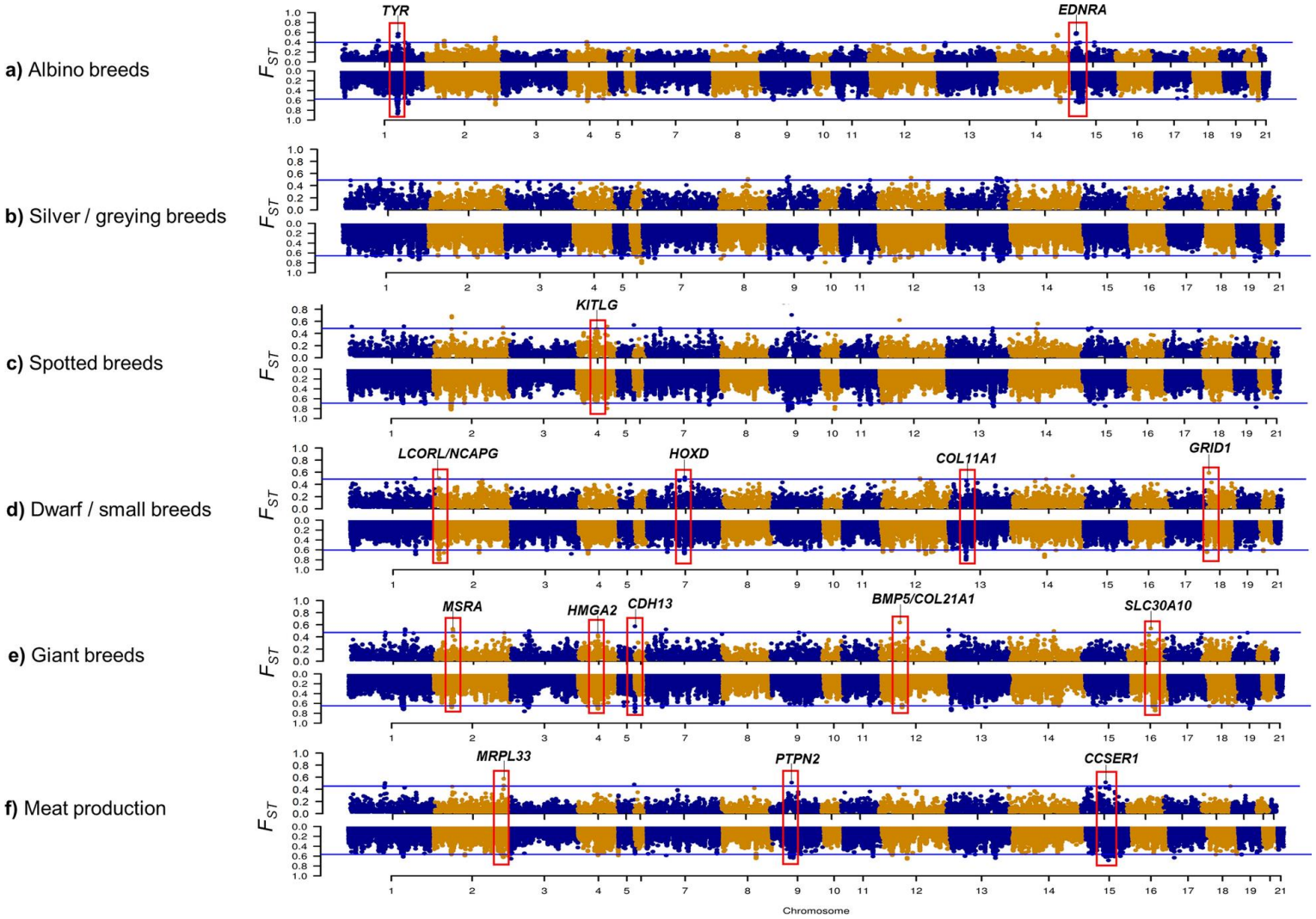
5 Rabbit

Population genomic analyses using SNP chip data

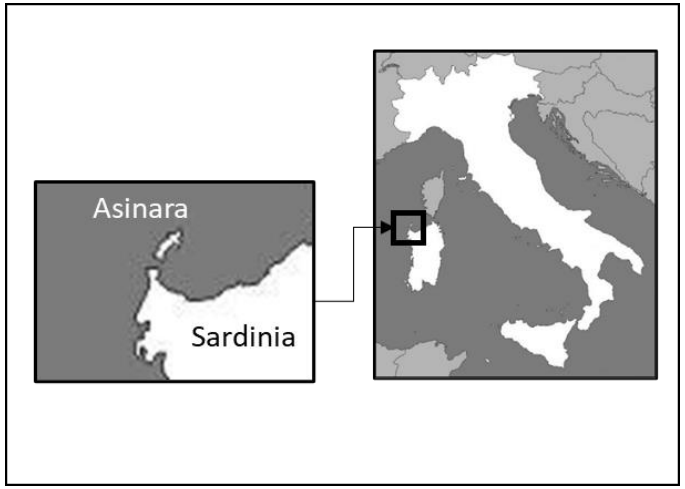


5 Rabbit

Population genomic analyses using SNP chip data



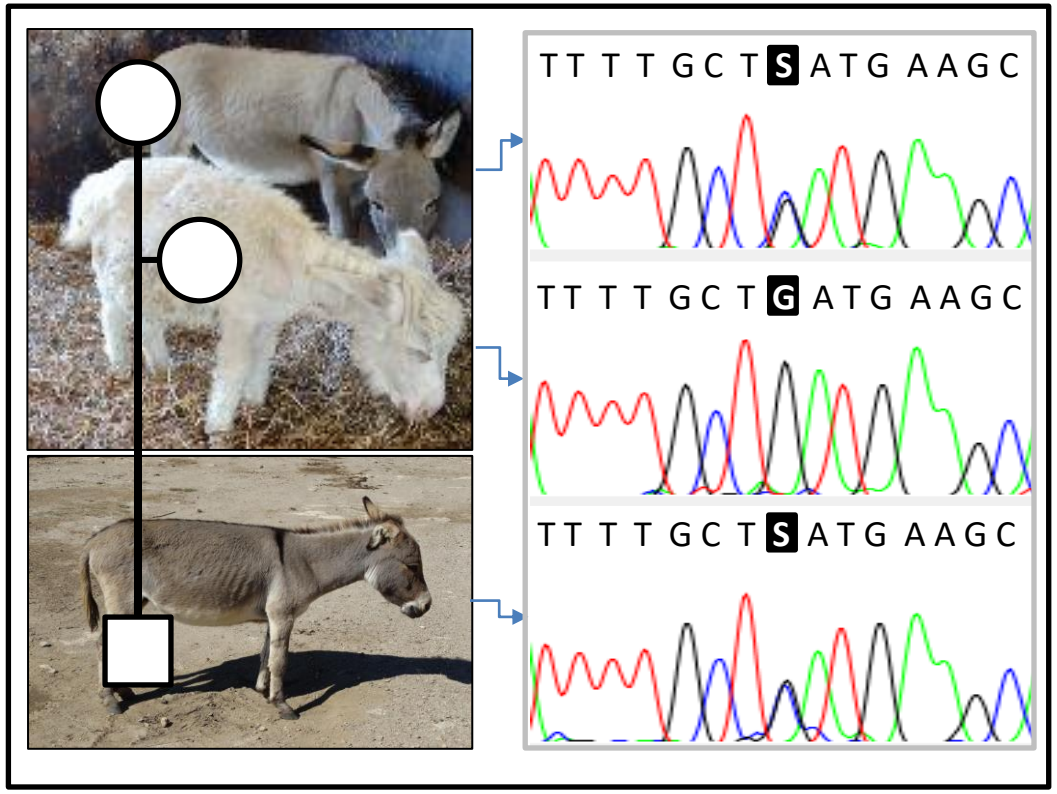
7 Donkey



Albinism

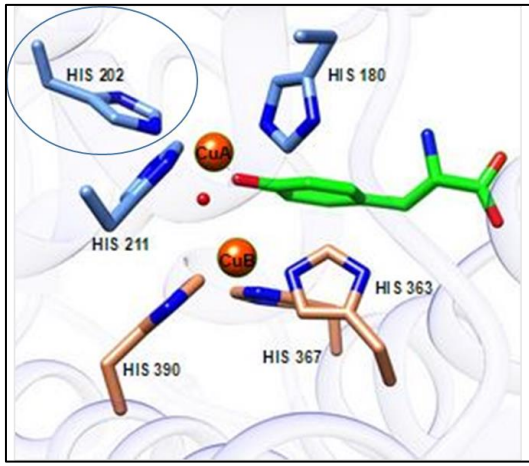
7 Donkey

Mutation in the tyrosinase (*TYR*) gene

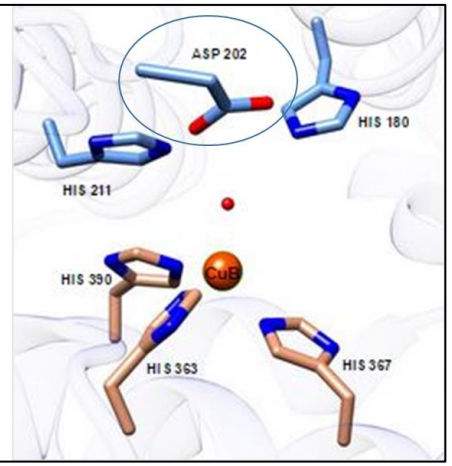


		H2A	H3A	
Equus asinus wild type	193	EVWKNIDFAHEAPGFLPWHRVFLLLWEQEIQK		228
***Equus asinus* albino**	193	.....D.....		228
Equus caballus	193	.....S.....		228
Homo sapiens	193	.I.RD.....A.....L...R.....		224
Mus musculus	193	.I.RD.....L.....RE		224
Bos taurus	193	...RD.....L.....		224
Oryctolagus cuniculus	193	...RD.....L.....		224
Gallus gallus	193	N..RD.....A.....R....		224
Xenopus laevis	197	AL.RD.....A.V....Y...H..H....		228
Danio rerio	193	N..AD.....SAA.....Y..F..H..R.		225
Neurospora crassa	88	SSGFGGYCT.SSIL.IT...PY.A.Y..ALYA		119
Ipomoea batatas	189	DYPDKEIQV.NSWL.F.F..WY.YFY.RILG.		220
Bacillus megaterium	51	PPGSDRNA..MSSA.....EY..RF.RDL.S		81

Wild type



Albino



Conclusions

- We have conducted throrough research on numerous Italian animal genetic resources

- By mining their genomes, we extracted a wealth of information and unique features that can **(i)** inform conservation programs and **(ii)** shed light on basic biological mechanisms

Animal and Food Genomics Group

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Associate Professor

Stefania Dall'Olio
Associate Professor

Paolo Zambonelli
Associate Professor

Samuele Bovo
Junior Assistant Professor
Bioinformatics – data analysis

Anisa Ribani
Junior Assistant Professor
Molecular genetics

Giuseppina Schiavo
Junior Assistant Professor
Bioinformatics – data analysis

Jacopo Vegni
Post-Doc
Data analysis

Valeria Taurisano
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Molecular genetics

Matteo Bolner
PhD student
Data analysis

Acknowledgements

"CUN-FU-2"

Co-funded by the European Union

Re-Livestock
RESILIENT FARMING SYSTEMS

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Animal and Food Genomics Group

Mining the genome of Italian animal genetic resources provided hints on peculiar genetic features: Examples of successful stories

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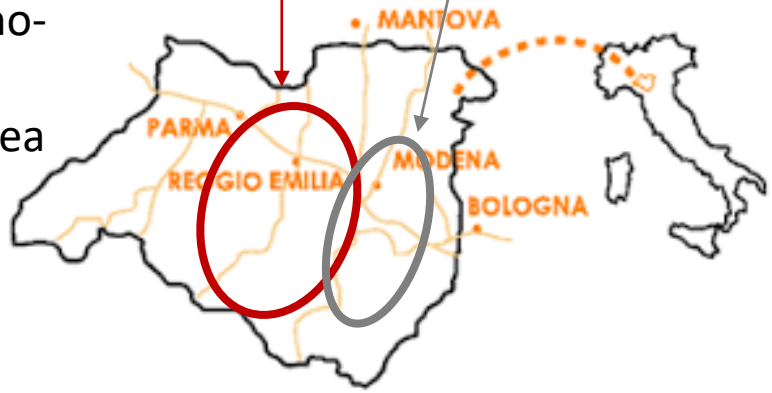
ERFP 2024 meeting - Florence, 29 August

1 Cattle (II)

Reggiana

Modenese (Bianca Val Padana)

Parmigiano-Reggiano cheese area



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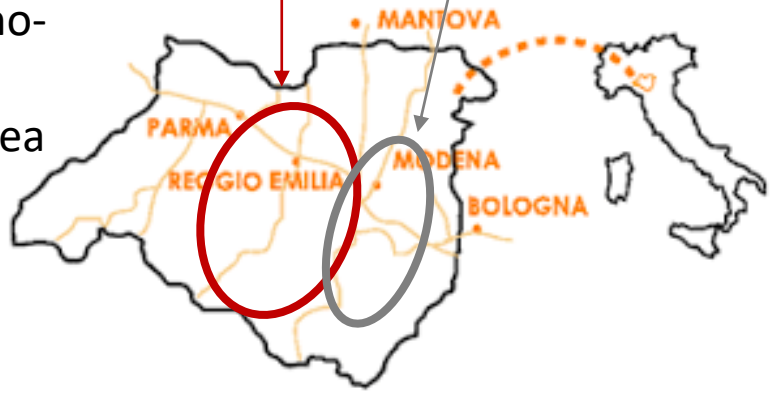
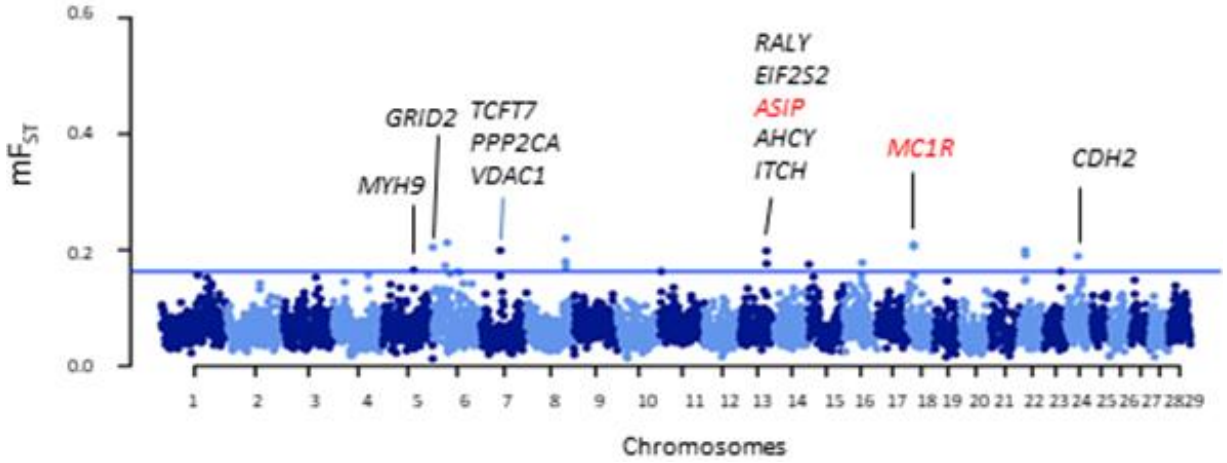




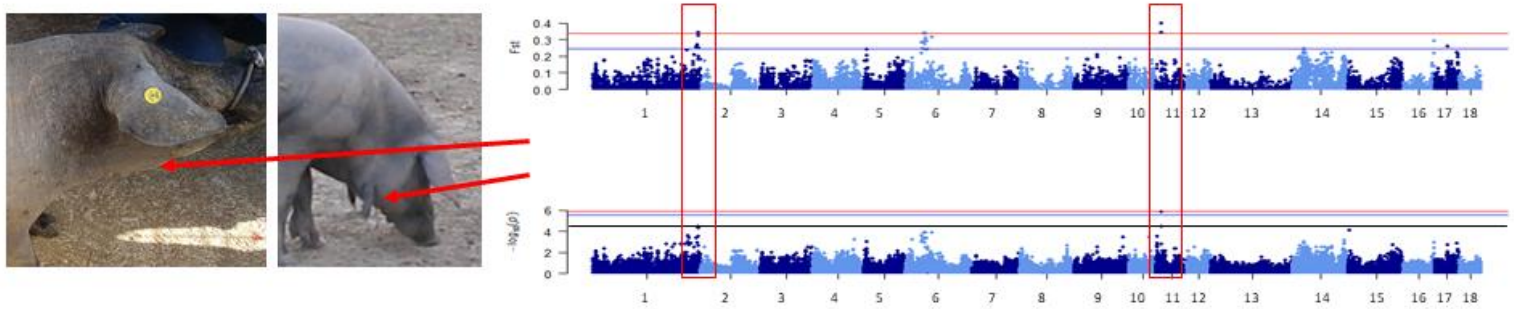
***MC1R e* allele**

***MC1R E*$^{+}$ allele**

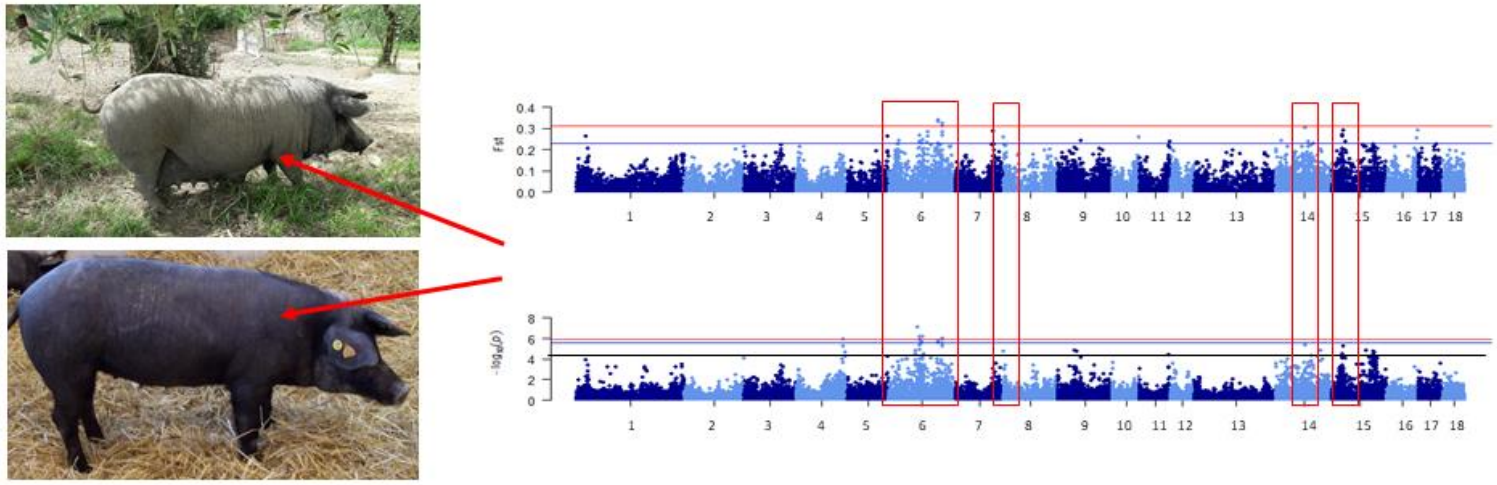
2 Pigs (II)

Phenotypic heterogeneity in Casertana population

Presence/absence of wattles



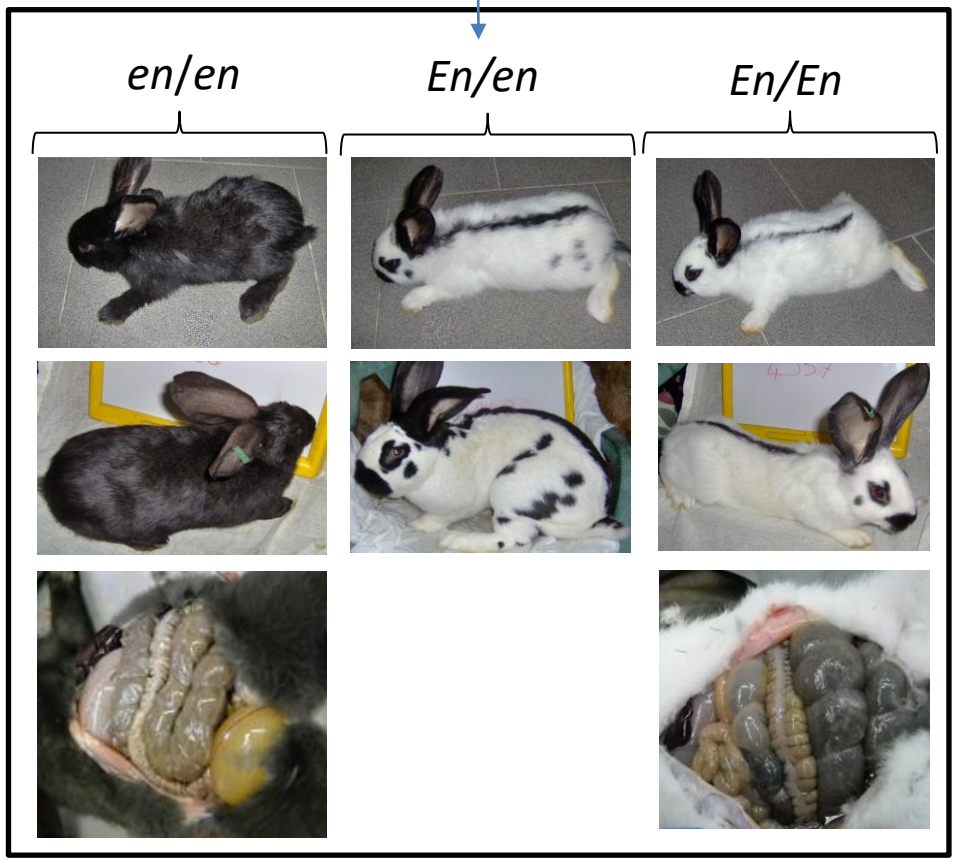
Slate-grey vs black coat colour



5 Rabbit

Spotted locus (*KIT*)

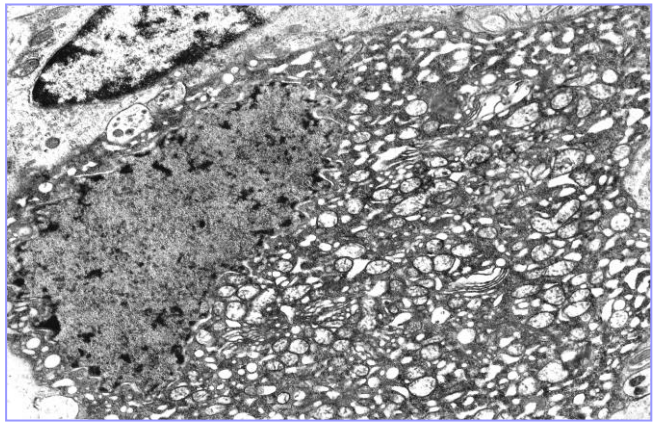
Checkered Giant rabbits



Megacolon

Ultrathin sections of colon specimens fixed for TEM

En/En



Particular of degenerated neuron