



ALMA MATER STUDIORUM
UNIVERSITÀ DI BOLOGNA



Animal and Food Genomics Group

A genomic-based approach to redefine herd book information in two local cattle breeds

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Reggiana and Modenese cattle breeds

Autochthonous cattle breeds from Emilia Romagna region, in the North of Italy

Historical traces go back to XII century



Reggiana



Modenese



**Unique mono-breed branded
Parmigiano-Reggiano cheese**

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Reggiana

- At present about 4500 cows raised in about 100 farms
- red coat color, “fromentino”, pink or pale muzzle



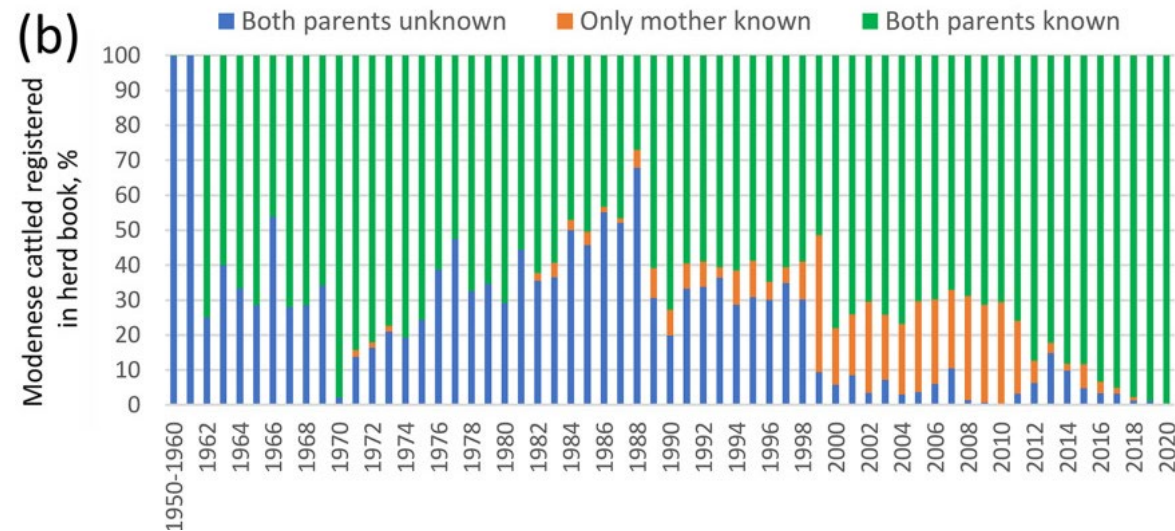
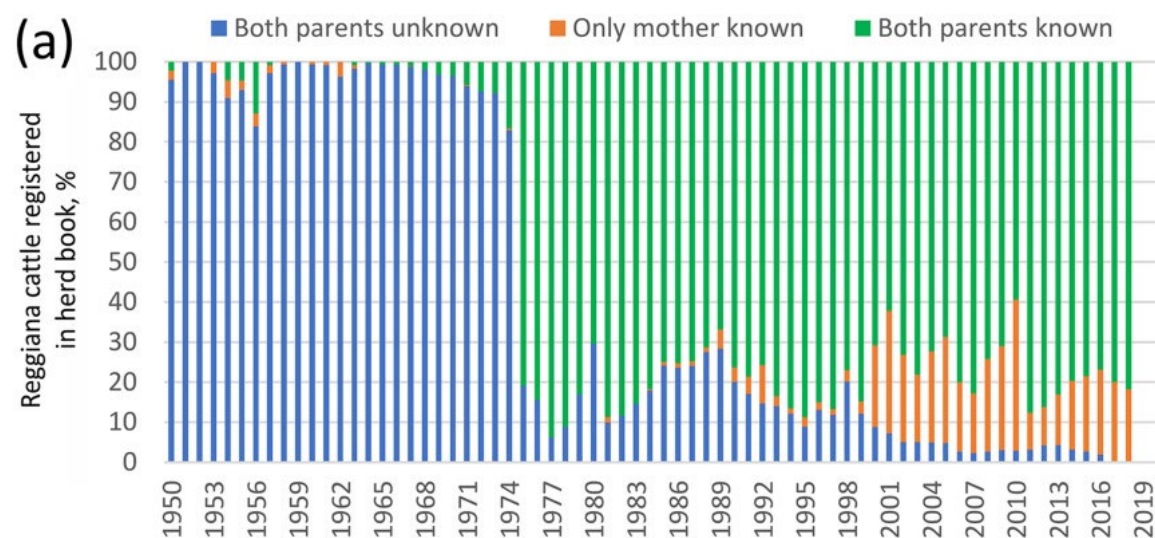
Modenese

- About 1200 cows
- White / pale grey color

Unique mono-breed branded Parmigiano-Reggiano cheese

Reggiana and Modenese pedigree

As in many local breeds, pedigree records in Reggiana and Modenese herd books are not always complete.



AIM

Identify inconsistencies and reconstruct pedigree
of Reggiana and Modenese cattle breed

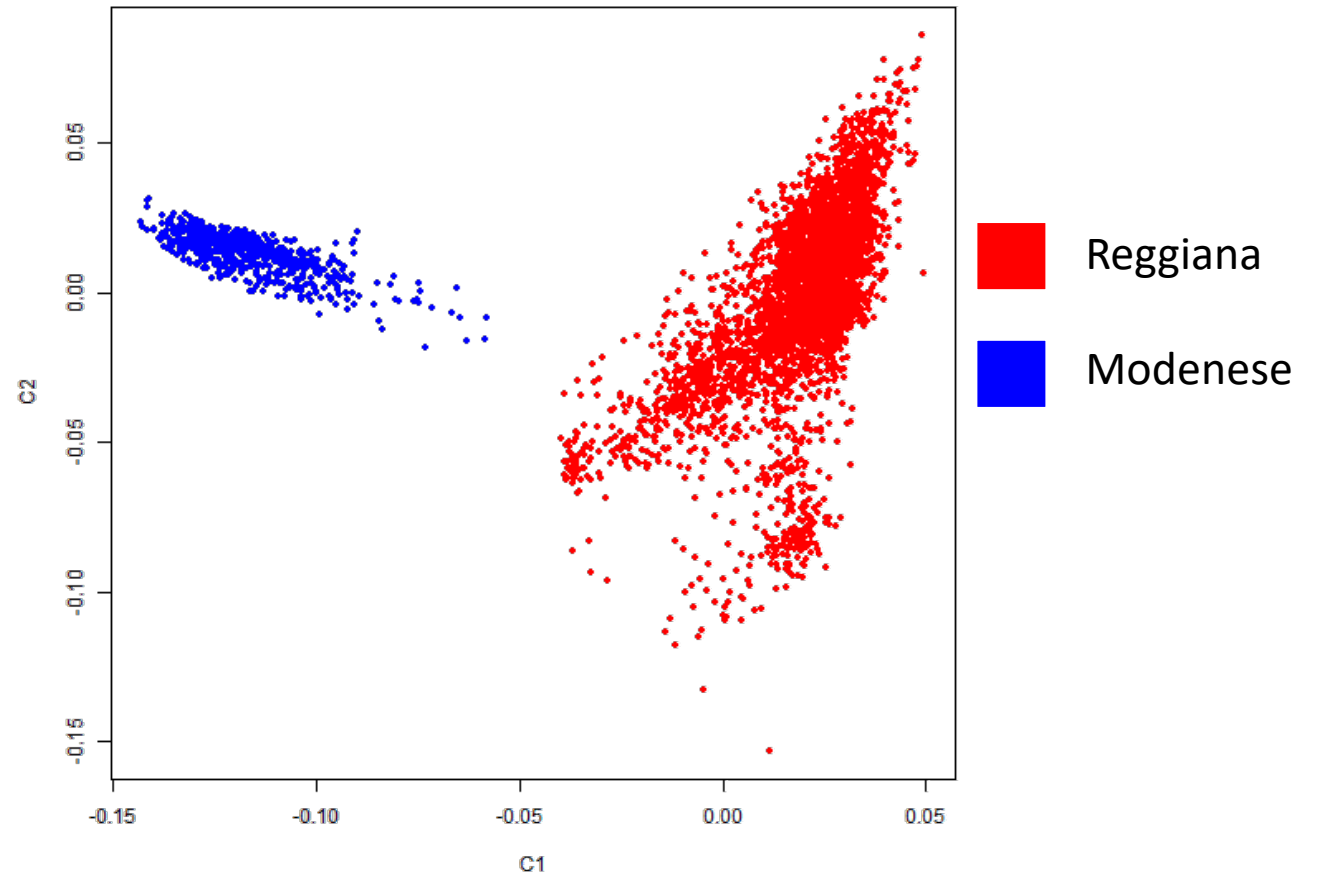
Materials and Methods: animals and genotyping

4025 Reggiana cattle,
including commonly used 288 sires
from 50 different farms

710 Modenese including 15 sires
from 20 different farms

GGP Bovine 150K SNP Chip
139464 autosomal SNPs remained after
PLINK filtering

In addition: 300 duplicates – evaluation of error rate



MDS of the individuals from both breeds

Materials and Methods: analysis

Combination of several approaches to estimate relatedness using DNA markers:

- 1) **Identity by Descent (IBD)** – *PLINK* software, *KING* software
- 2) **Pairwise comparisons based on Opposing Homozygotes (OH)**
 - Define inconsistencies in parent-offspring pairs
 - Define inconsistencies between pairs of duplicates to estimate error rate
 - Evaluate mapping problems and/or structural variants in the individuals
- 3) **Comparison with genomic inbreeding** and pedigree based inbreeding after the corrections
- 4) **Correction/Reconstruction** of the pedigree records

**Relationship
estimation**

**Correction and
integration**



Results

Estimated degrees of relatedness and related statistics

Degree of relatedness	Mean IBD	Min / Max IBD	Mean Number of OH SNPs	Min / Max #OH SNPs
Unrelated	0	0 / 0	11002.3 $\pm$ 989.1	9916 / 12453
3rd	0.019 $\pm$ 0.03	0 / 0.05855	9266.9 $\pm$ 773.7	7444 / 11351
2nd	0.072 $\pm$ 0.05	0.0392 / 0.7145	7095.3 $\pm$ 964.6	4598 / 8596
Full Siblings	0.132 $\pm$ 0.05	0.1242 / 0.14105	2104.5 $\pm$ 405.2	1818 / 2391
Parent-Offspring	0.259 $\pm$ 0.02	min=0.24365,max=0.3126	35.4 $\pm$ 10.4	min=11, max=155

Call rate evaluation:

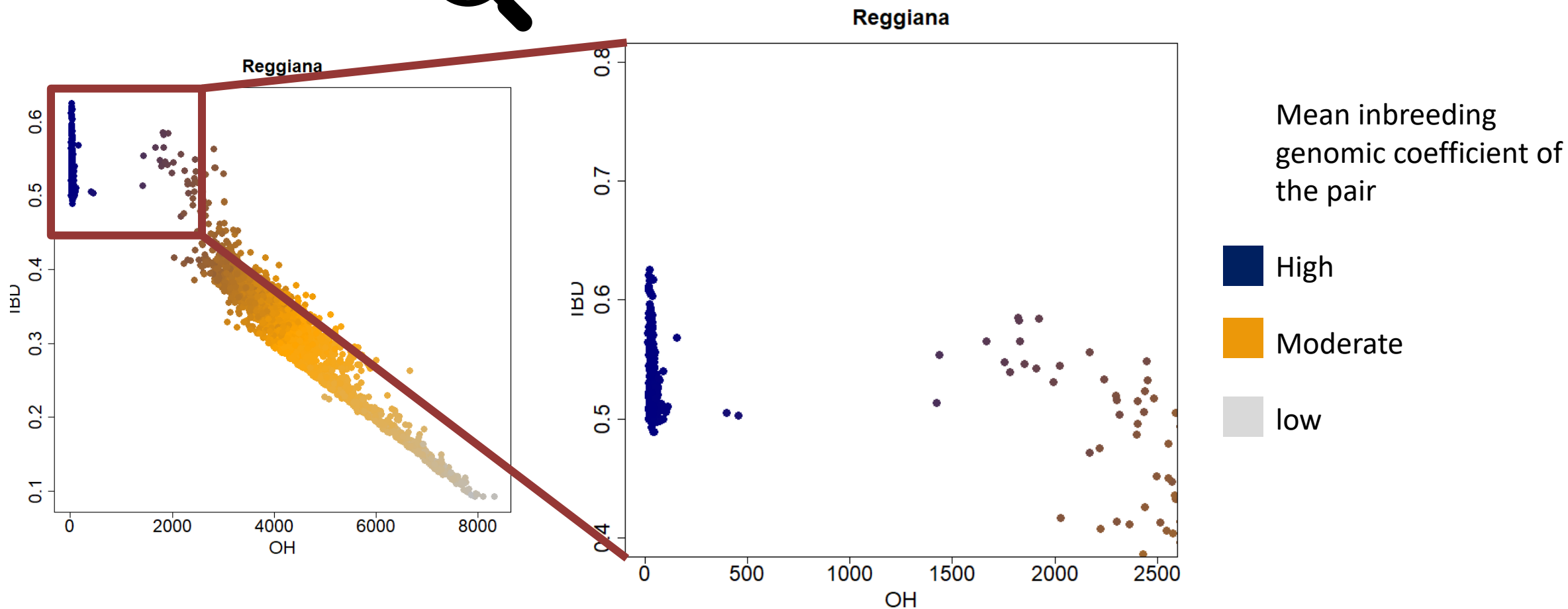
for duplicate samples, OH SNPs ranged from 0 to a maximum of 9 with an average of 4

Combination of IBD estimation and OH to validate the Parent-Offspring relationship

Some cases:

IBD $\geq$ 0.25 **but** OH > 1800

Results

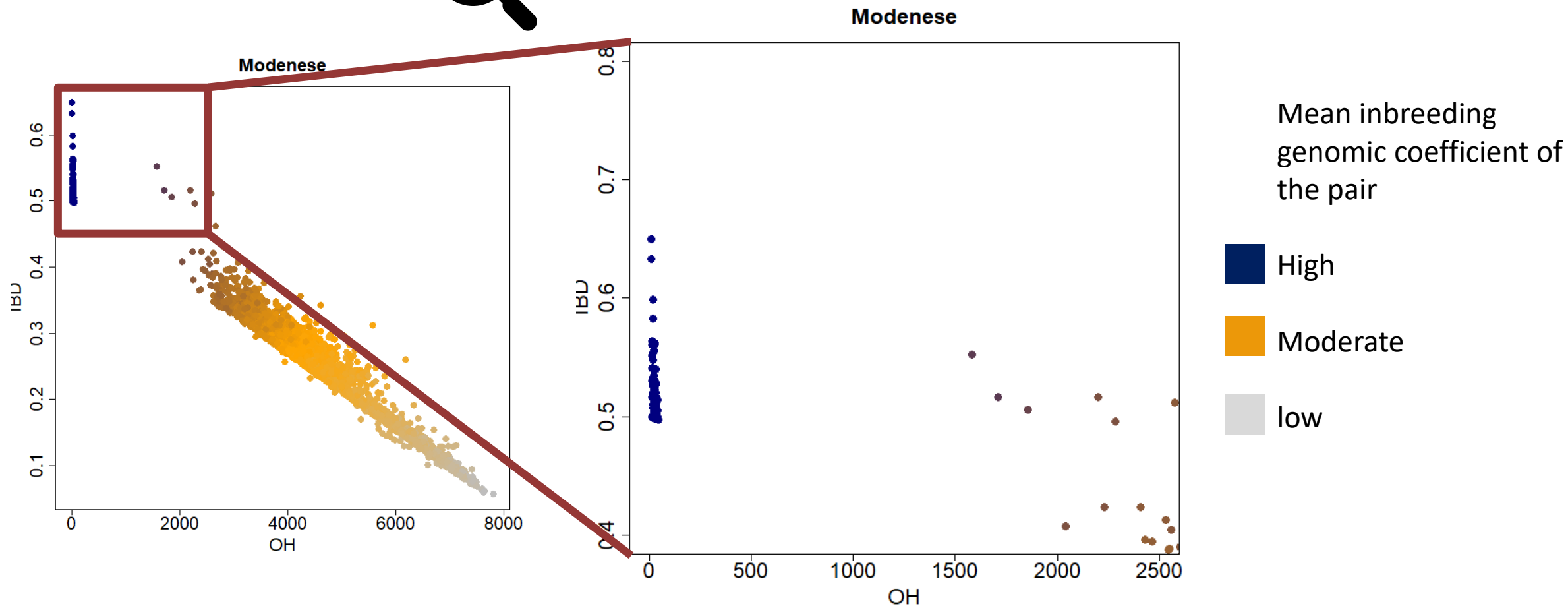


X-axes: number of Opposite Homozygous SNPs.
Y axes: PLINK – IBD

**False Parent-Offspring
estimation if only looking at
Plink - IBD**



Results

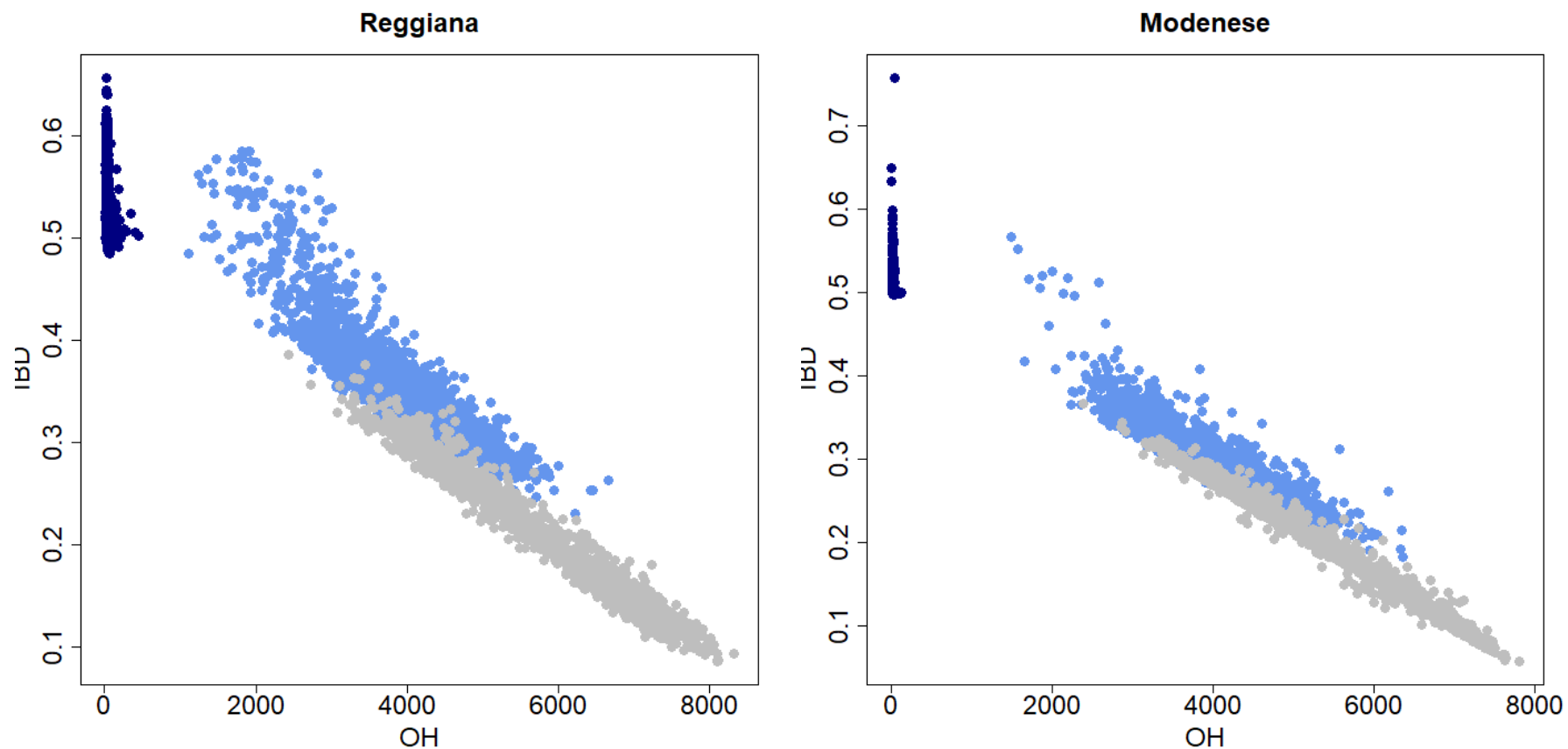


X-axes: number of Opposite Homozygous SNPs.
Y axes: PLINK – IBD

**False Parent-Offspring
estimation if only looking at
Plink - IBD**



Results



Relationships
inferred by KING
software

- Parent – Offspring
- Full-siblings
- 2° degree

X-axes: number of Opposite Homozygous SNPs.
Y axes: PLINK – IBD



Results

Parentage information from pedigree records was not correct for about 10% of the comparisons.

	Fathers					
	Correct	Wrong	Swap	Reconstructed	Not genotyped	Total
Mothers	Correct	928	131	112	13	1359
	Wrong	129	79	86	12	363
	Swap	144	94	125	9	434
	Reconstructed	1	1	2	11	46
	Not genotyped	796	248	176	76	1823
	Total	1998	553	501	121	4025

	Fathers					
	Correct	Wrong	Swap	Reconstructed	Not genotyped	Total
Mothers	Correct	8	0	0	179	187
	Wrong	2	0	1	40	43
	Swap	2	0	0	55	57
	Reconstructed	0	0	0	0	0
	Not genotyped	0	1	1	421	423
	Total	12	1	2	695	710



Different scenarios for pedigree correction and reconstruction



Results

Parentage information from pedigree records was not correct for about 10% of the comparisons.

		Fathers				
Mothers		Correct	Wrong	Swap	Reconstructed	Not genotyped
	Correct	928	131	112	13	175
	Wrong	129	79	86	12	57
	Swap	144	94	125	9	62
	Reconstructed	1	1	2	11	31
	Not genotyped	796	248	176	76	527
Total		1998	553	501	121	852
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Mothers		Correct	Wrong	Swap	Reconstructed	Not genotyped
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	Wrong	2	0	1	0	40
	Swap	2	0	0	0	55
	Reconstructed	0	0	0	0	0
	Not genotyped	0	1	1	0	421
Total		12	1	2	0	695
						710



Genomic relation Parent-Offspring matches with Pedigree information:



The records are validated and used as comparison for uncertain records



Results

Parentage information from pedigree records was not correct for about 10% of the comparisons.

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Incomplete pedigree records:



The records are updated with the correct parent information



Results

Parentage information from pedigree records was not correct for about 10% of the comparisons.

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	Correct	928	131	112	13	175
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	Reconstructed	0	0	0	0	0
	Not genotyped	0	1	1	0	421
	Total	12	1	2	0	695
		Total				
		710				



Wrong pedigree records:

Among the genotyped animals, none of them results in a Parent-Offspring relationship



The records are updated as missing



Results

Parentage information from pedigree records was not correct for about 10% of the comparisons.

		Fathers				
Mothers		Correct	Wrong	Swap	Reconstructed	Total
	Correct	928	131	112	13	175
	Wrong	129	79	86	12	57
	Swap	144	94	125	9	62
	Reconstructed	1	1	2	11	31
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						710



Reggiana – fathers : 0.17
Reggiana – mothers : 0.16

Proportion of wrong records
among the pairs of genotyped
animals



Modenese – fathers : 0.07 (few sires)
Modenese – mothers: 0.14



Results

Parentage information from pedigree records was not correct for about 10% of the comparisons.

	Correct	Not correct	Reconstructed	Not genotyped	Total
Correct	84	10	6	27	127
Not correct	10	2	1	7	20
Reconstructed	6	0	5	2	13
Not genotyped	41	17	9	36	103
Total	141	29	21	72	263

Some examples
divided by farm

	Correct	Not correct	Reconstructed	Not genotyped	Total
Correct	9	1	1	4	15
Not correct	3	8	11	4	26
Reconstructed	4	4	11	4	23
Not genotyped	27	20	12	20	79
Total	43	33	35	32	143



Results

Different situations for fathers' and mothers' records depending on the farm and on their farming system

Correct fathers proportion	Farm	Total genotyped
1	Farm1	13
0.93	Farm2	91
0.902	Farm3	51
0.891	Farm4	50
0.857	Farm5	18
0.844	Farm6	171
0.842	Farm7	22
0.833	Farm8	16
0.828	Farm9	29
0.825	Farm10	103
0.8	Farm11	74
0.8	Farm12	28
0.765	Farm13	75
0.757	Farm14	39

Correct mothers proportion	Farm	Total genotyped
1	Farm7	22
1	Farm33	8
0.923	Farm30	62
0.91	Farm6	171
0.889	Farm25	17
0.857	Farm10	103
0.85	Farm13	75
0.833	Farm12	28
0.788	Farm17	263
0.758	Farm2	91
0.75	Farm9	29
0.741	Farm35	72
0.739	Farm16	29
0.72	Farm18	66

Correct mothers proportion	Farm	Total genotyped
1	Farm14	11
1	Farm24	2
1	Farm11	1
1	Farm21	1
0.866667	Farm9	27
0.75	Farm19	12
0.722222	Farm2	28
0.6875	Farm6	19
0.666667	Farm4	370
0.666667	Farm20	3
0.666667	Farm22	3
0.655172	Farm1	32
0.5	Farm12	26



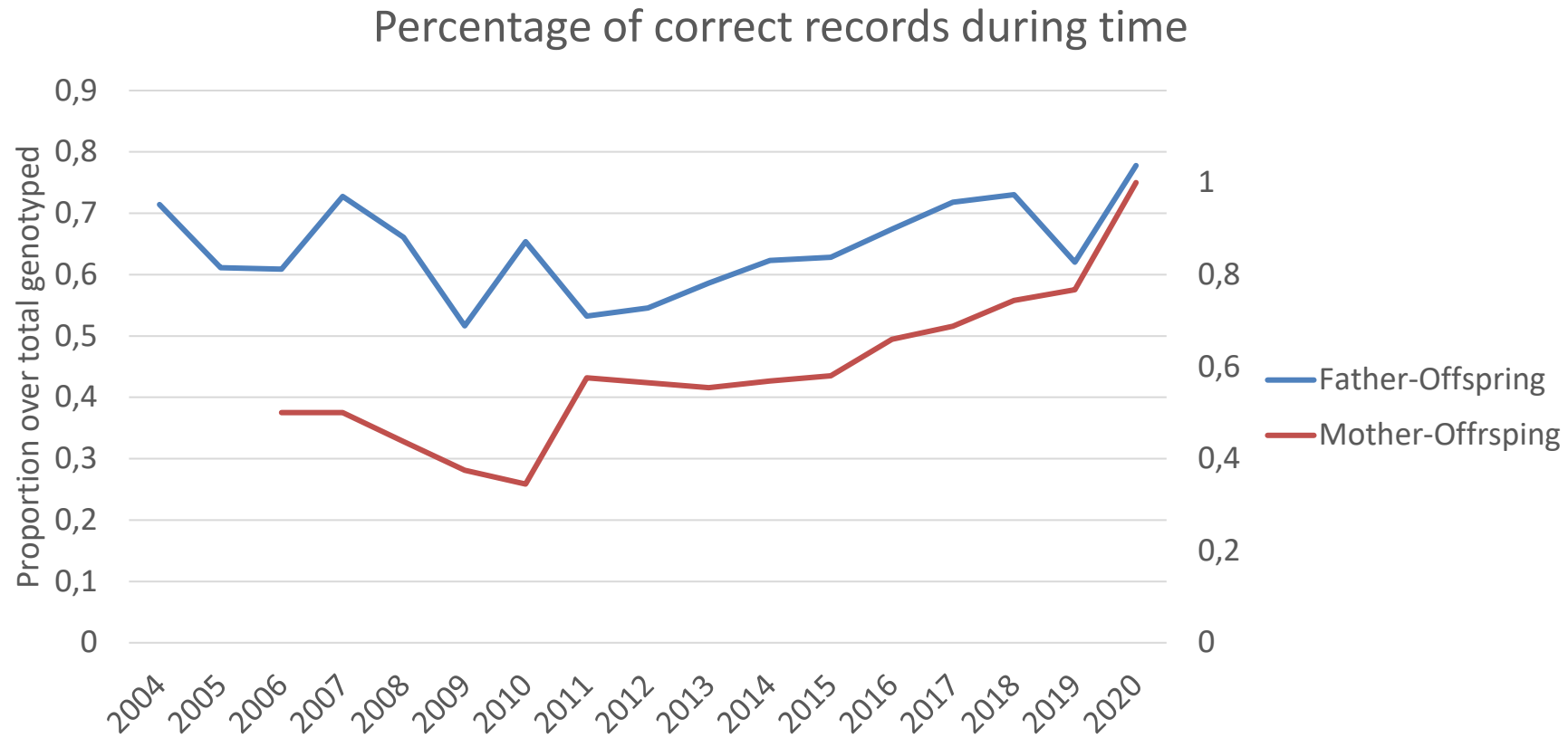
Top farms with the highest proportion of correct pedigree

For Modenese:
13 corrected/15
total sires



Reconstruction and correction

Correctness improves during time (Reggiana)



Information about farms were used to correct pedigree
(i.e. change of personnel, lactation and delivery records)

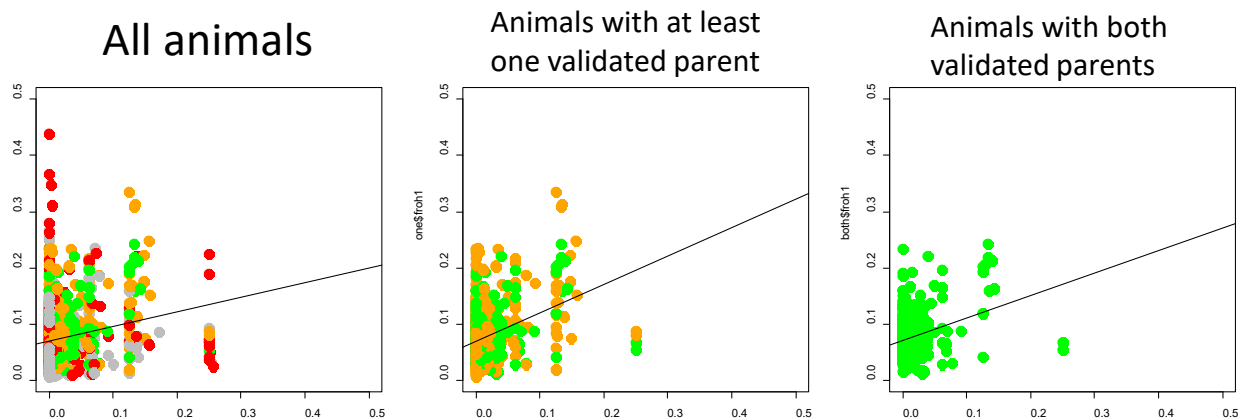


Recostruction and correction

After the corrections – **Proof of concept** :

Improvement of the correlations between pedigree based and genomic inbreeding

Y axes:
Pedigree based
inbreeding coefficient
before corrections
X axes: genomic
inbreeding (FROH)

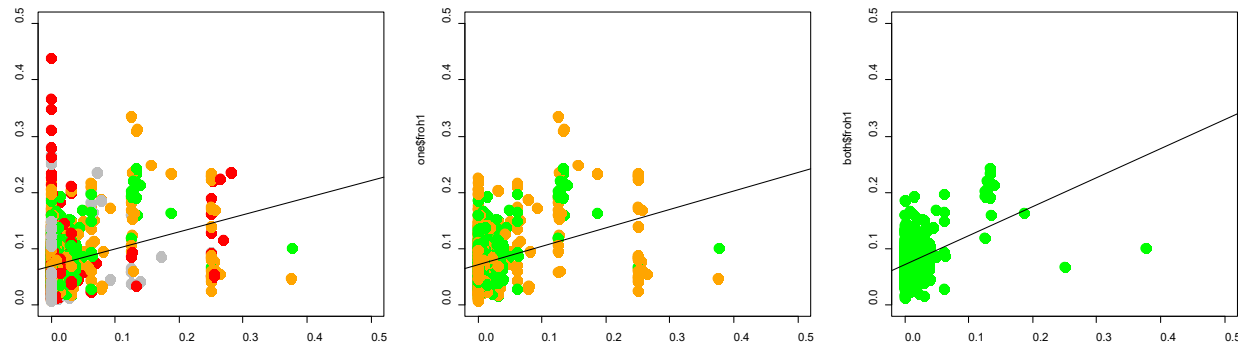


0.19

0.34

0.31

Y axes:
Pedigree based
inbreeding coefficient
after corrections
X axes: genomic
inbreeding (FROH)



0.27

0.34

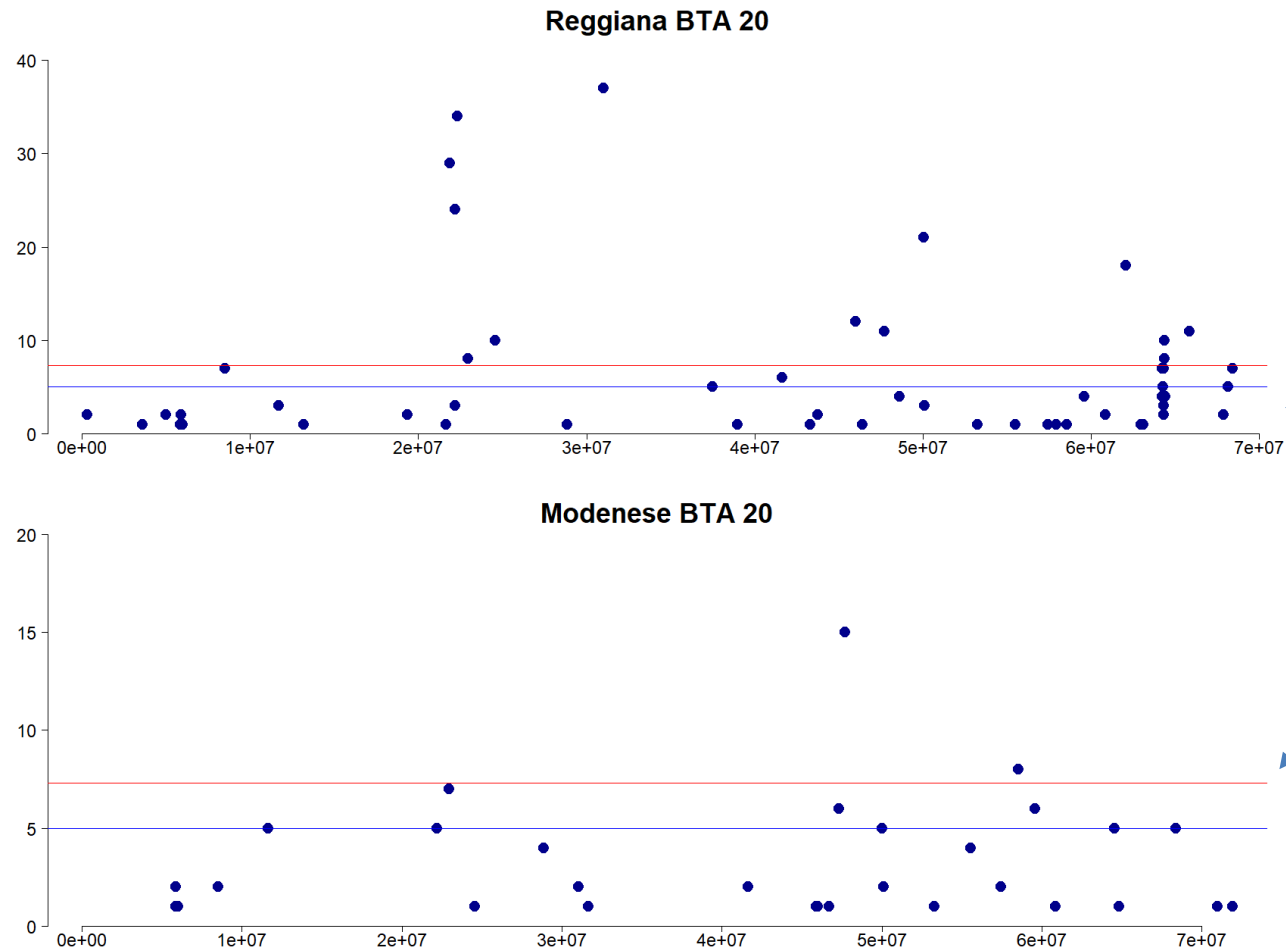
0.45

Pearson correlation coefficients

- Animal with wrong records
- Animal with one validated parent
- Animal with both validated parents
- Animals without genotyping information



Results – Regions of incompatibility



- Incompatible opposite SNPs shared by all animals in both breeds may indicate mapping/assembly issues;
- Differences in the Incompatibility patterns May indicated structural variations that are specific for the breed



Conclusion and Perspectives

- Combining IBD estimates and Opposing Homozygotes can be helpful in the estimation of the degree of relationship
- By combining information about the farm and genotyping data it is possible to reconstruct problematic pedigree information
- Around 4% of the Reggiana and Modenese pedigree of the active has been reconstructed or corrected
- Genotyping of the totality of the breeds is ongoing
- The routine application of this method will improve both the managing and conservation of Reggiana and Modenese cattle breed and the accuracy of estimated breeding values.



Animal and Food Genomics Group



Luca Fontanesi
Full Professor



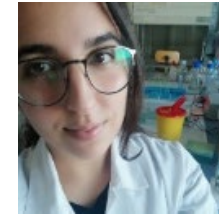
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Acknowledgments

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Thank you ! Questions ?





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