



GEORG-AUGUST-UNIVERSITÄT
GÖTTINGEN

QUALITAS⁺

HD genotype imputation in 54k and ungenotyped Original Braunvieh and Brown Swiss cattle

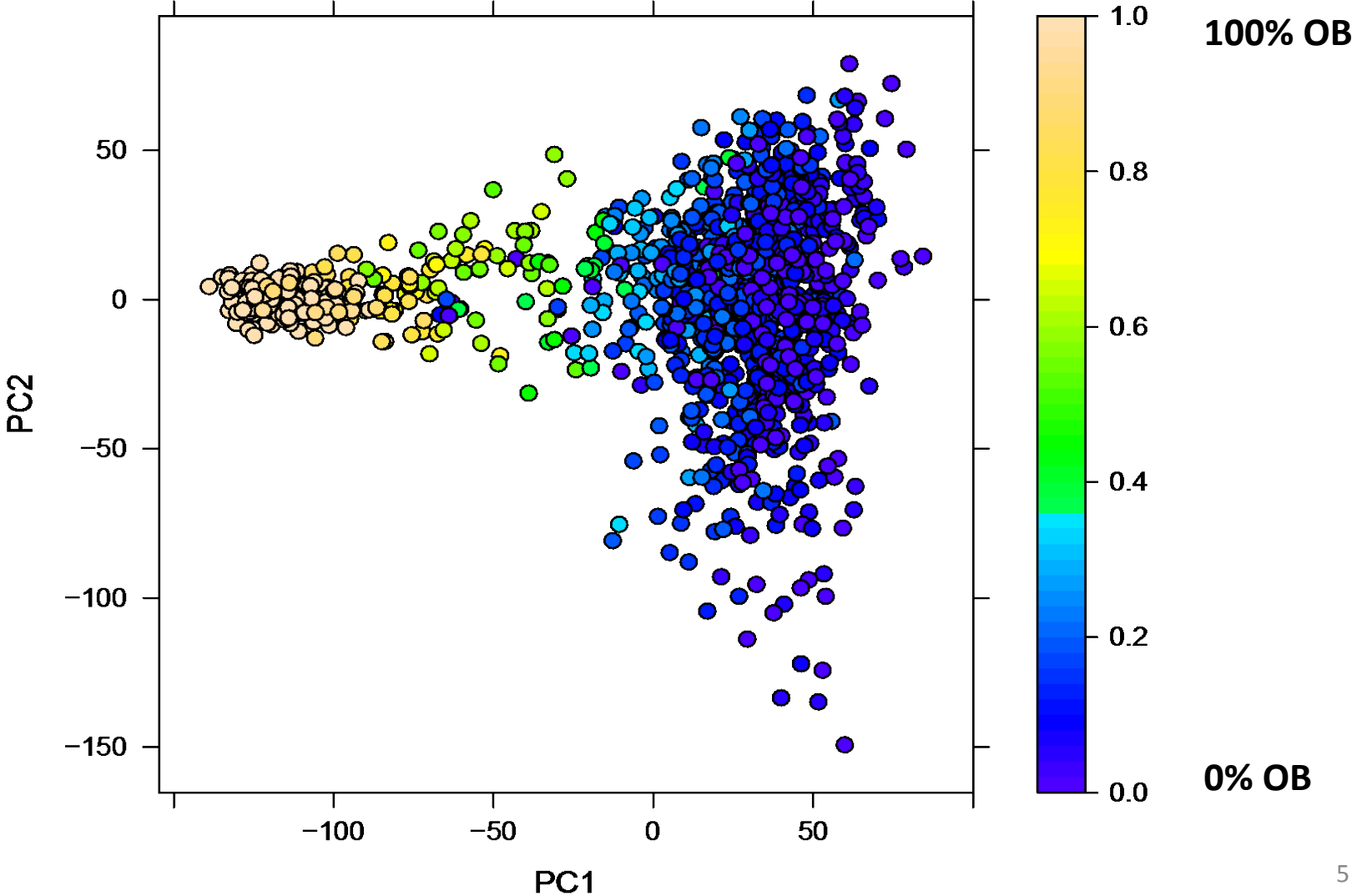
Birgit Gredler, Mehdi Sargolzaei, Beat Bapst, Anna
Bieber, Henner Simianer, Franz Seefried

- Genotype imputation reduce the cost of genotyping
- A common application is to impute genotypes from low density to high density
- Genotype imputation of non-genotyped animals can be attractive for enlarging the reference population
 - for interesting phenotypes
 - of small breeds
- Original Braunvieh (OB) is a small breed in Switzerland
- Low number of bulls genotyped; genomic evaluation is not implemented

- **How accurately can we perform genotype imputation from 50k to HD in Original Braunvieh?**
- **How accurately can we impute non-genotyped animals based on progeny information?**

- 1,173 Illumina HD genotyped bulls (n=307) and cows (n=866)
- 40 Brown Swiss, 173 OB, 960 Braunvieh
- Data editing: MAF \geq 0.5%
SNP-CR, Animal CR \geq 90%
only SNP with known coordinates
- 629,611 SNP

Brown Swiss population structure **QUALITAS**⁺ based on HD genotypes

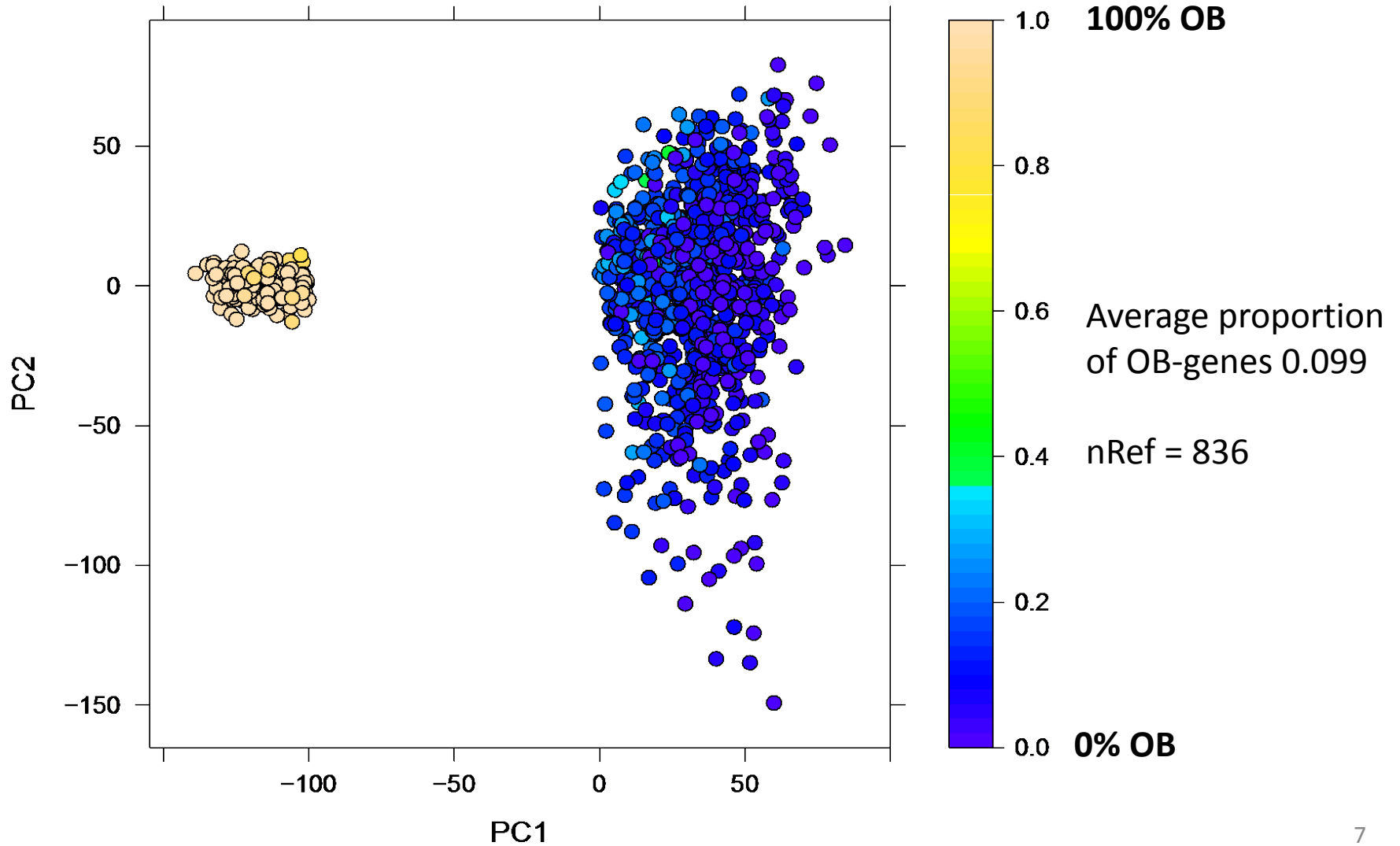


Imputation scenarios – imputation of OB animals

- 43 OB animals were discarded from the testset, since they had progenies in the HD genotyped reference population
- HD genotypes of 130 OB animals were masked to mimic genotyping with the Illumina 50k-chip (40,127 SNP)
- Imputation was performed in 4 different scenarios, where the HD reference population was set up according to proportion of OB-genes

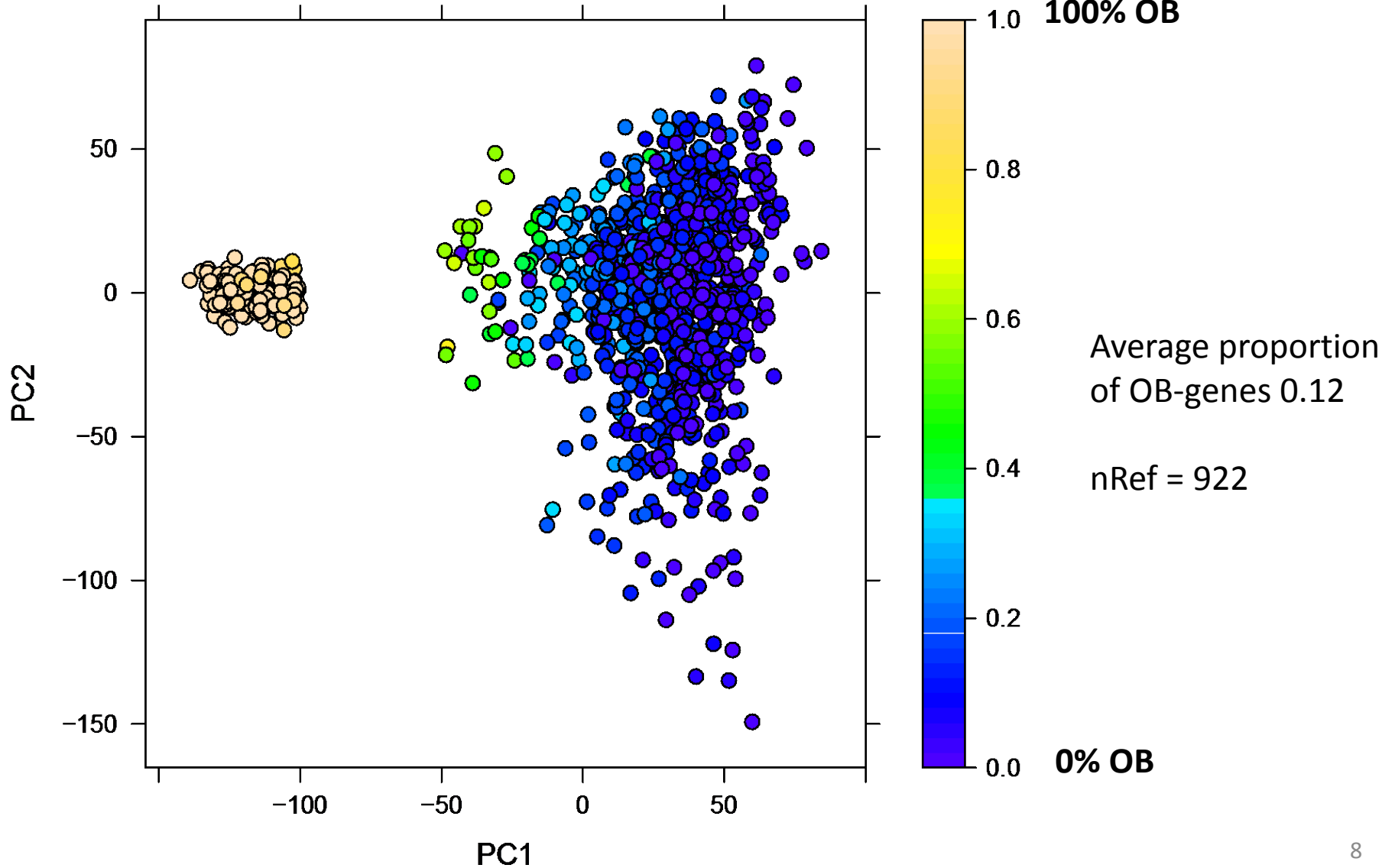
Scenario 1

QUALITAS⁺

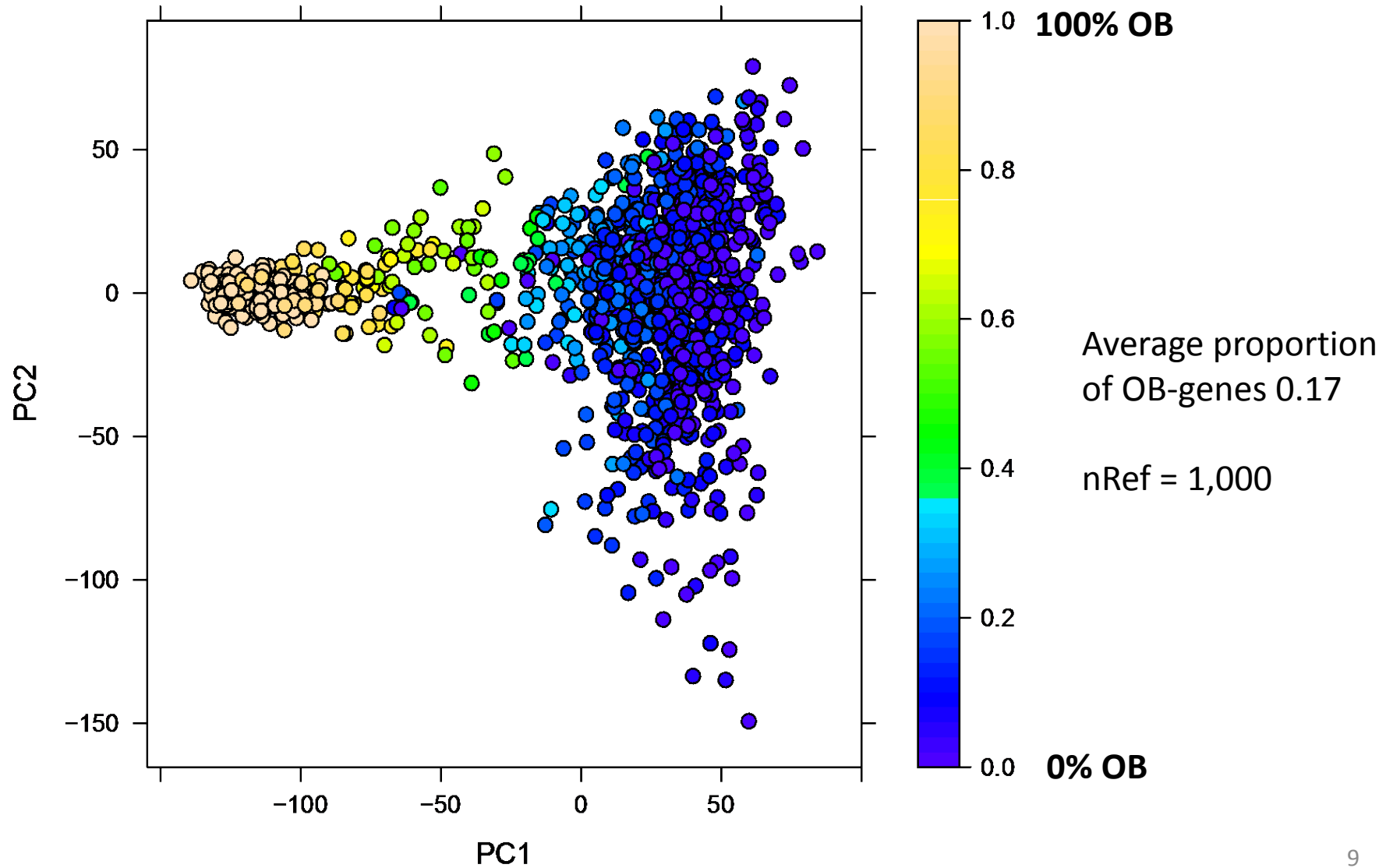


Scenario 2

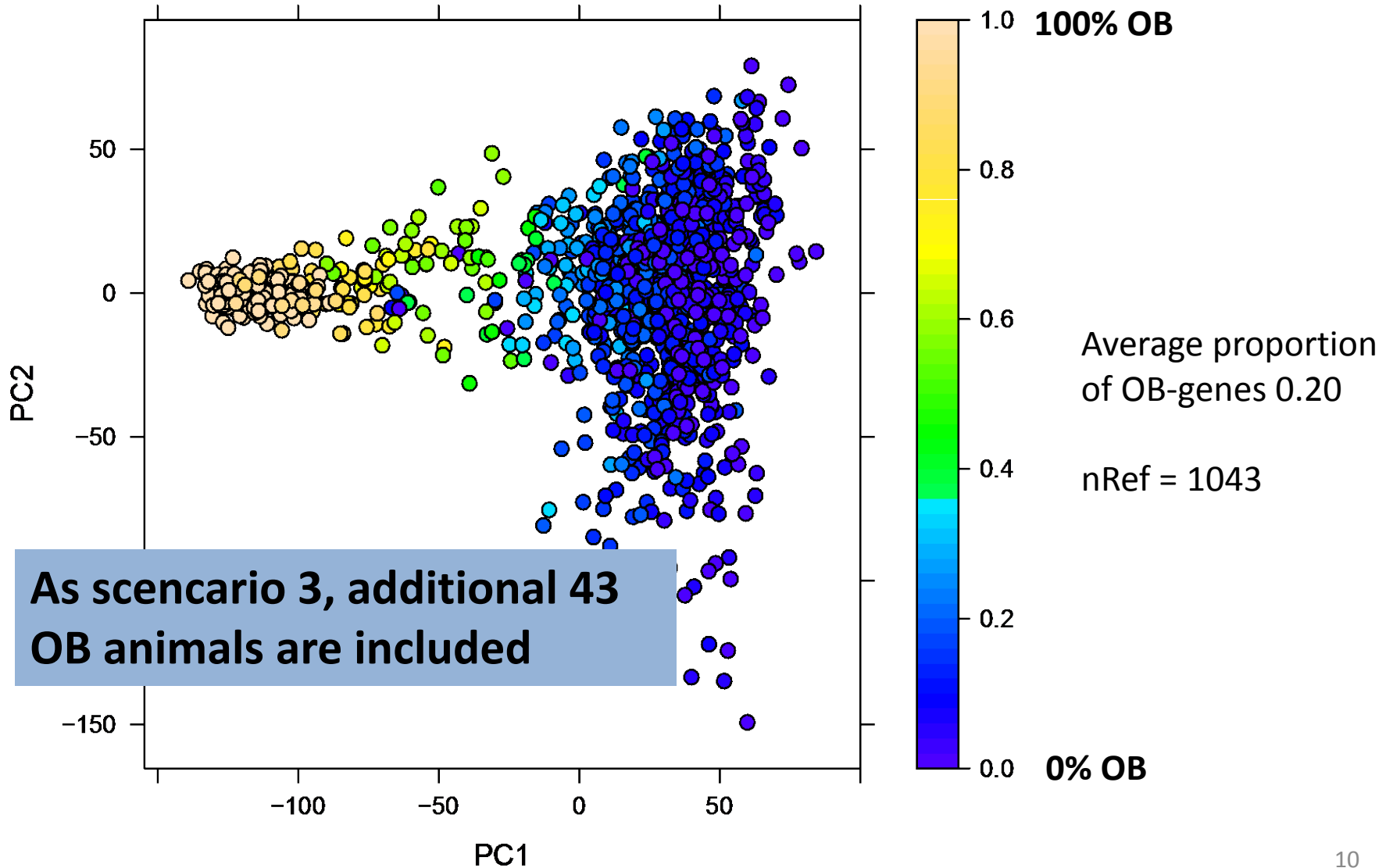
QUALITAS⁺



Scenario 3



Scenario 4



Imputation scenarios – imputation of non-genotyped animals

- Bulls and cows having HD genotyped progenies were removed from the reference population
- These animals were still in the pedigree
- 79 bulls and 15 cows had at least two progenies HD genotyped
- Imputation was based on a reference population of 829 animals

- **FImpute** (Sargolzaei et al., 2012)
- Pedigree information is used to impute genotypes with high certainty
- Secondly, haplotypes are constructed based on a sliding window approach
- Finally, missing genotypes are filled in according to the haplotype library
- Imputation accuracy was assessed by the squared correlation between true and imputed genotypes (r^2), % correctly and incorrectly imputed genotypes

Accuracy of imputation for OB animals

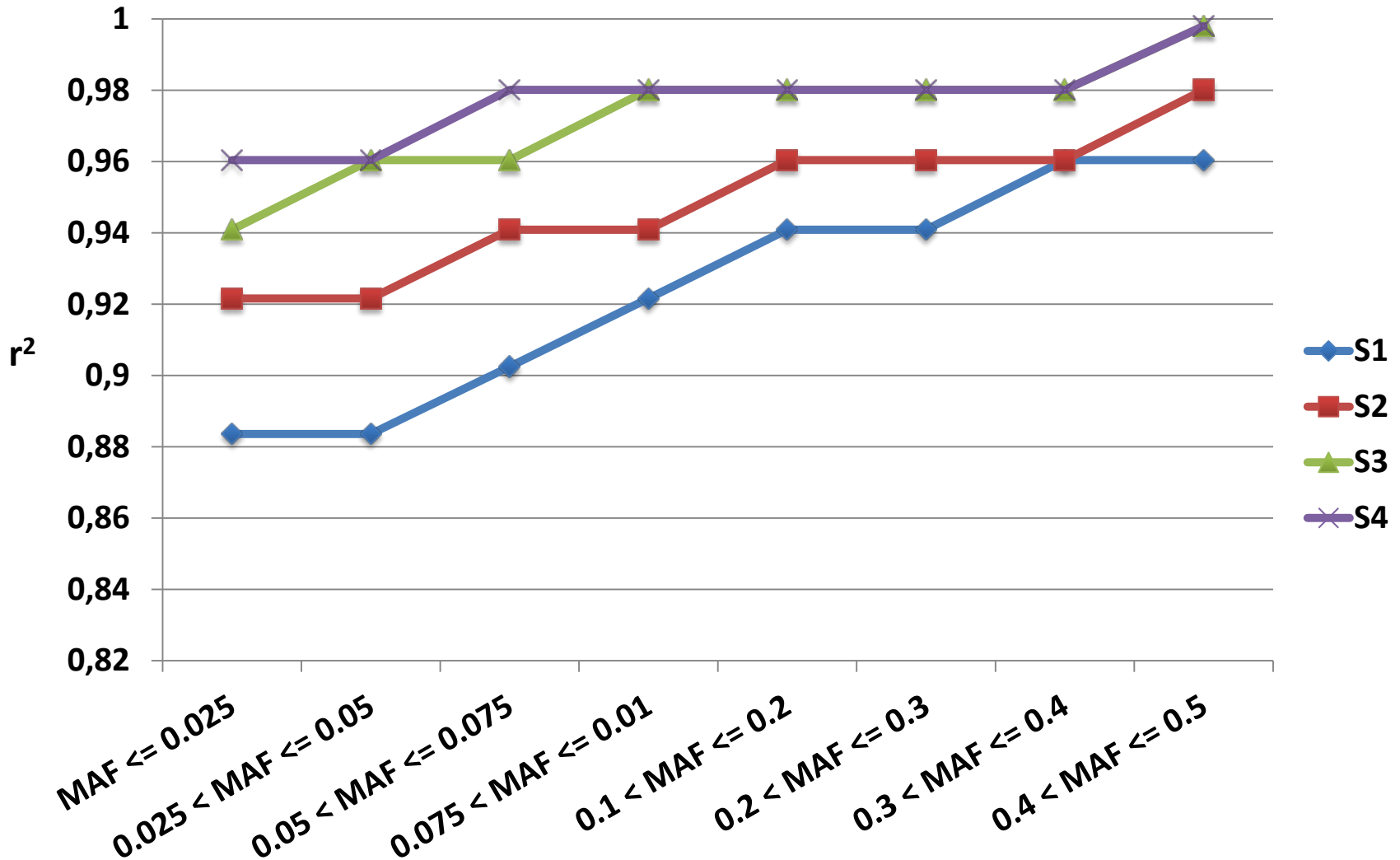


	Scenario 1				Scenario 2		
	nRef=836	avgOB=0.099			nRef=922	avgOB=0.12	
	n	r ²	% Corr		n	r ²	% Corr
Sire & dam							
Sire & MGS							
Sire							
Dam							
Other	130	0.921	94.7		130	0.948	96.4

Accuracy of imputation for OB animals

	Scenario 3 nRef=1000 avgOB=0.17				Scenario 4 nRef=1043 avgOB=0.20		
	n	r ²	% Corr		n	r ²	% Corr
Sire & dam					3	0.999	99.6
Sire & MGS					11	0.987	98.9
Sire					19	0.978	98.6
Dam	4	0.970	98.1		1	0.980	99.3
Other	126	0.968	97.9		96	0.971	98.1

Accuracy of imputation according to MAF



Accuracy of imputation of non-genotyped animals

No. progeny	n	r ²	% Correct	% Incorrect
2	39	0.58	64.2	26.4
3	14	0.76	78.5	18.1
4	14	0.74	76.4	13.9
5	4	0.75	78.2	17.6
6 - 10	12	0.83	86.0	12.2
11 - 19	7	0.89	91.7	6.8
>=20	4	0.97	97.6	2.3

Accuracy of imputation of non-genotyped animals

No. progeny	Brown Swiss		Braunvieh		Original Braunvieh	
	n	r ²	n	r ²	n	r ²
2	10	0.63	19	0.57	10	0.53
3	3	0.76	7	0.74	4	0.78
4	3	0.76	6	0.74	5	0.74
5	2	0.71	2	0.79	-	-
6 - 10	2	0.83	7	0.85	3	0.79
11 - 19	-	-	7	0.89	-	-
>=20	1	0.96	3	0.97	-	-

Summary

- High accuracy of genotype imputation of OB animals
- Imputation accuracy of non-genotyped animals was good when several progenies were genotyped
- Adding sire, dam and/or MGS information will improve accuracy of non-genotyped animals (Session 55, Bouwman et al., 2013)



Thank you!

Genotypes were provided by:

Braunvieh Schweiz

Genotype pool Germany-Austria

Associazione Nazionale Allevatori

Bovini della Razza Bruna

Beltsville Agricultural Research Centre

LowInputBreeds, FP7 – project No KBBE

222 632

The views expressed by the authors do not necessarily reflect the views of the European Commission, nor do they in any way anticipate the Commission's future policy in this area.