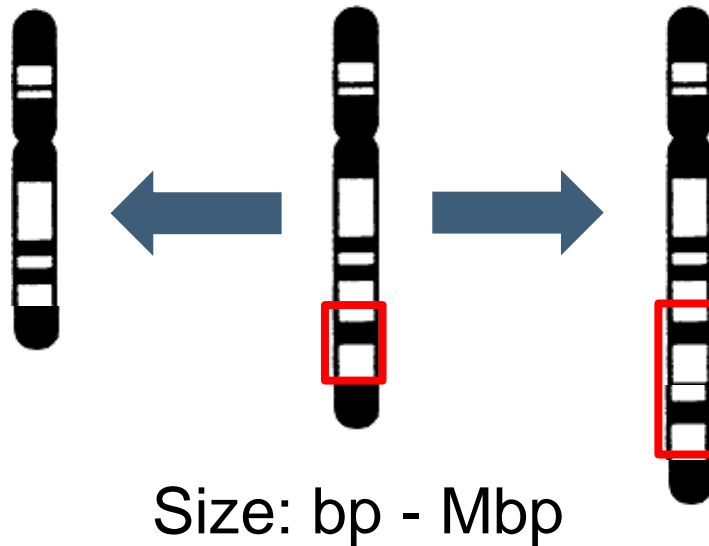


Objectives

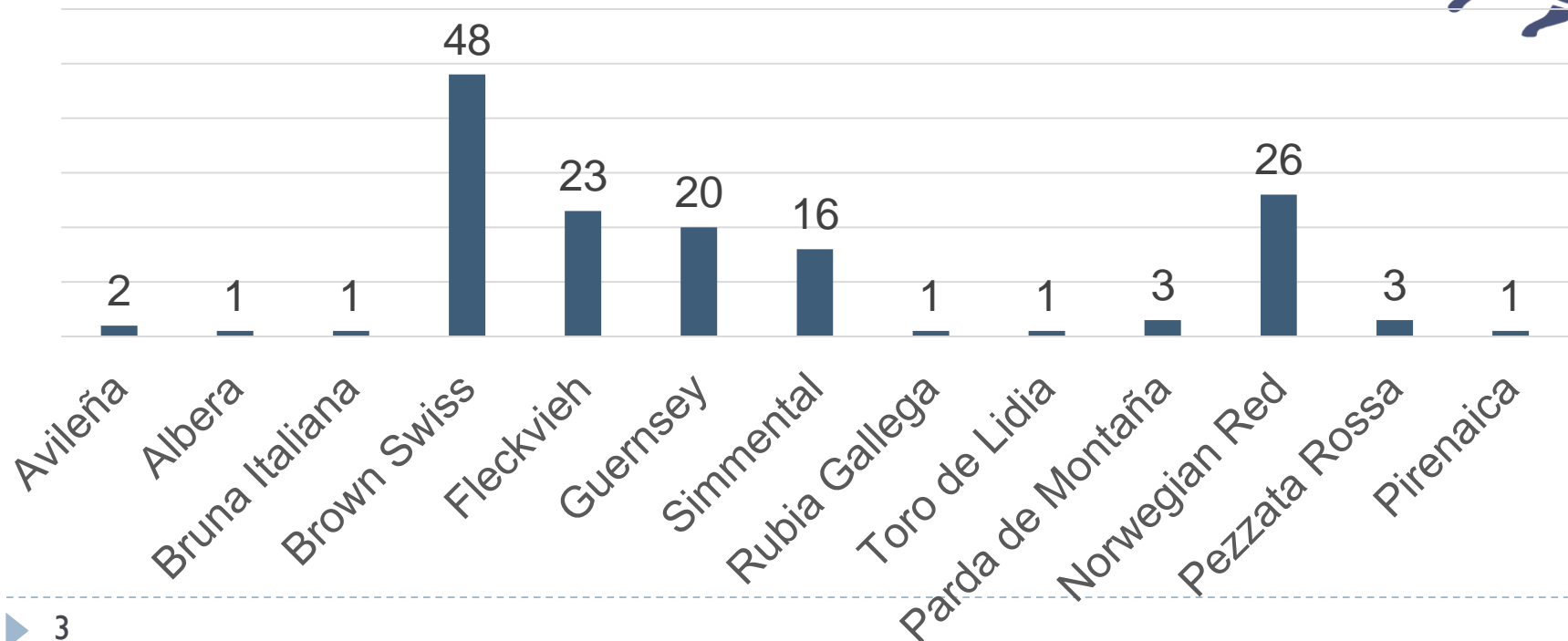
- ▶ detecting of CNV polymorphisms



- ▶ describing the **inter-individual** and **inter-breed** variation

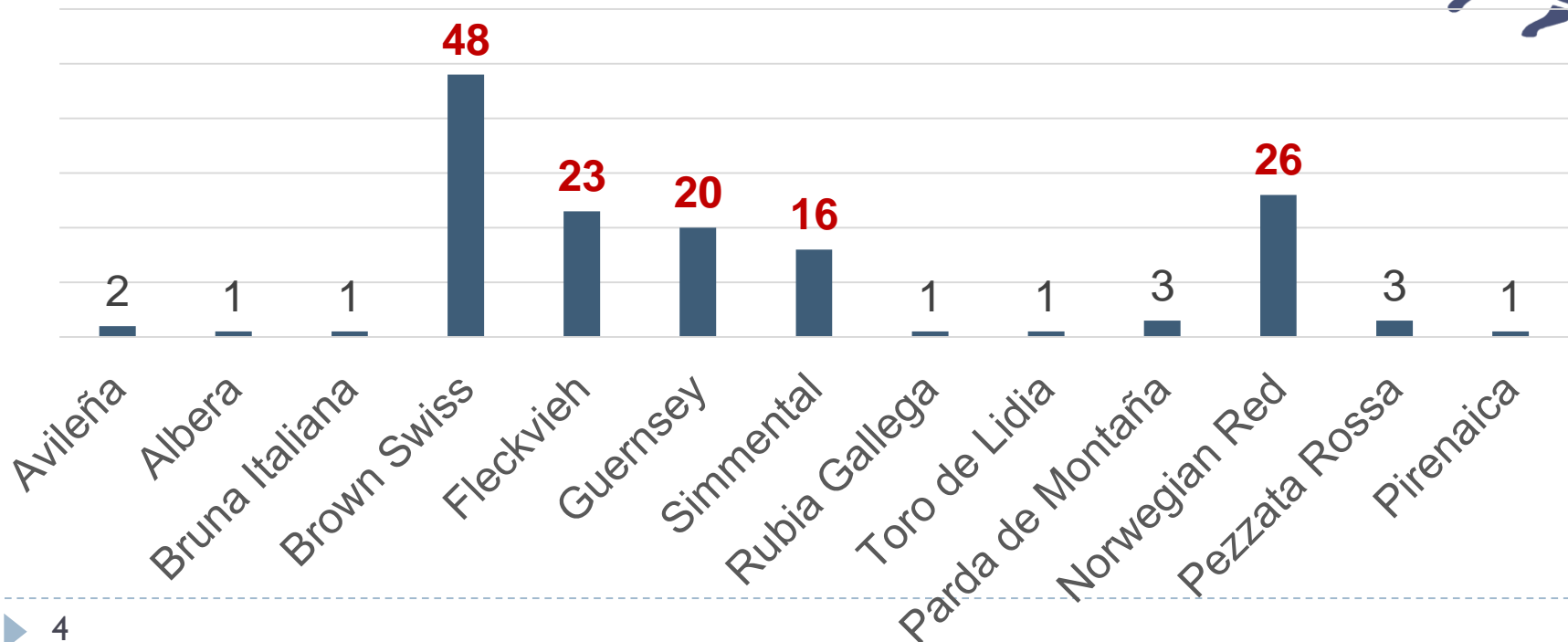
Dataset

- ▶ Whole genome DNaseq – **146** bulls
- ▶ Illumina HiSeq next-generation sequencing platform
- ▶ 7-28 x genome average coverage
- ▶ 13 breeds / 5 breeds

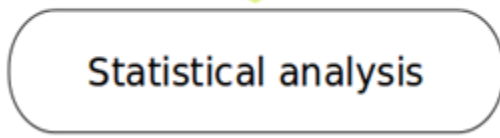
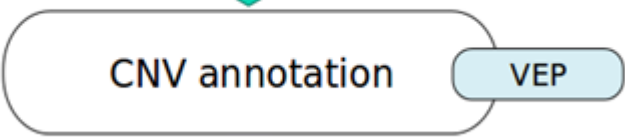
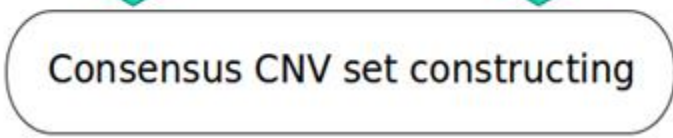
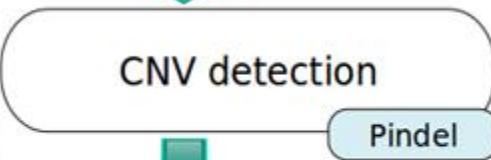
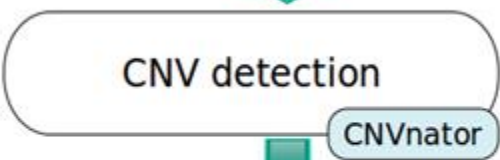
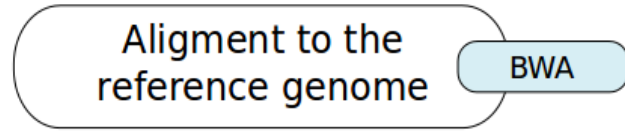


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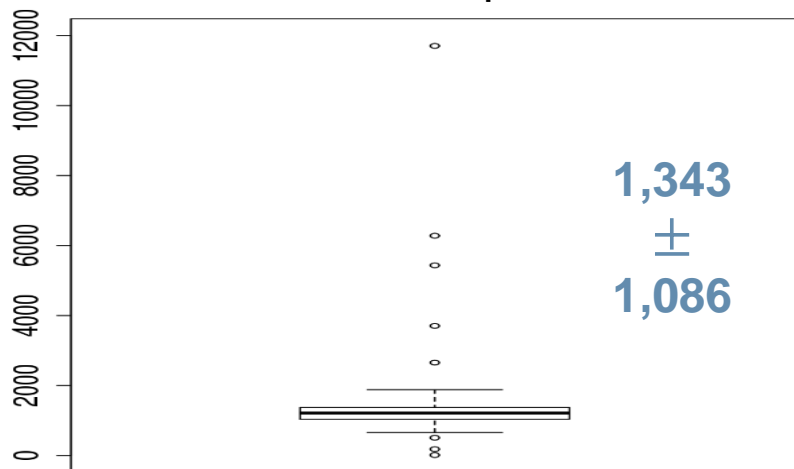
Methods



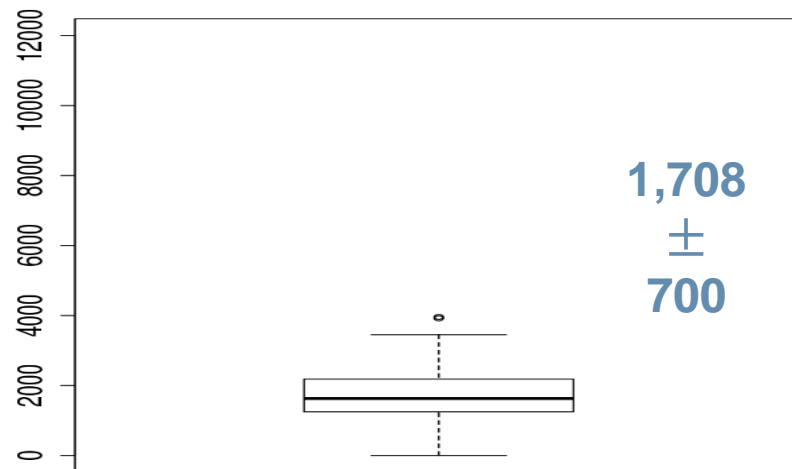
Results

Total **number**/**length** of dup/del called

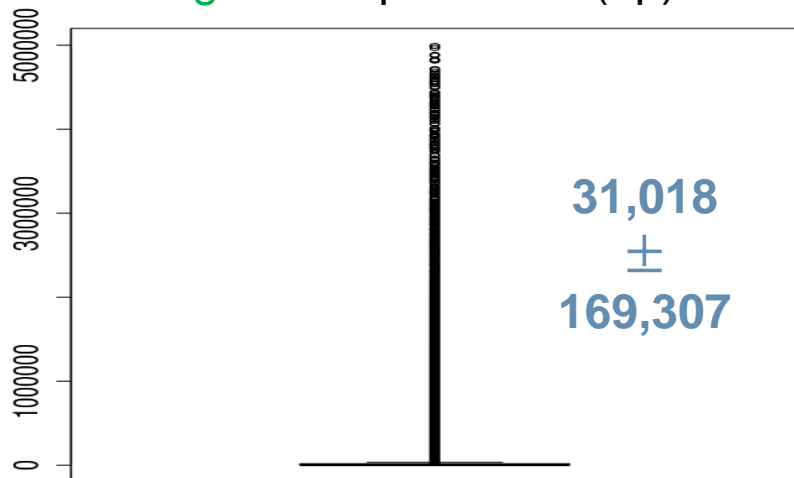
number of duplications



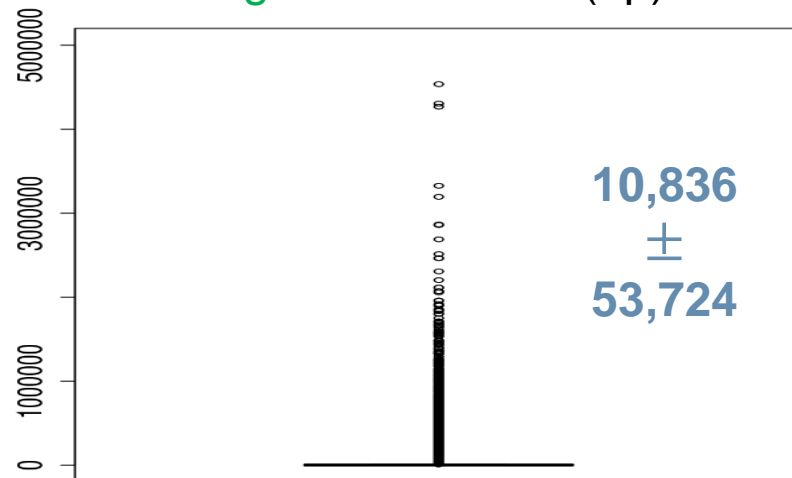
number of deletions



length of duplications (bp)



length of deletions (bp)

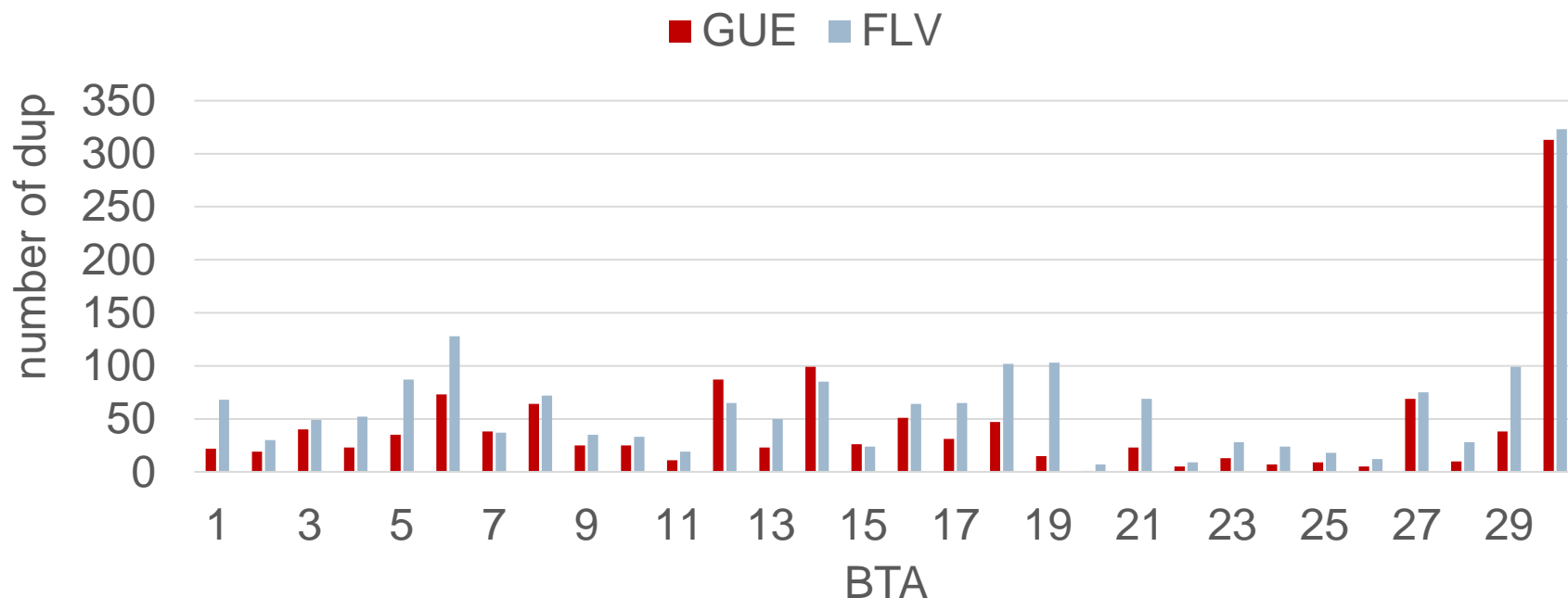


Results

Number of dup/del

- not uniformly distributed across 146 individuals

dup: $P < 2.20 \cdot 10^{-16}$, del: $P < 2.20 \cdot 10^{-16}$



Results

Number of dup/del

- ▶ not uniformly distributed across 146 individuals
dup: $P < 2.20 \cdot 10^{-16}$, del: $P < 2.20 \cdot 10^{-16}$
- ▶ not uniformly distributed across individuals within each breed
dup: $P < 2.20 \cdot 10^{-16}$, del: $P < 2.20 \cdot 10^{-16}$
(for each breed)
- ▶ variability between breeds exists
dup: $P=0.01932$, del: $P=0.01006$ ($\alpha=0.05$)

Results

Length of dup/del

- ▶ distribution not the same for 146 individuals

$$\text{dup: } P < 2.20 \cdot 10^{-16}, \text{ del: } P < 2.20 \cdot 10^{-16}$$

- ▶ distribution not the same across individuals within each breed

$$\text{dup: } P \in (3.02 \cdot 10^{-94}, 0.1 \cdot 10^{-12})$$

$$\text{del: } P \in (1.23 \cdot 10^{-192}, 0.1 \cdot 10^{-12})$$

- ▶ variability between breeds exists

$$\text{dup: } P < 2.20 \cdot 10^{-16}, \text{ del: } P < 2.20 \cdot 10^{-16}$$

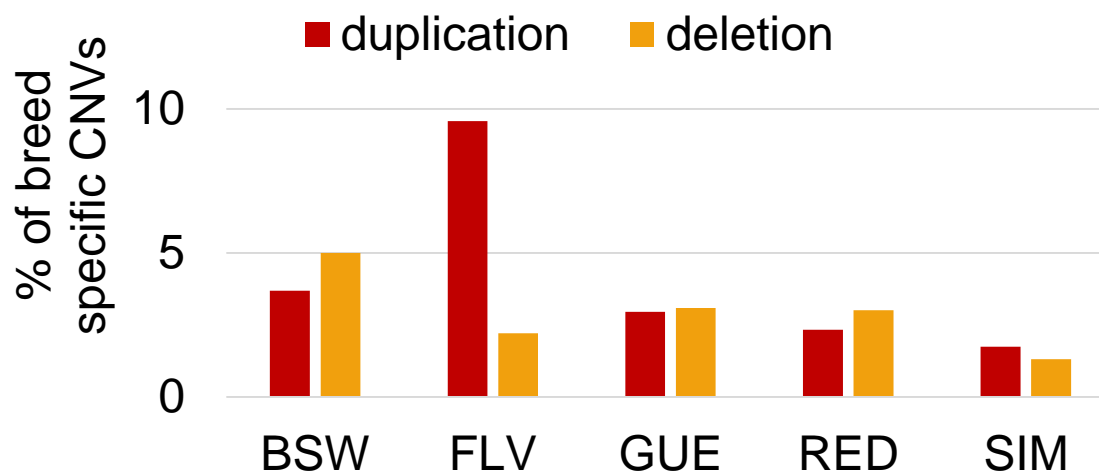
Results

Unique, shared and breed specific duplications

▶ **Unique** (1/146): dup 84.85 %, del 77.22 %

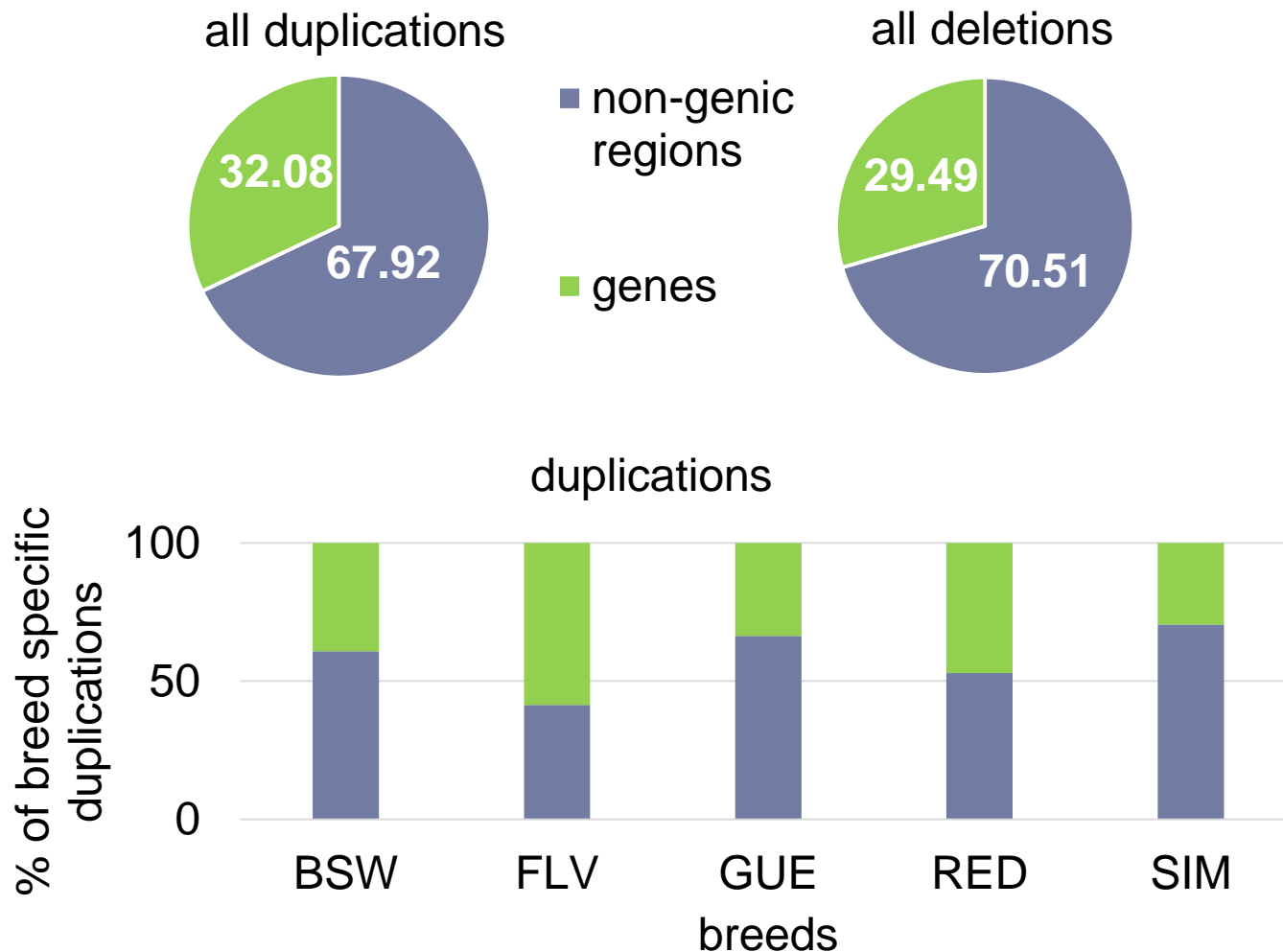
▶ Max **shared**: dup 117/146, del - 140/146

▶ **Breed specific**: min $2/N_{\text{breed}}$







Results

Functional CNV annotation



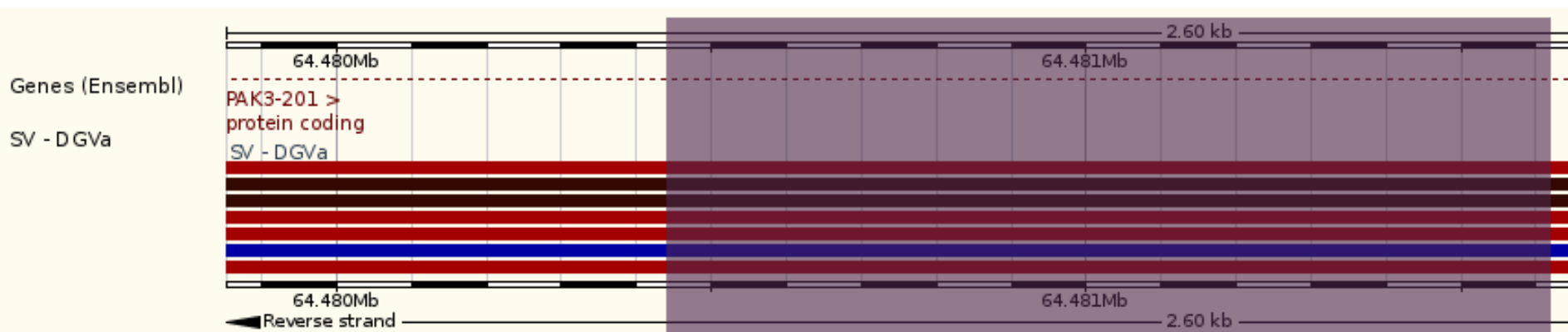
Results




20 most common dup (117-74 bulls) and del (140-117 bulls)

	intergenic	intron	protein coding	non-protein coding
dup	13	4 	1 	2 
del	19	1 	-	-

 modifier  high

Common variants overlapp with DGVa



 gain (Bickhart *et al.* 2012)  tandem duplication (Boussaha *et al.* 2015)
 deletion (Liu *et al.* 2010, Boussaha *et al.* 2015)

Conclusions

- ▶ high complexity of a CNV landscape in *Bos taurus* genomes
- ▶ varying density of CNV depends on genome function
- ▶ the breed-specific phenomenon in the Fleckvieh
- ▶ deletion events in coding regions less evolutionary accepted than duplications



Thank you!



Acknowledgements

National Science Centre in Poland:

Describing the genome-wide distribution of copy number variations in various breeds of domestic cattle based on the next-generation sequencing data of 121 individuals

National Science Centre in Poland:

Resolving genetic predisposition to clinical mastitis based on whole genome sequences of 32 cows

European Union's Seventh Framework Programme

Poznan Supercomputing and Networking Center



Conclusions

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