

# The analysis of CNV regions in five breeds of bulls



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## CONCLUSIONS

- Deletion regions shorter than duplication regions
- More deleted regions than duplicated regions
- Unique regions in particular breeds mostly located in intergenic regions
- BSW animals more similar within group than other breeds

## MATERIAL

- Whole genome sequence with IlluminaHiSeq2000
- 7-28 x genome average coverage
- five breeds: Brown Swiss (BSW) – 48, Fleckvieh (FLV) – 29, Guernsey (GUE) – 20, Simmental (SIM) - 16, Norwegian Red (RED) – 19

## METHODS

### Bioinformatics pipeline

- Alignment to UMD3.1 (BWA-MEM)
- Post alignment filtering (Picard & Samtools)
- CNV calling (CNVnator & Pindel)

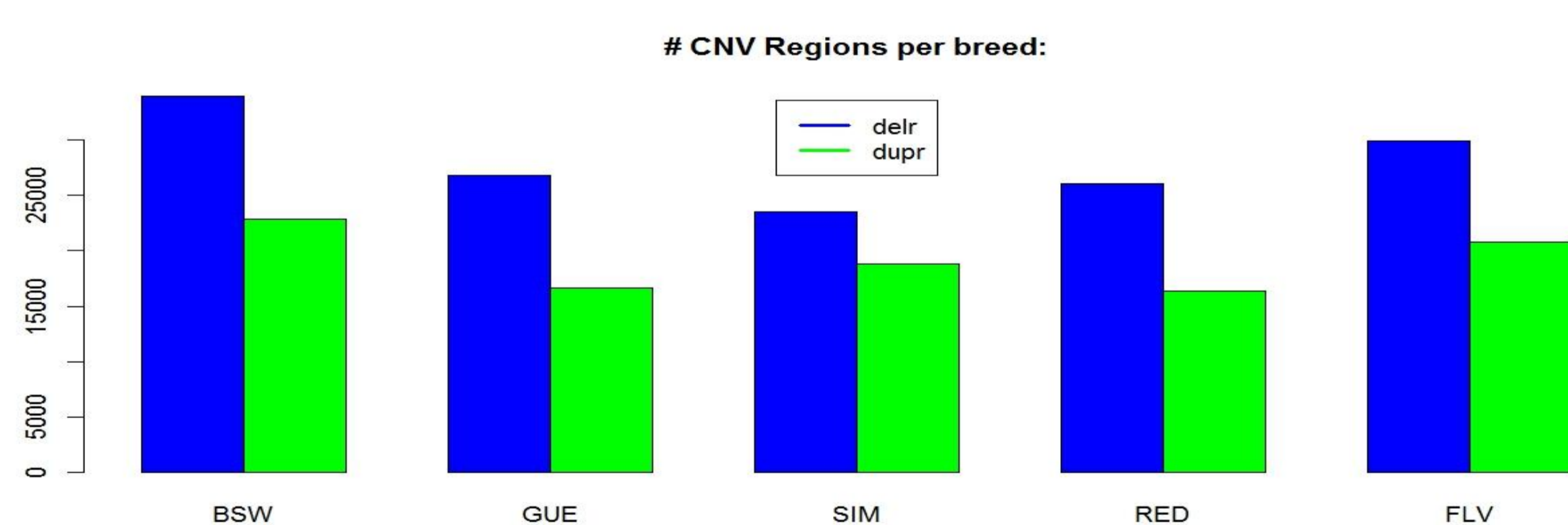
### CNV regions analysis

- Descriptive statistics
- Jaccard similarity measure
- Non – metric multidimensional scaling
- Mann-Whitney-Wilcoxon tests
- Functional annotation

## RESULTS

Validation (CNVnator in 70% overlapped by Pindel)

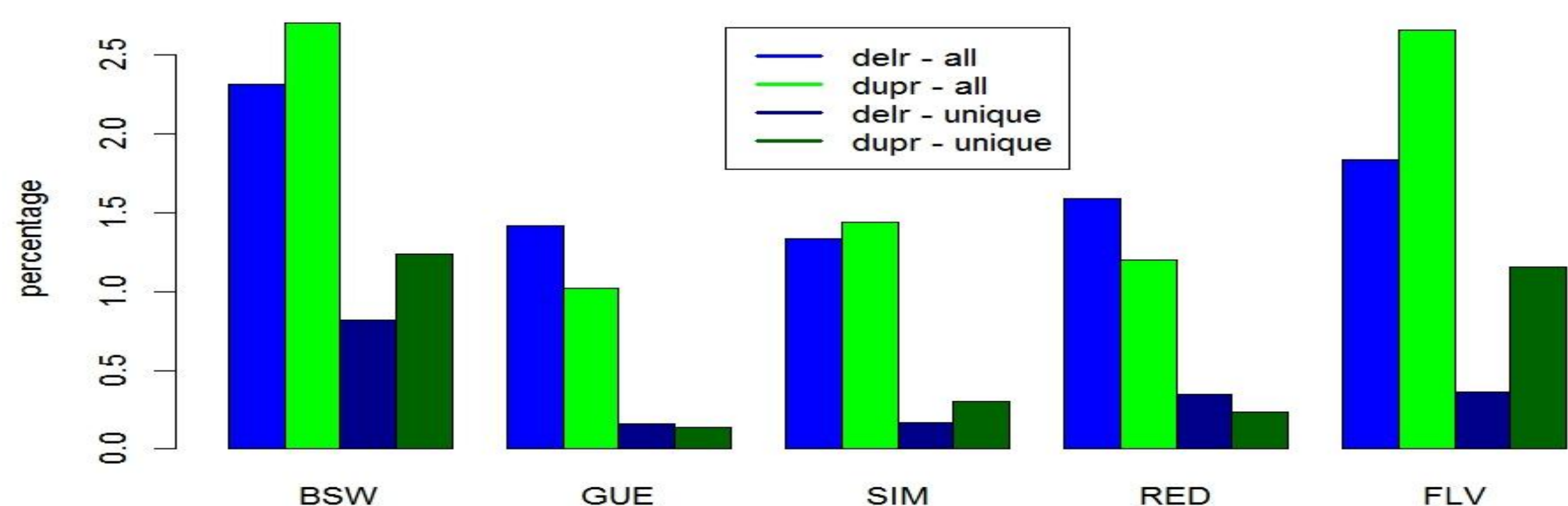
10.20% deletions & 20.26% duplications



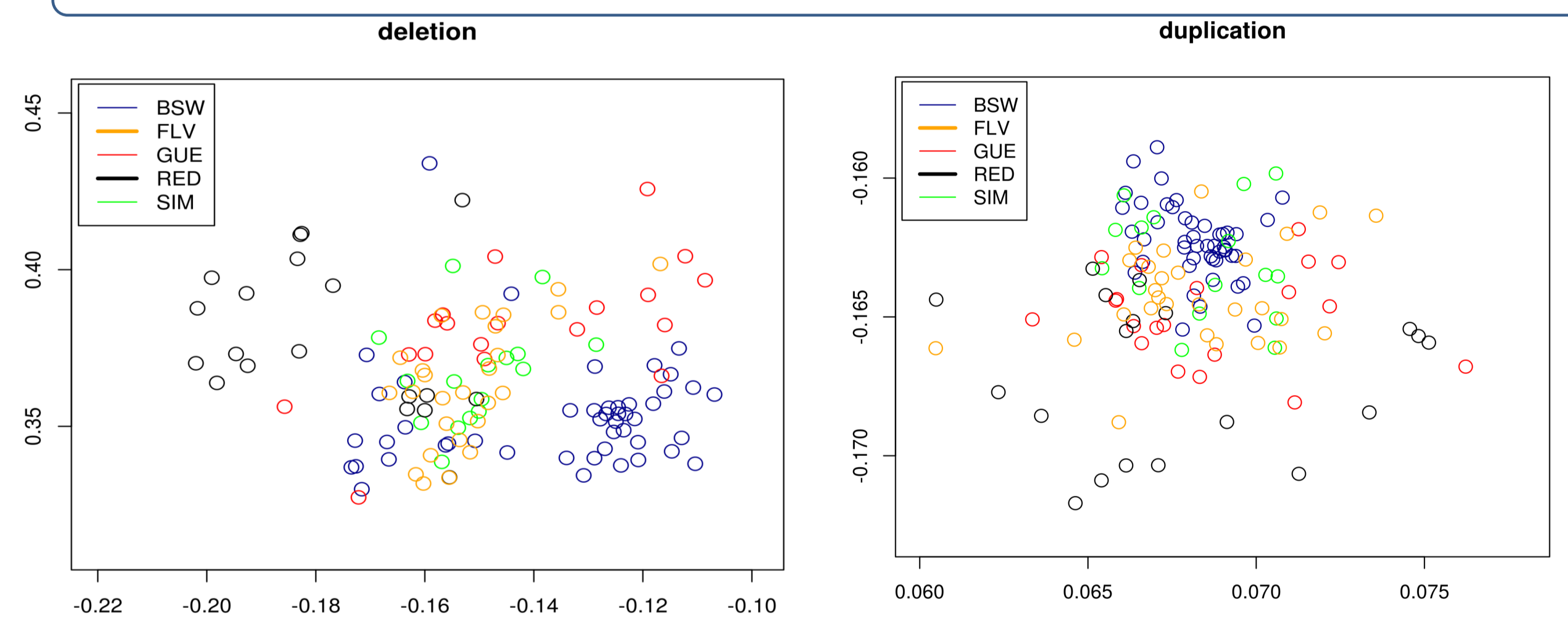
# unique CNVR per breed:

	BSW	GUE	SIM	RED	FLV
DELR	6227	2318	1363	2838	3161
DUPR	2976	409	1006	609	1739

## Genome occupation



## Non - metric MDS

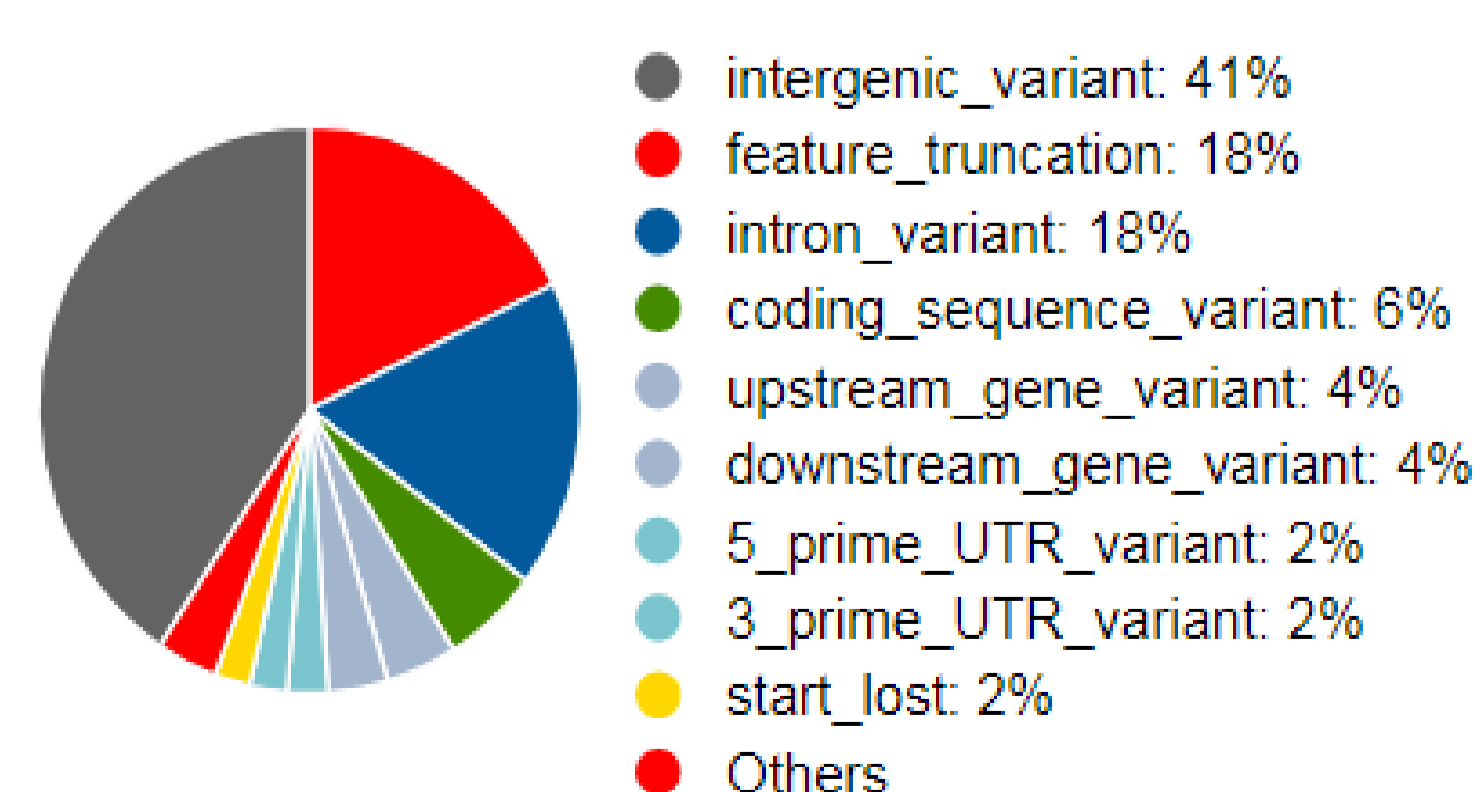


BSW mostly similar within the group (DEL:  $P=4.95 \cdot 10^{-5}$ , DUP:  $P=6.1 \cdot 10^{-87}$ )

## Functional annotation

CNVR – unique per breed

deletion



duplication

