

Analysis of copy number variation regions in bovine genome



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CONCLUSIONS

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

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

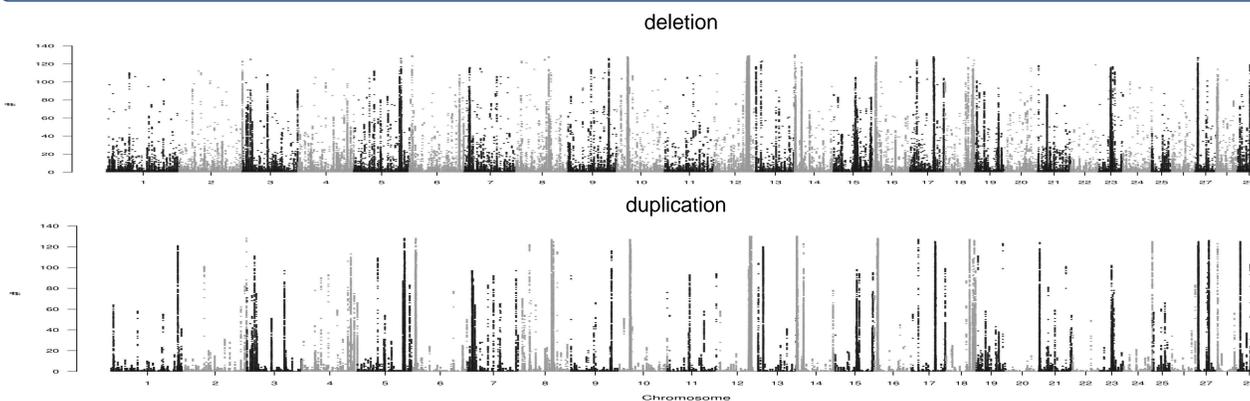
- Descriptive statistics
- Jaccard similarity measure
- Non – metric multidimensional scalling
- Mann-Withney-Wilcoxon tests
- Standard genetic distance of Nei
- Functional annotation

RESULTS

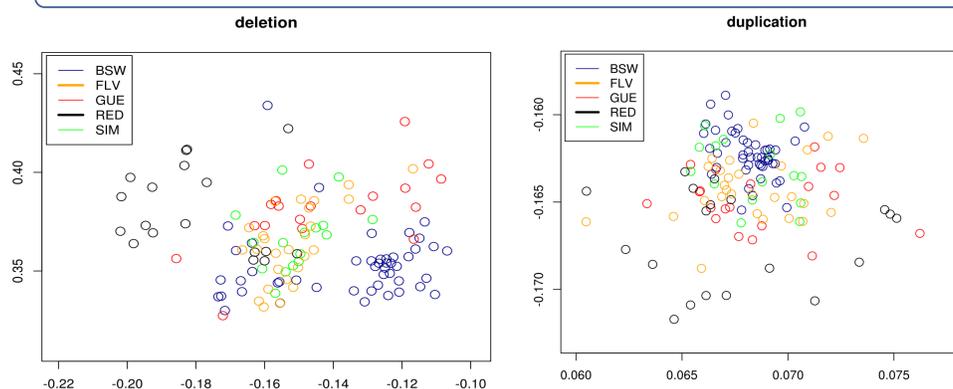
Validation (CNVnator in 70% overlapped by Pindel)

DEL: 2 003 701 → 191, 125 (9.53%) & DUP: 492 417 → 95 587 (19.41%)

Number of individuals sharing CNV in each region of genome



Non - metric MDS



BSW mostly similar within the breed

DEL: $P=4.95 \cdot 10^{-5}$,
DUP: $P=6.1 \cdot 10^{-87}$

DUPR longer than DELR

BSW: $P=1.84 \cdot 10^{-924}$
FLV: $P=8.98 \cdot 10^{-240}$
SIM: $P=2.78 \cdot 10^{-71}$
RED: $P=3.56 \cdot 10^{-65}$
GUE: $P=2.61 \cdot 10^{-44}$

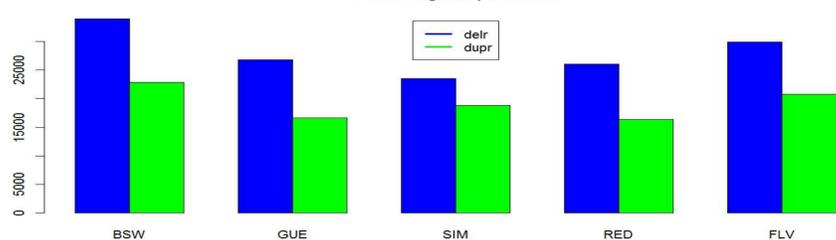
Nei genetic distance

		DEL				
		BSW	FLV	GUE	SIM	RED
DUP	BSW	-	0.106599	0.1558	0.12951	0.1792
	FLV	0.081023	-	0.11916	0.084303	0.11724
	GUE	0.133975	0.091897	-	0.167443	0.17051
	SIM	0.099133	0.082934	0.151943	-	0.15313
	RED	0.156794	0.111289	0.146719	0.160601	-

CNVR – unique per breed

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

CNV Regions per breed:



Functional annotation

- GO: Olfactory system (DUPR – RED (-), GUE (+), SIM (+))
- KEGG: Olfactory transduction (DUPR – GUE, SIM; DELR – RED)
- KIT gene (DELR – BSW)

Acknowledgements:

