Characterization of copy number variants in a large multi-breed population of beef and dairy cattle using high-density single nucleotide polymorphism genotype data

P. Rafter*†, D. C. Purfield*, D. P. Berry*2, A. C. Parnell†, I.C. Gormley†, J.F. Kearney#, M.P. Coffey¥, T. R. Carthy*

*Teagasc, Animal & Grassland Research and Innovation Centre, Moorepark, Fermoy, Co. Cork, Ireland.

† UCD School of Mathematics and Statistics, Insight Centre for Data Analytics, University College Dublin, Belfield, Dublin 4, Ireland.

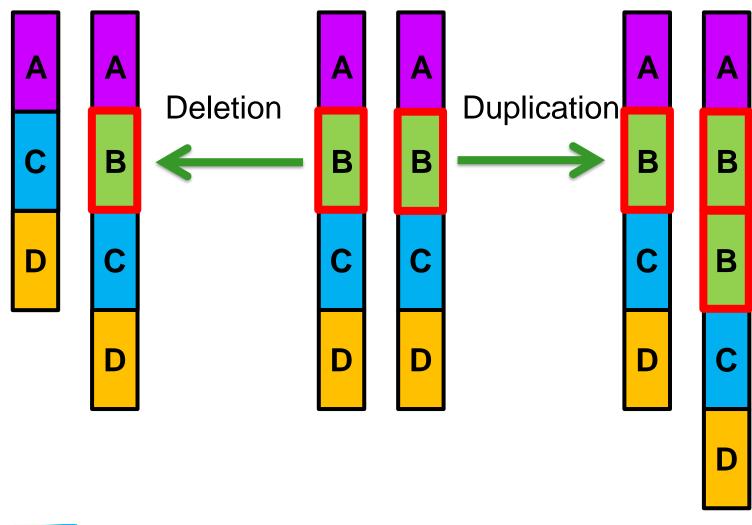
¥ Animal & Veterinary Sciences, SRUC, Roslin Institute Building, Easter Bush, Midlothian, Scotland. EH25 9RG.

ICBF, Highfield House, Shinagh, Bandon, Co.Cork, Ireland.





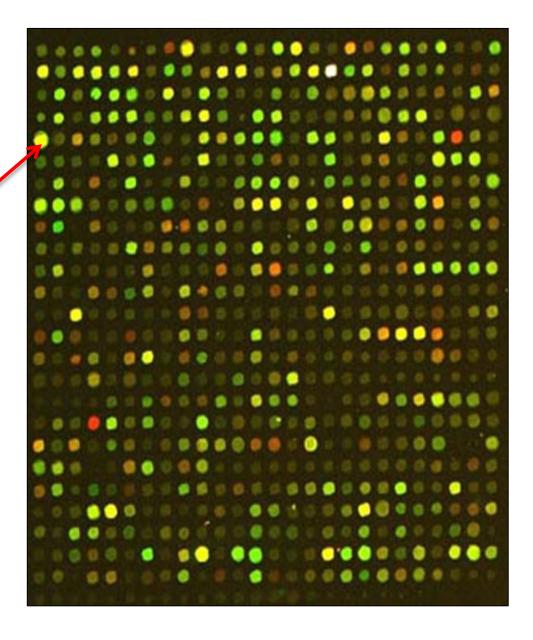
What is a copy number variant (CNV)?







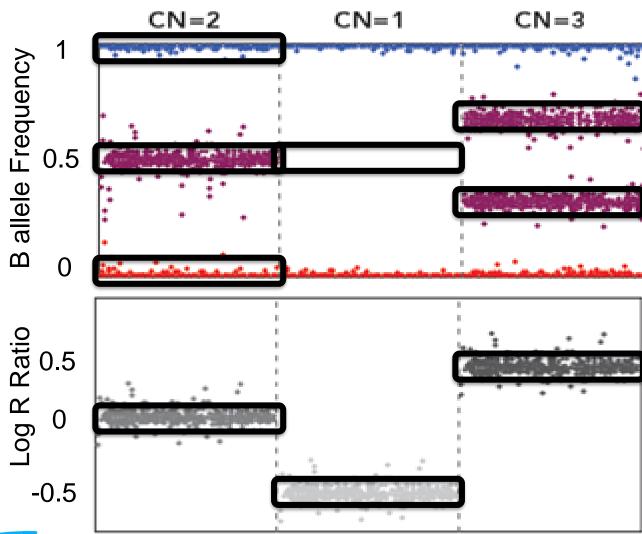
 $\log \left(\frac{Observed\ Fluorescence}{Expected\ Fluorescence} \right)$







Copy number analysis

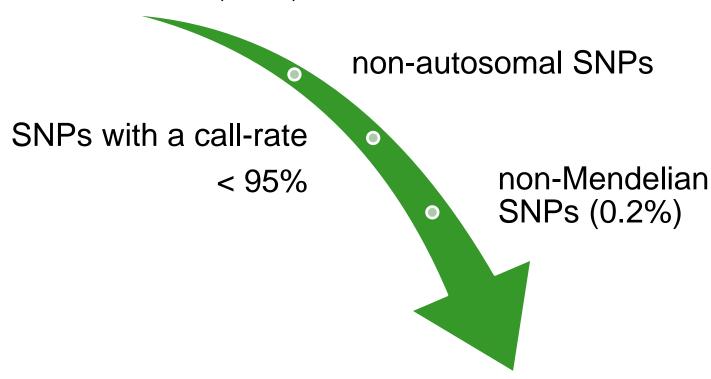






Genotype data

Illumina BovineHD, 777,962 SNPs

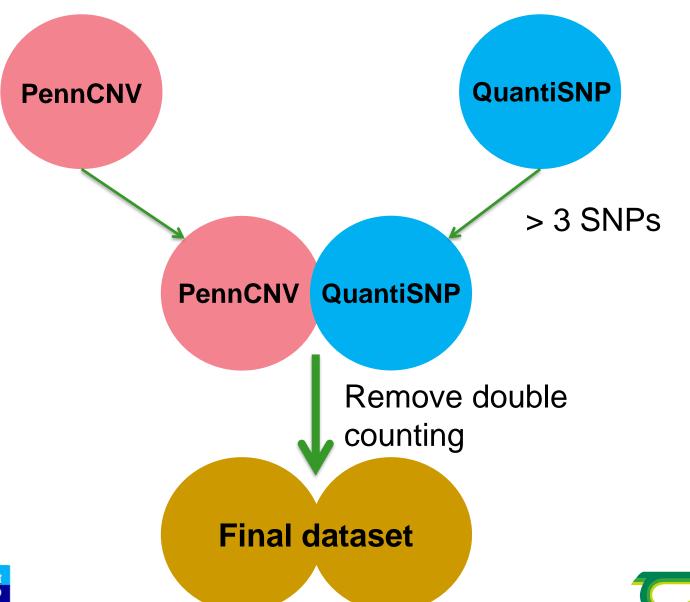


713,162 SNPs





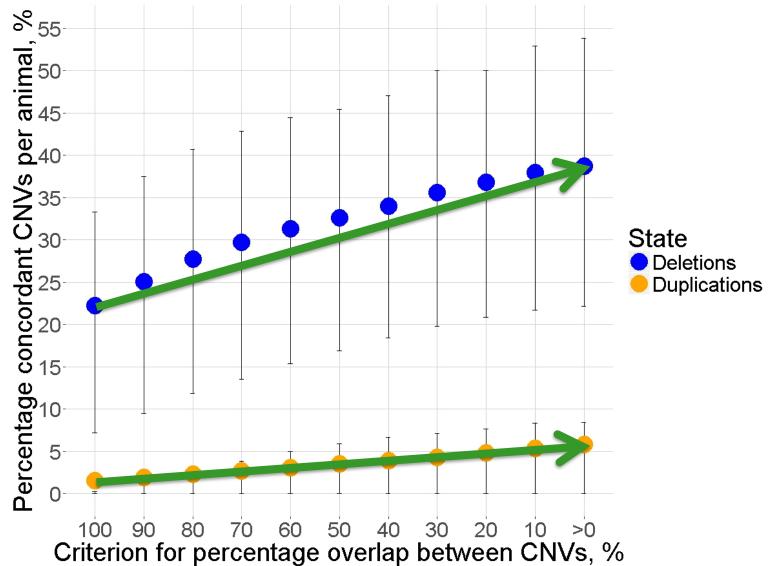
The CNV dataset







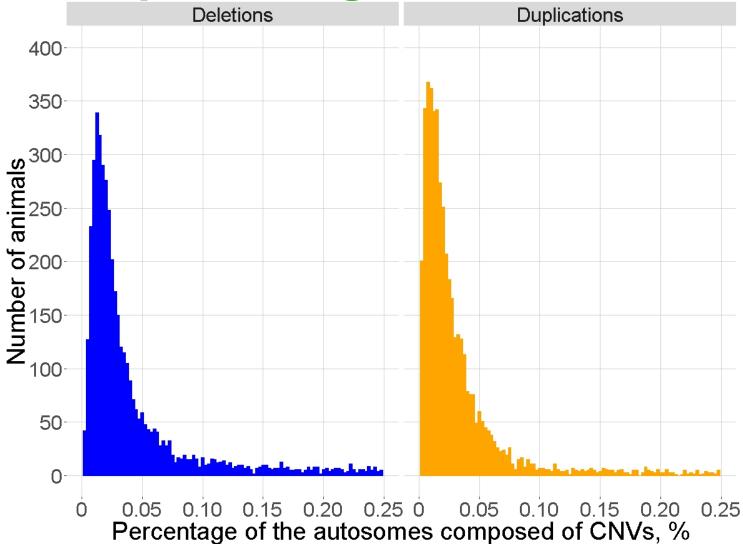
Concordance between PennCNV and QuantiSNP







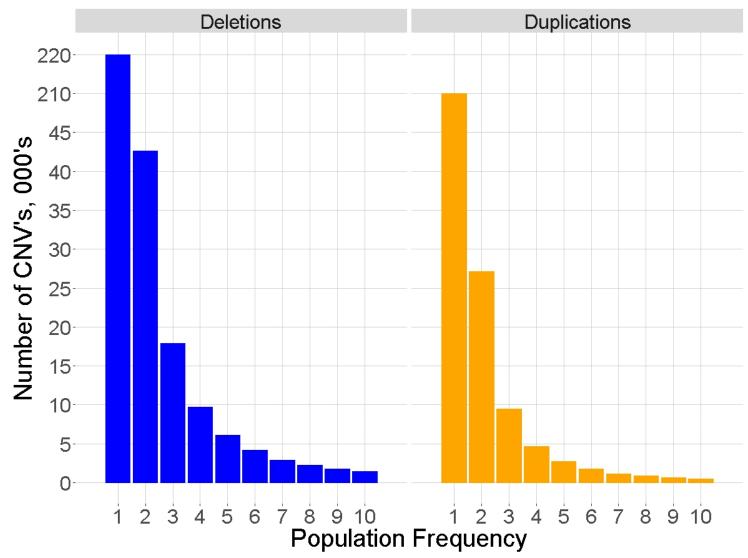
CNV percentage of autosomes





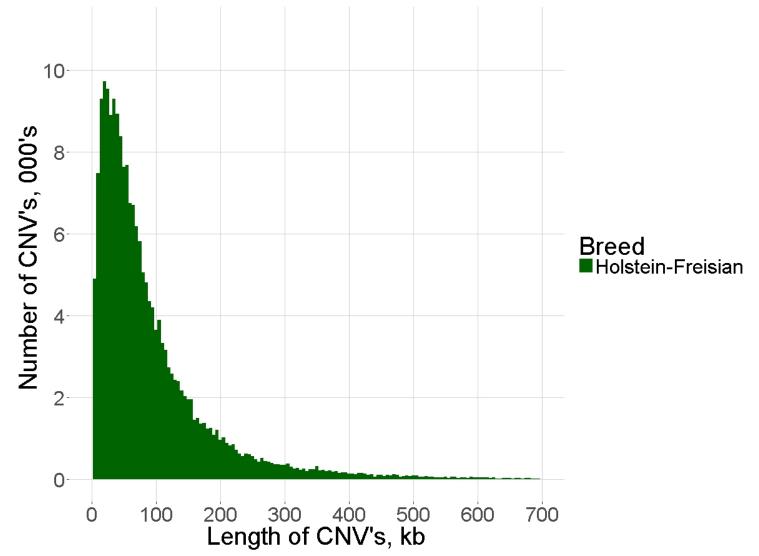


Frequency of CNVs



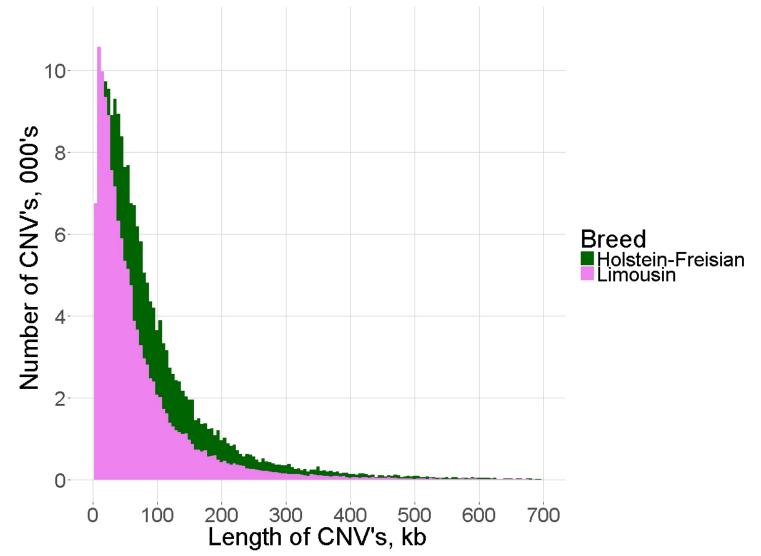






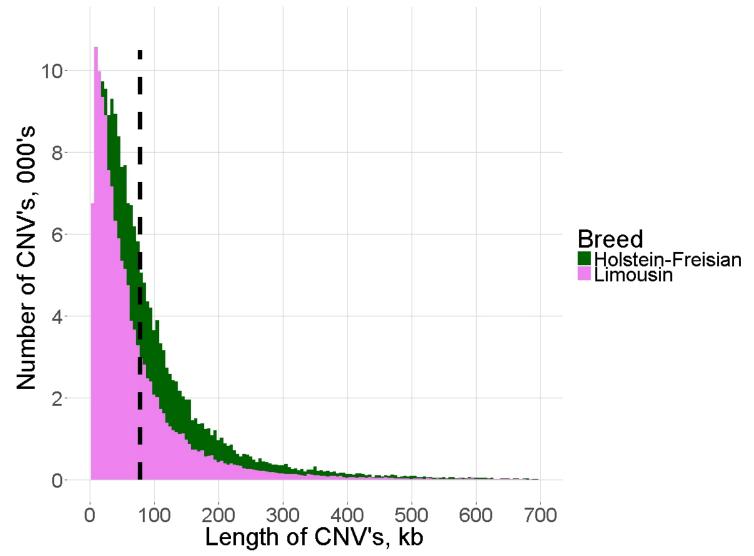






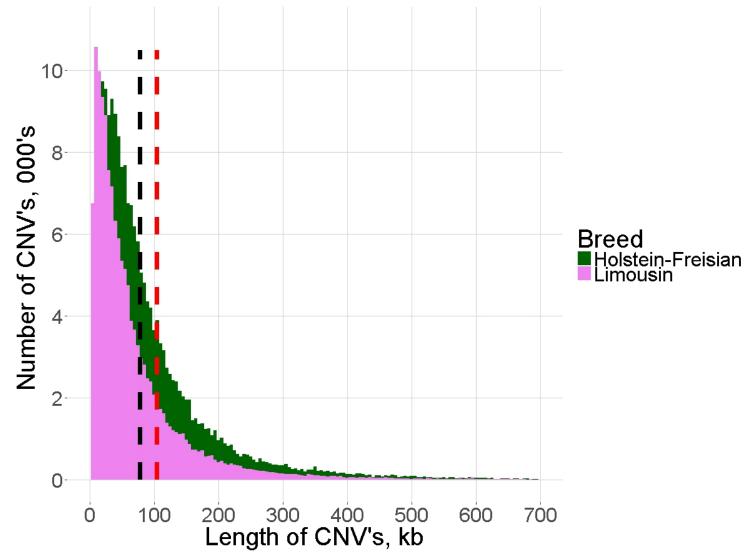






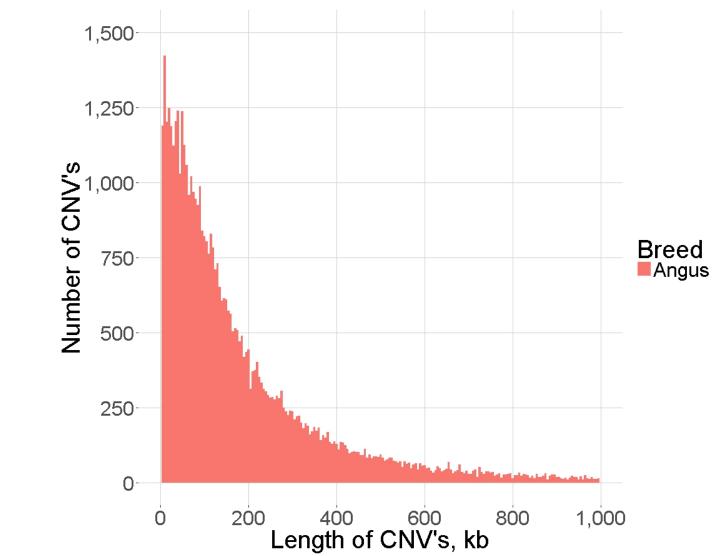






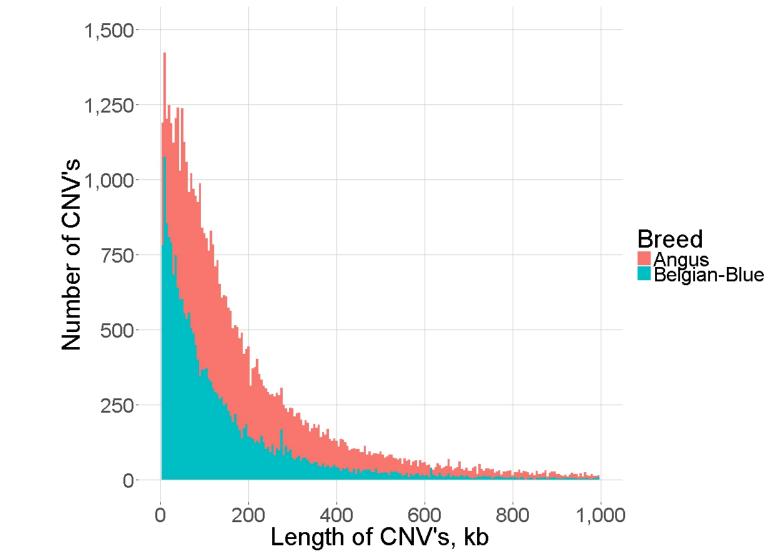






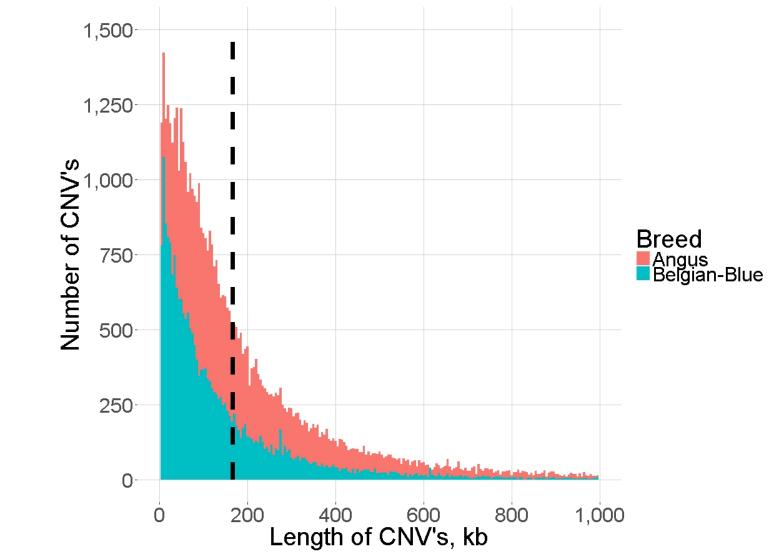






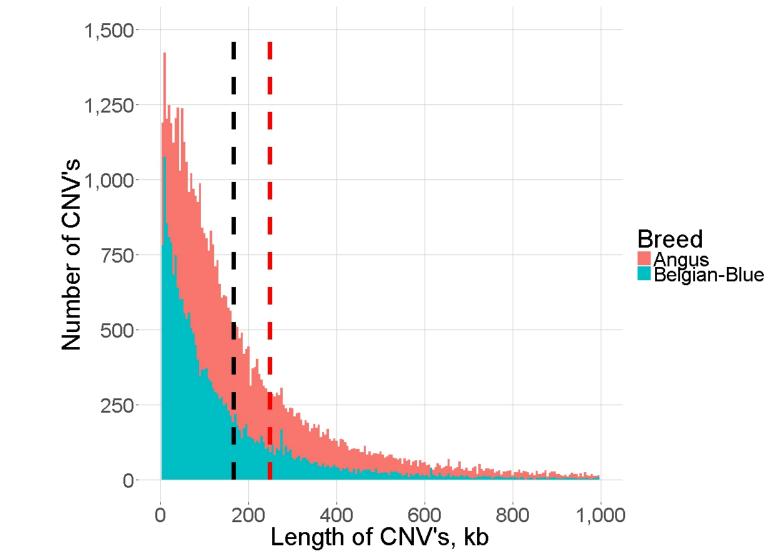








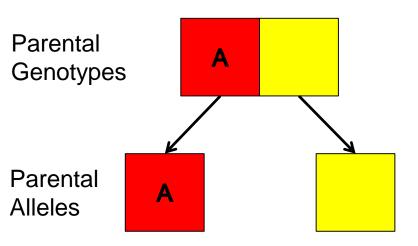


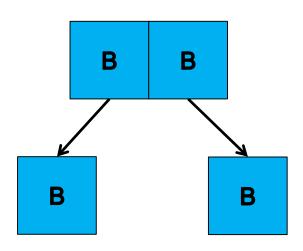


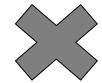




Non-Mendelian Inheritance







Progeny Genotypes





Observed Progeny Genotypes









Conclusions

- For most animals a small proportion of their genome were composed of CNVs.
- Small differences did exist between some breeds for mean length of CNVs, and mean number of CNVs per animal.
- The presence of CNVs can give rise to apparent Mendelian inconsistencies in SNP genotype data.



