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

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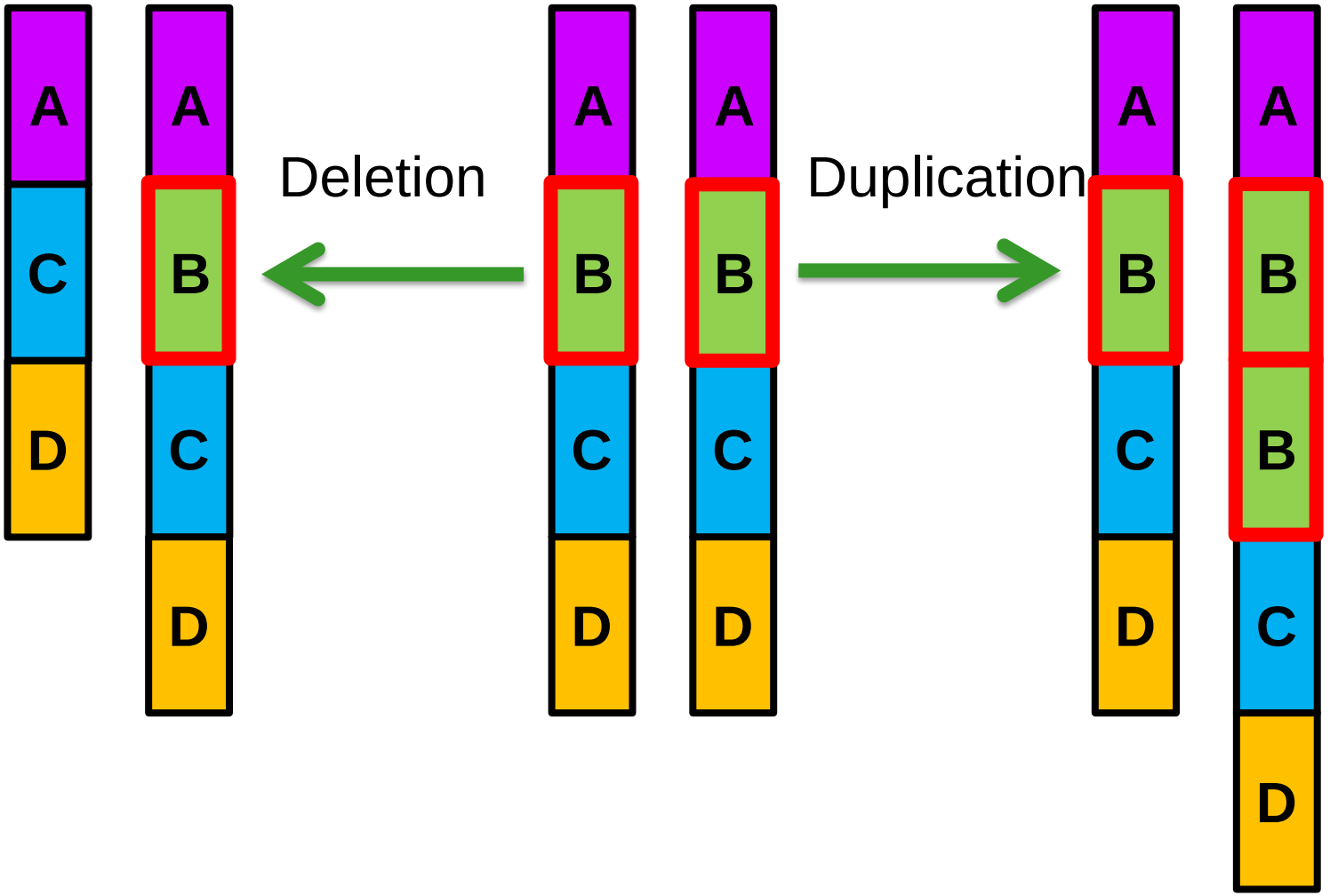
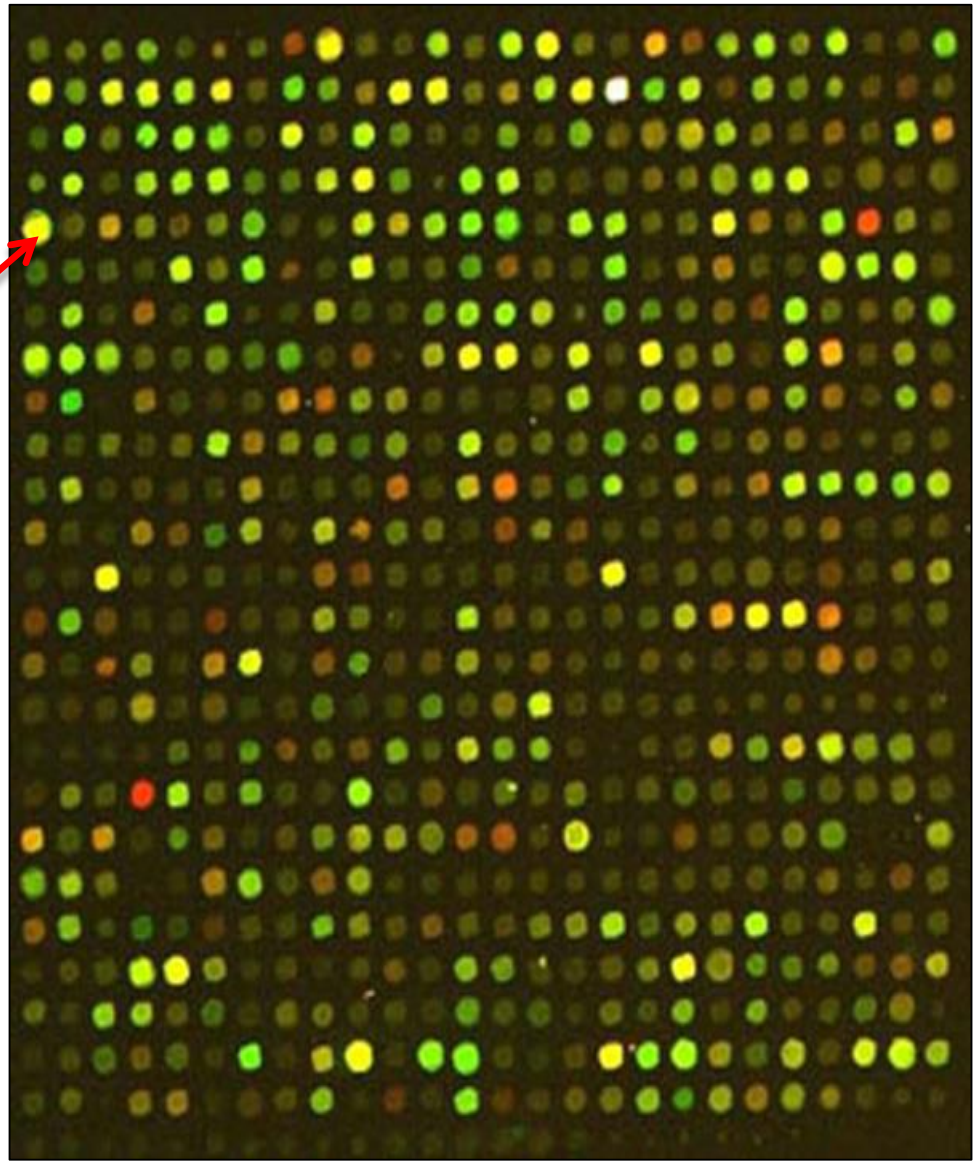
What is a copy number variant (CNV)?



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



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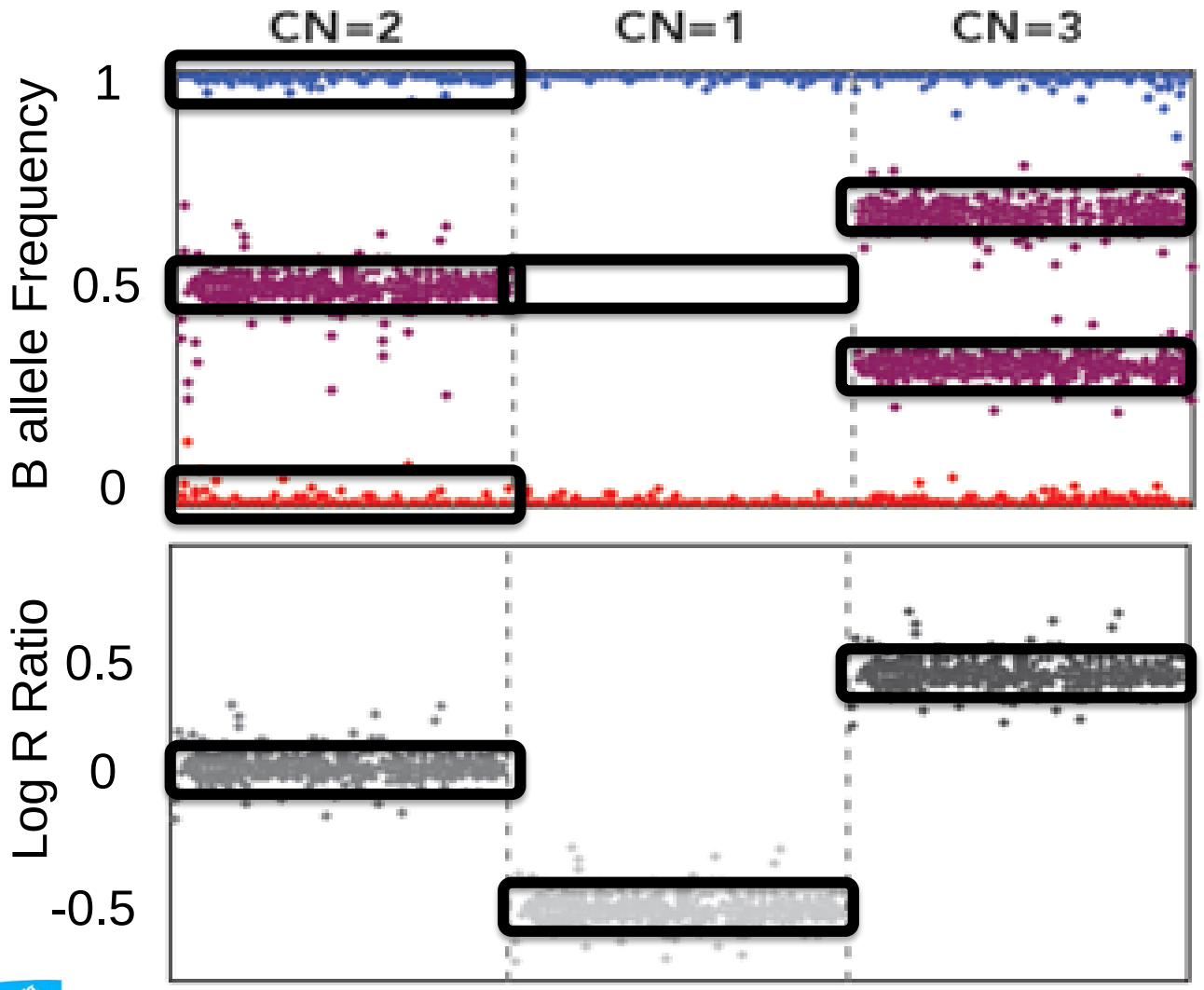
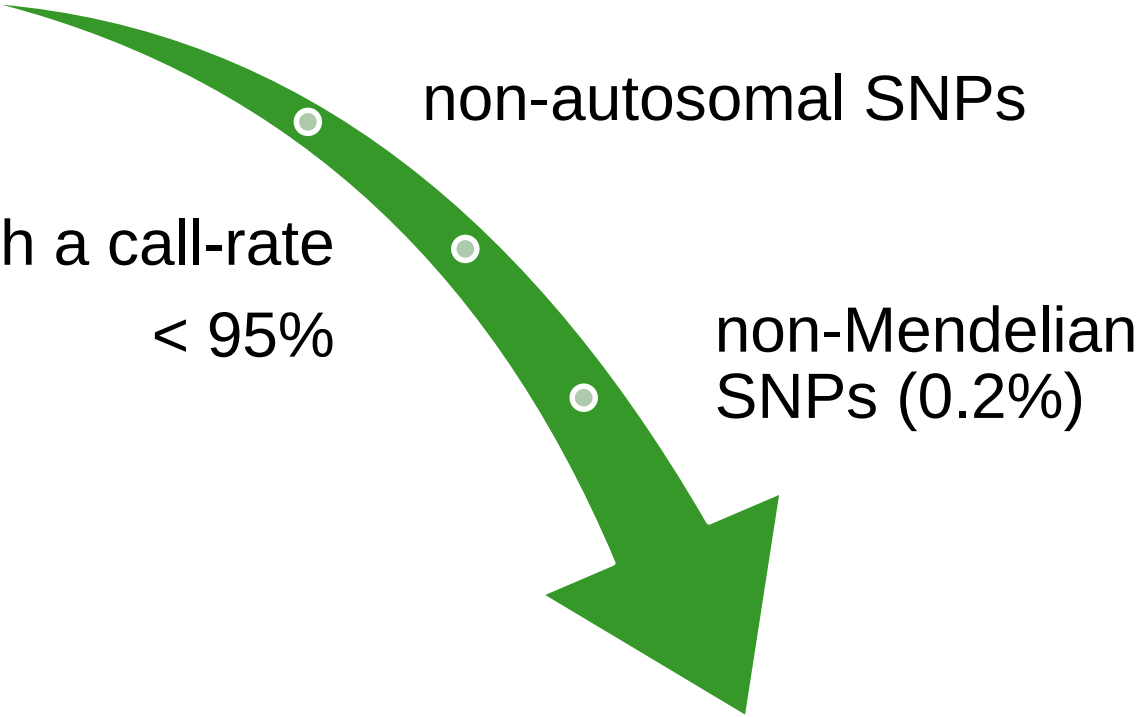
Copy number analysis



Genotype data

Illumina BovineHD, 777,962 SNPs



713,162 SNPs

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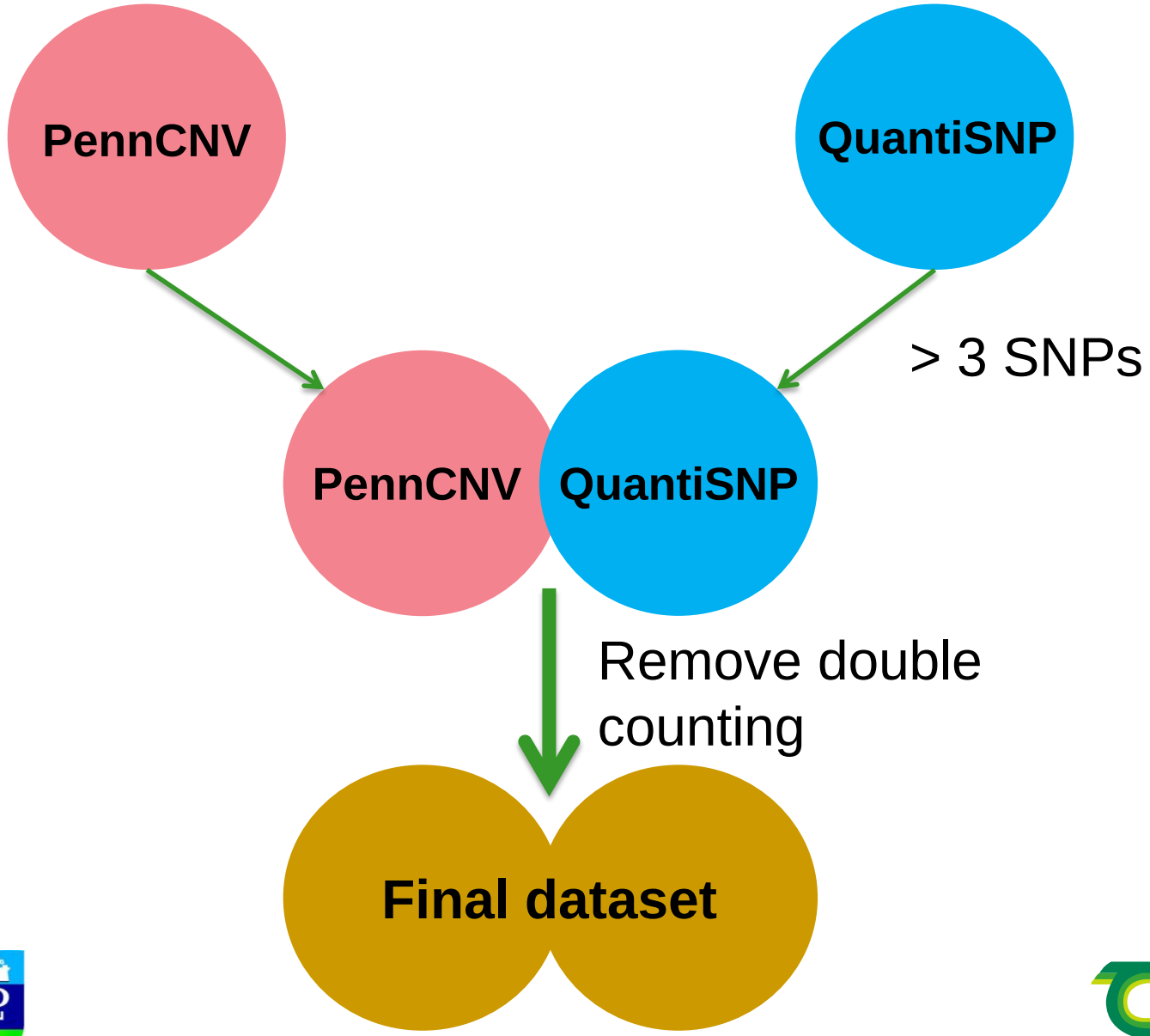
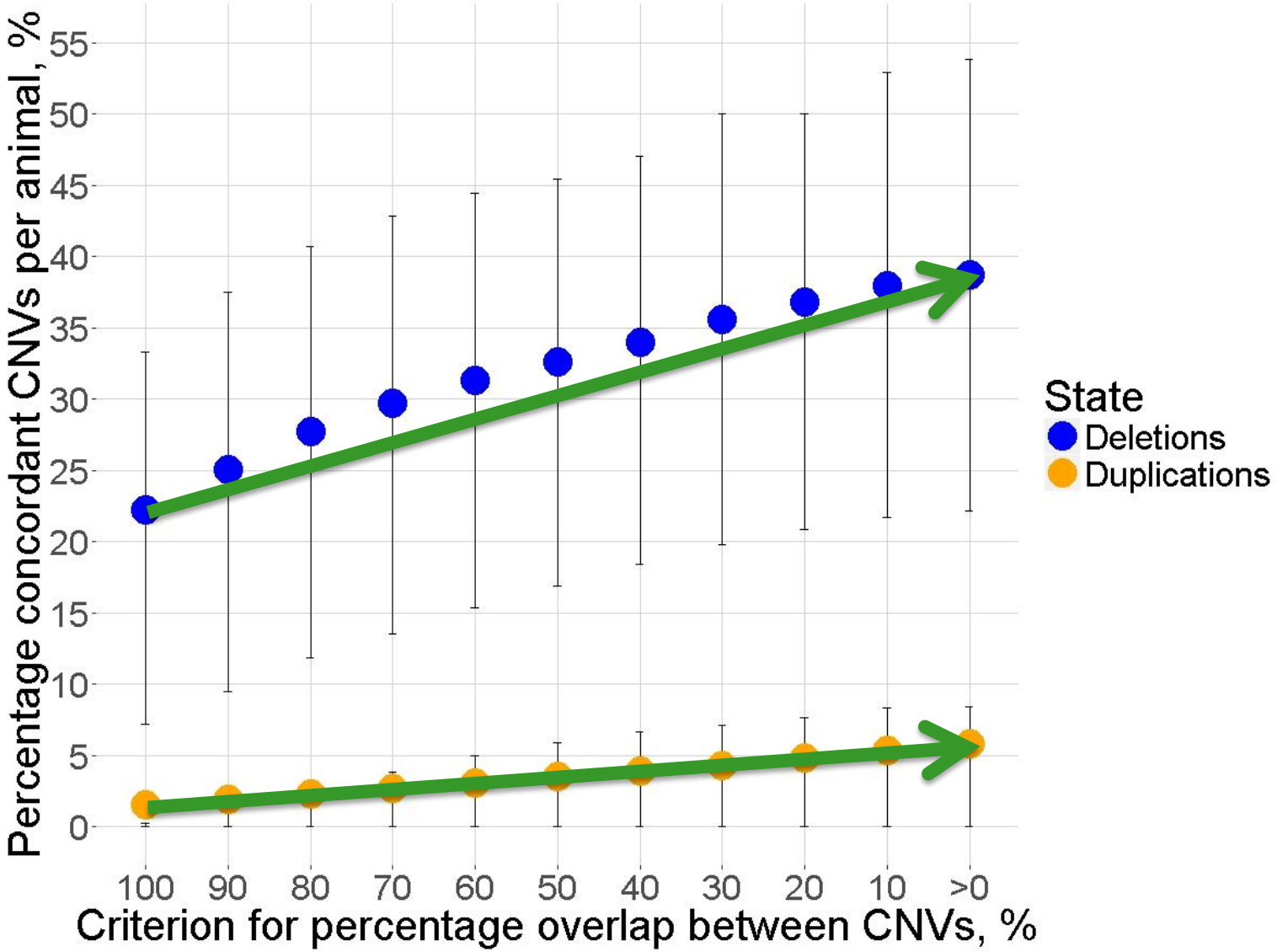
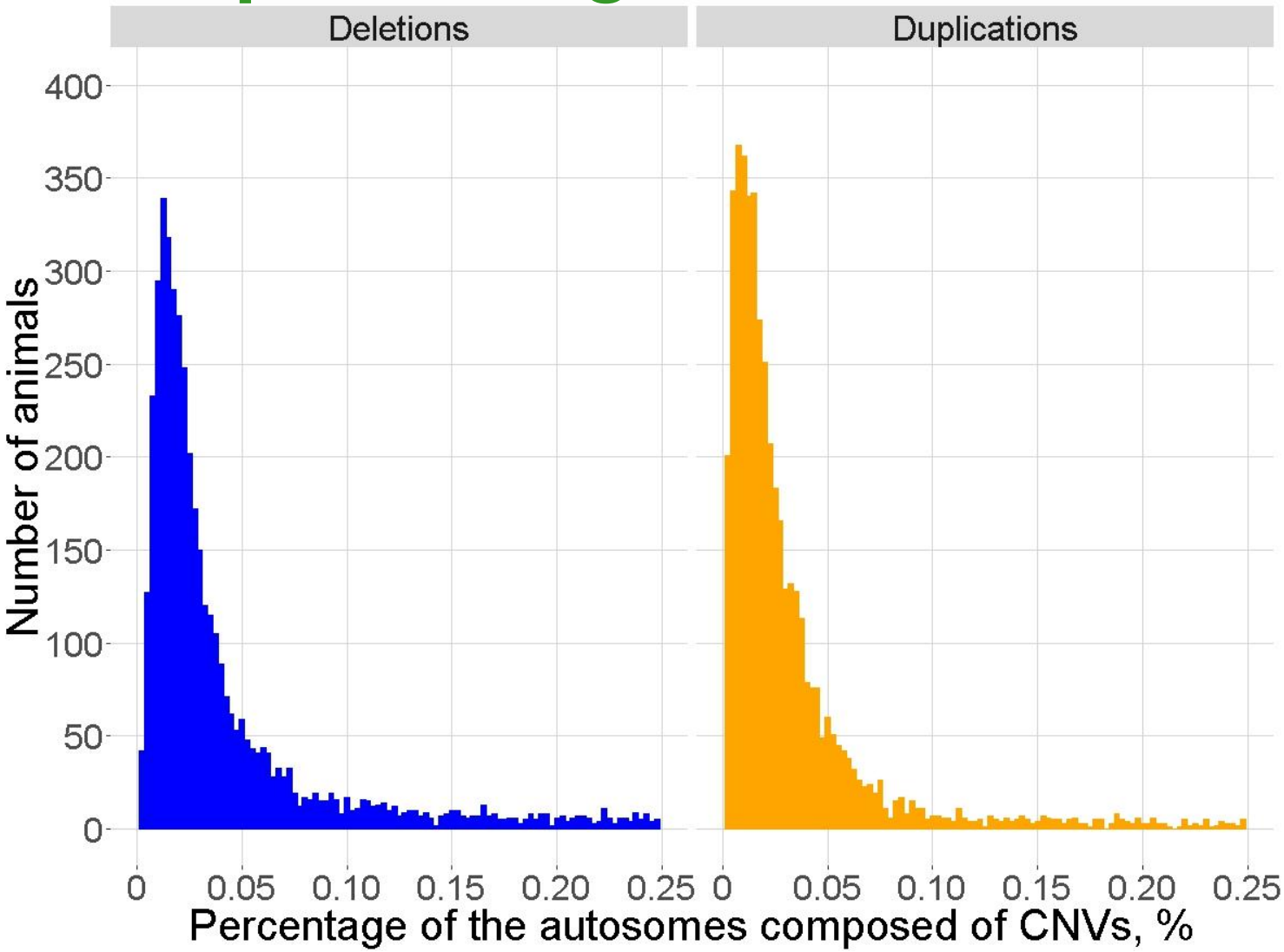
The CNV dataset



Concordance between PennCNV and QuantiSNP



CNV percentage of autosomes



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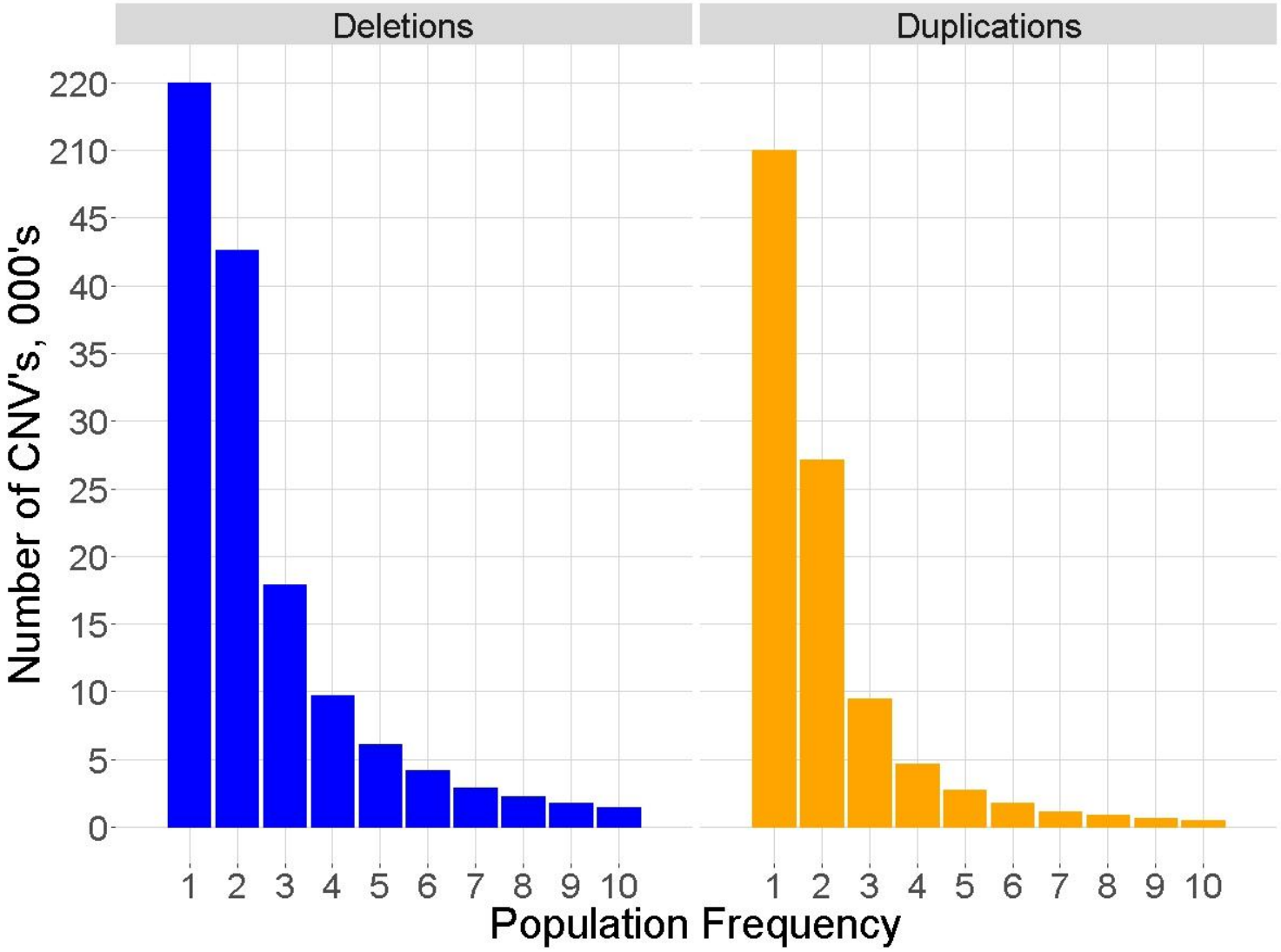
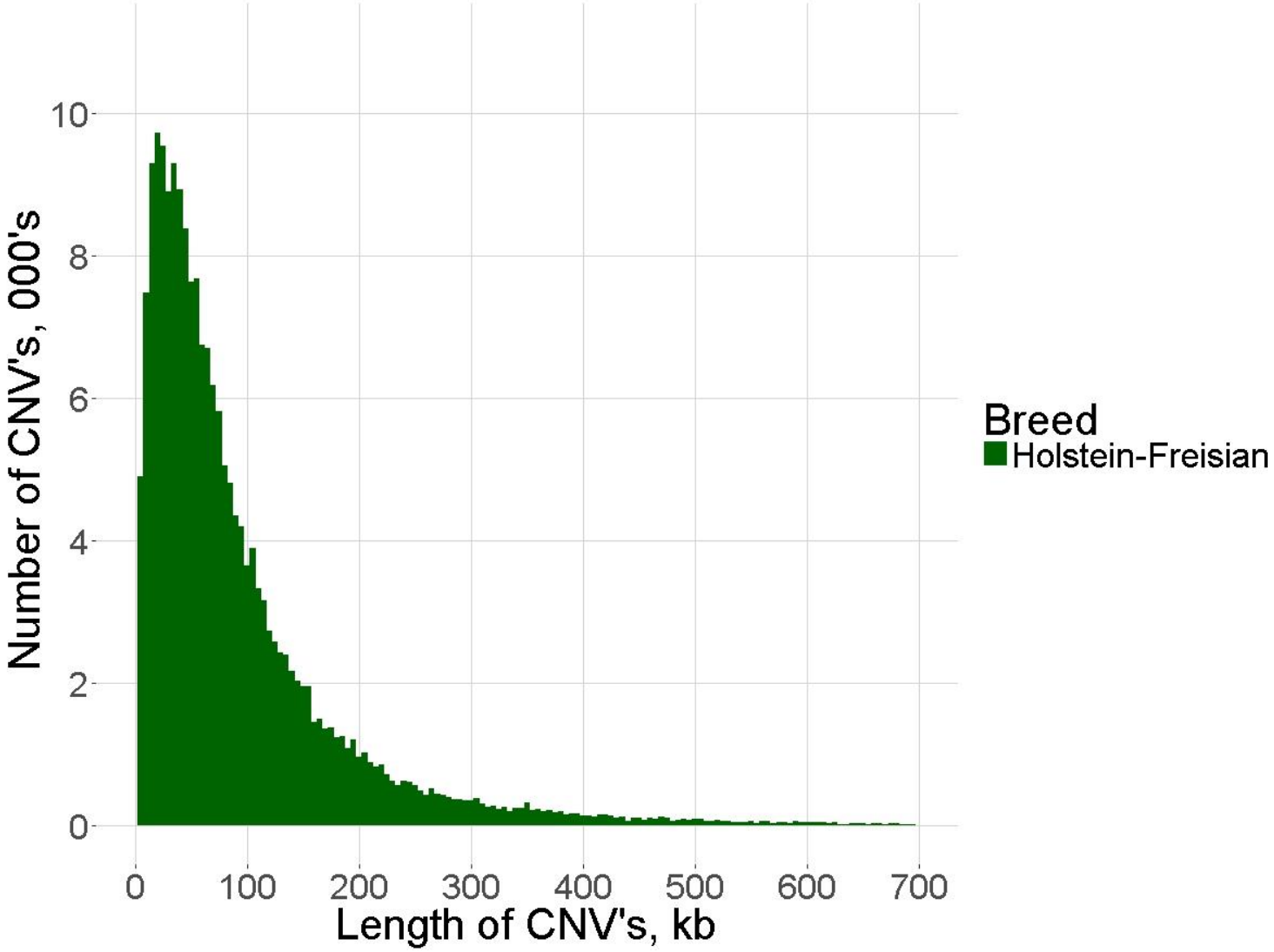
Frequency of CNVs



Breed comparison – deletion length



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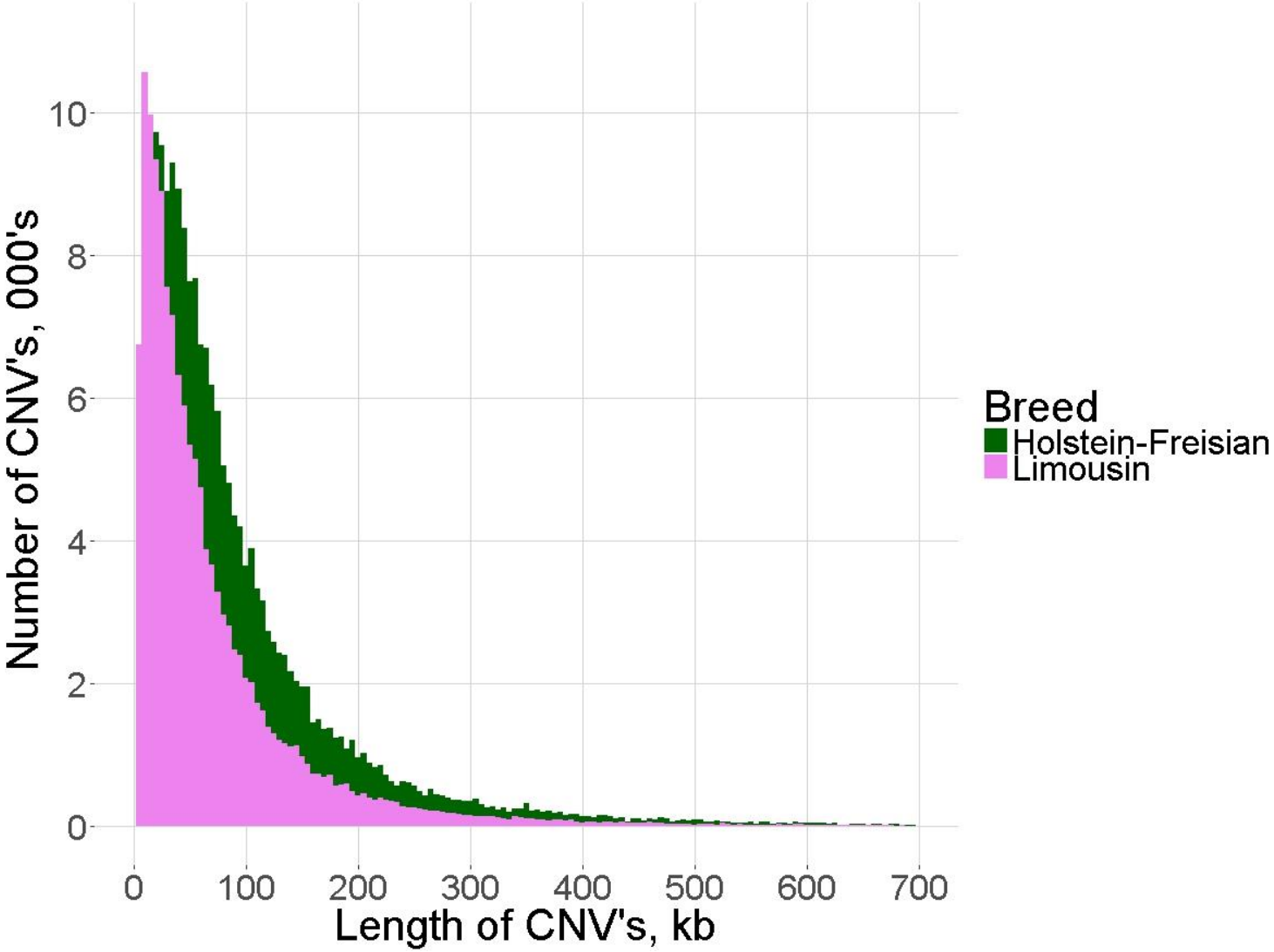

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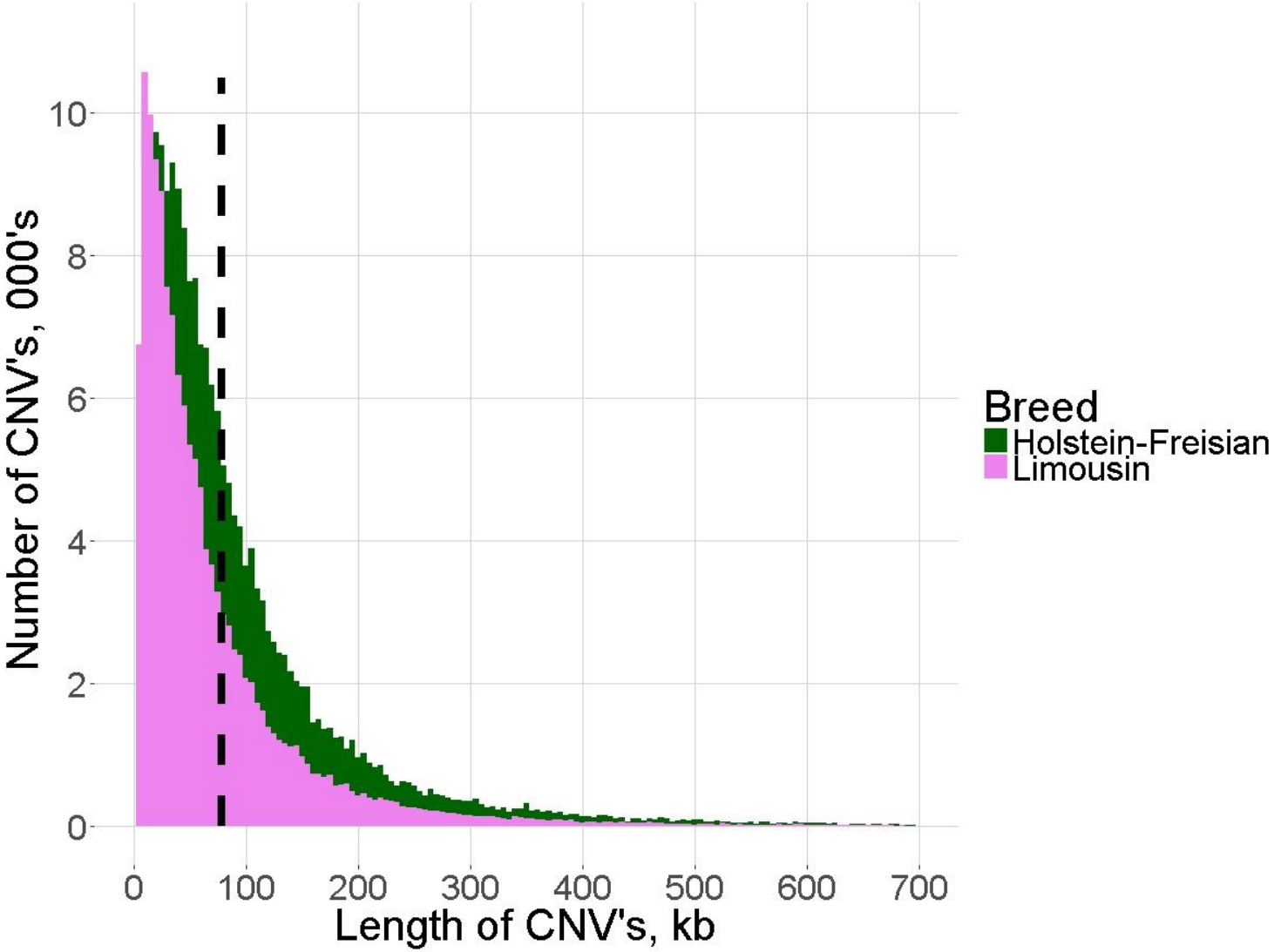
Breed comparison – deletion length



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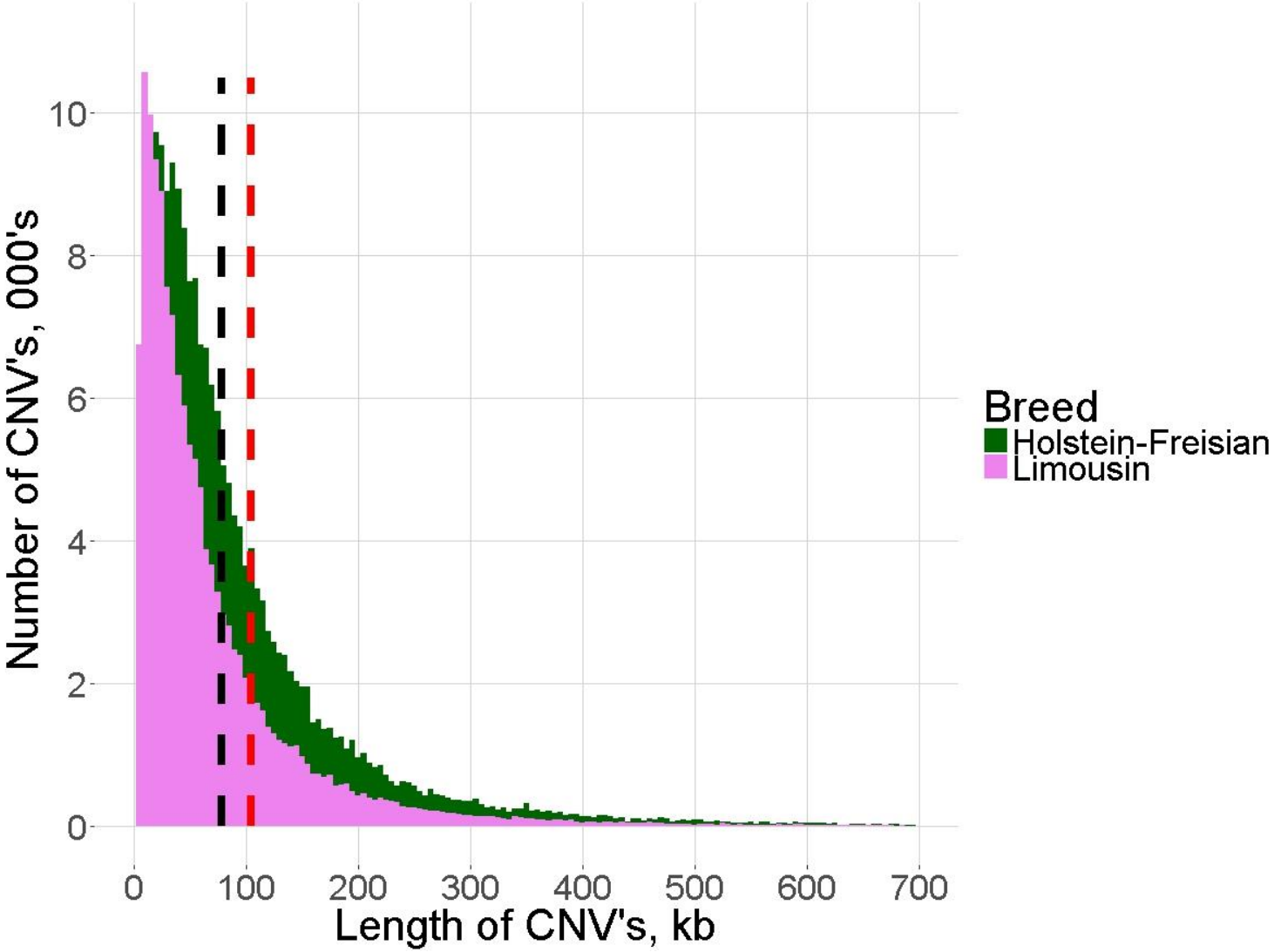
Breed comparison – deletion length



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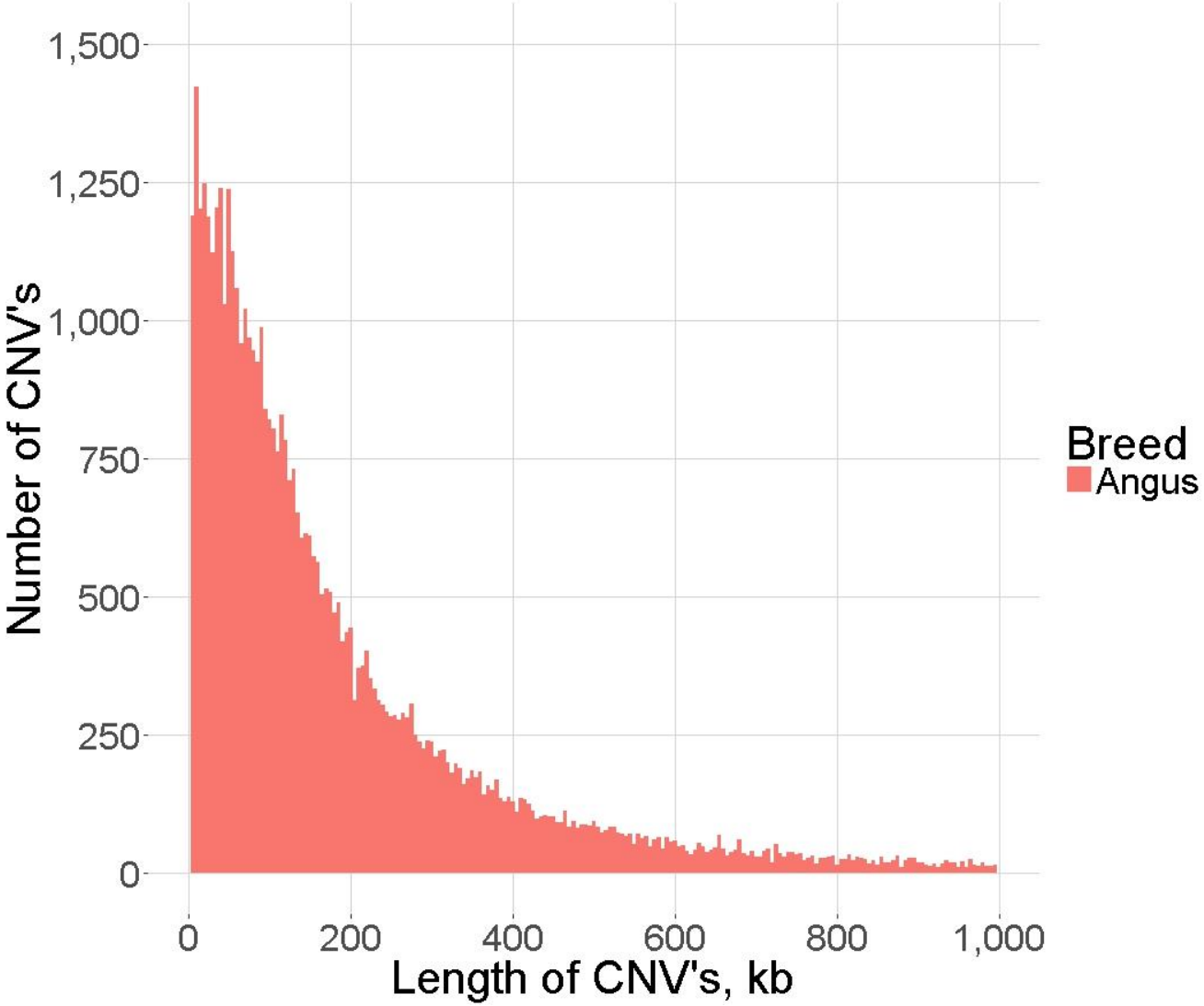
Breed comparison – duplication length



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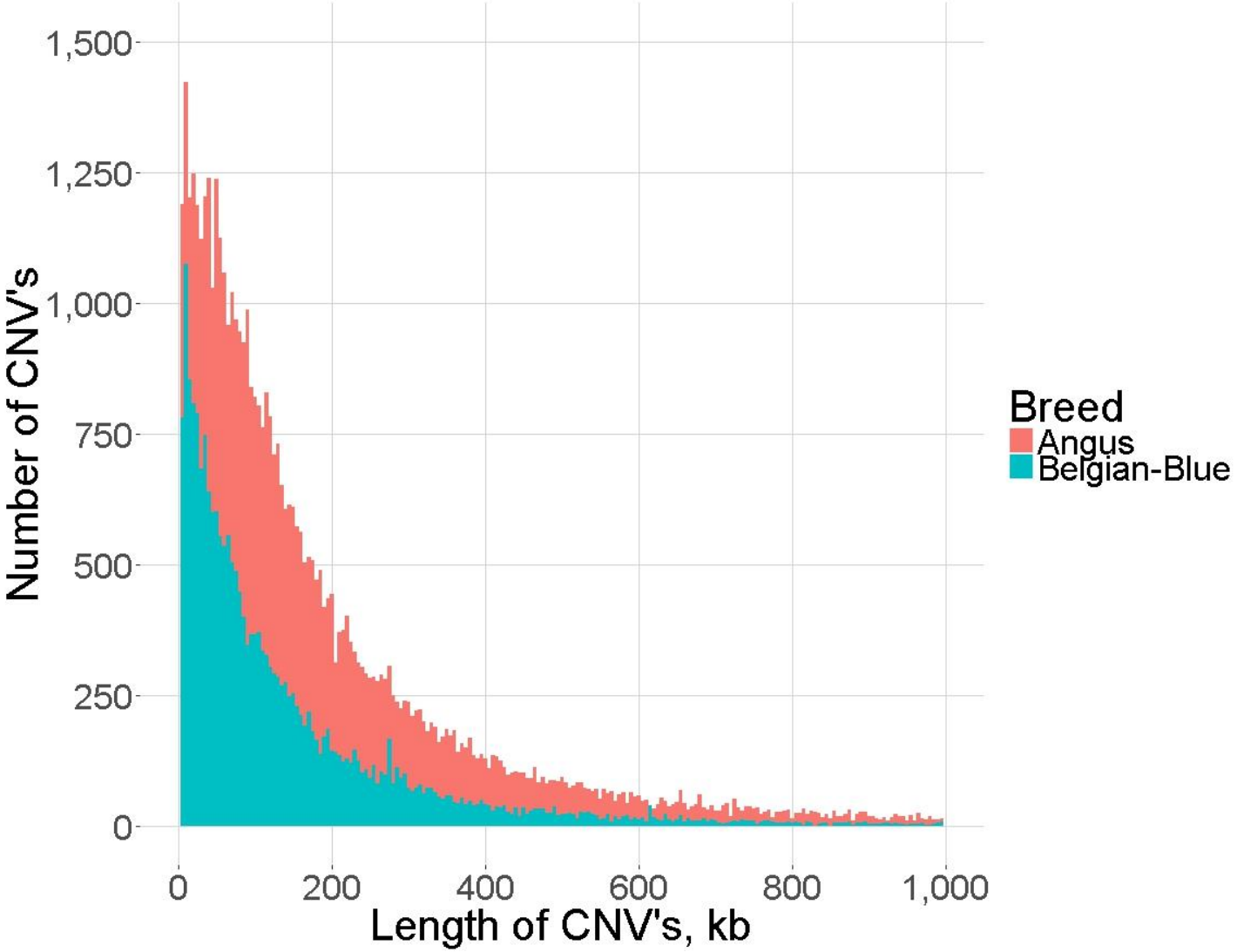
Breed comparison – duplication length



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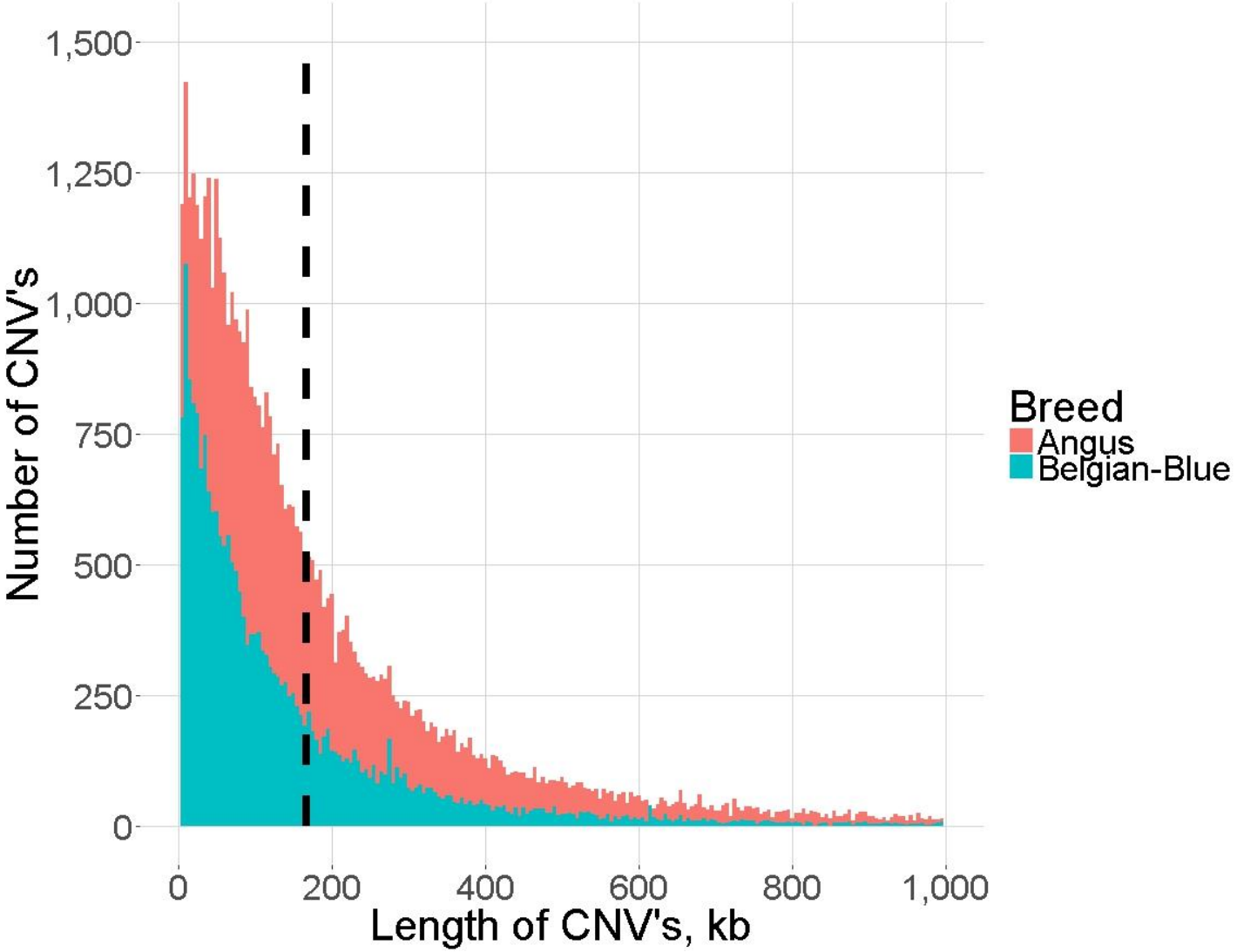
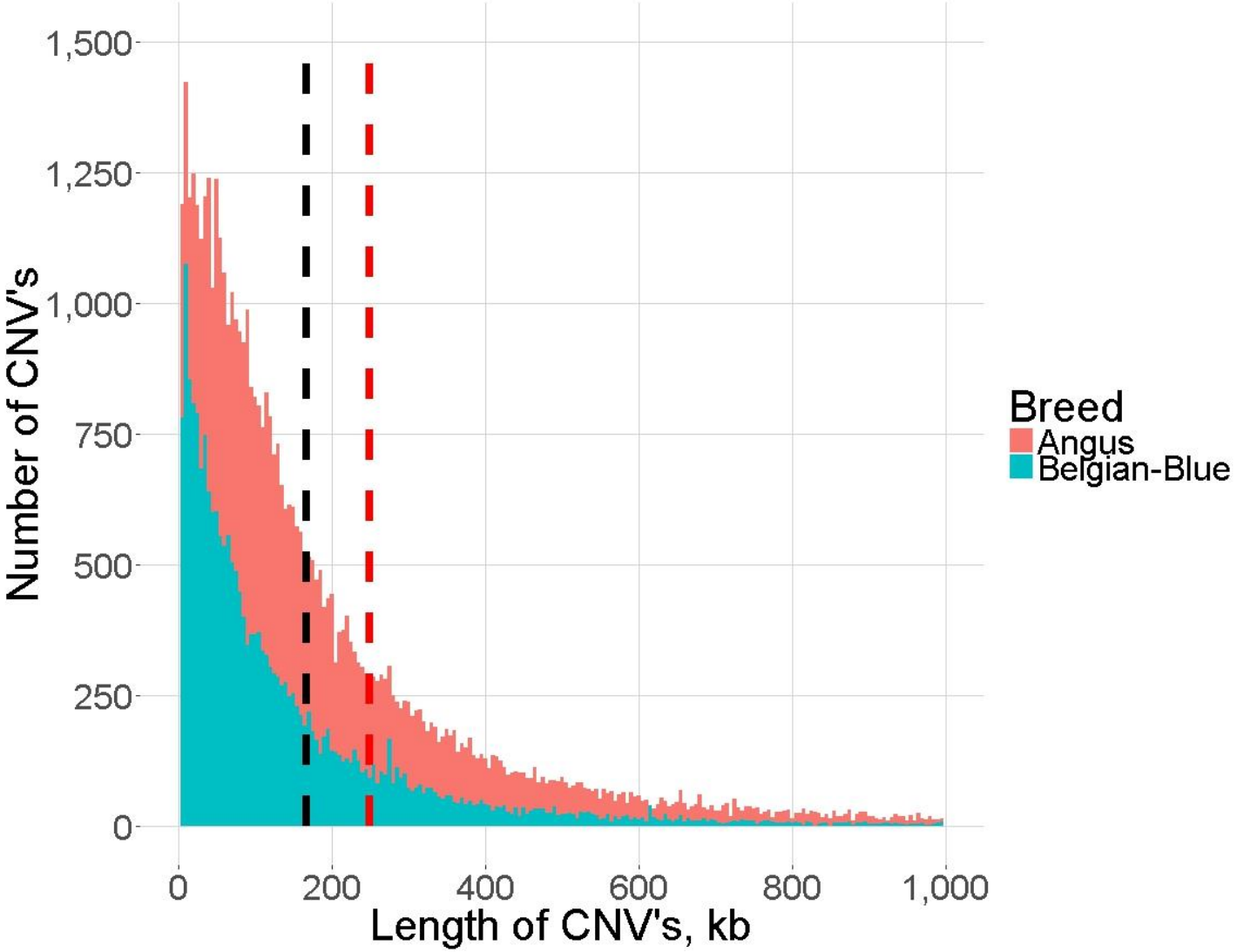
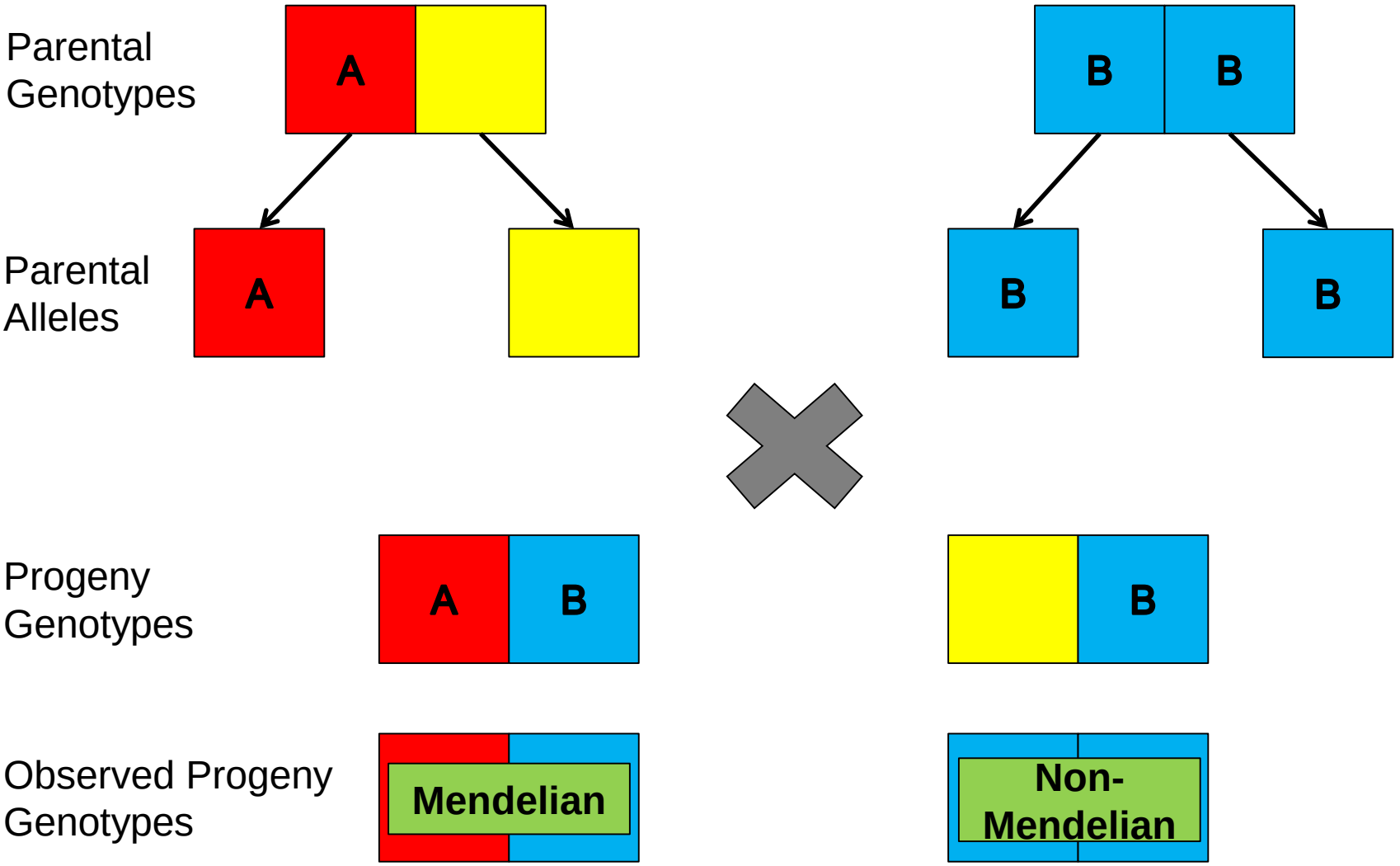


Breed comparison – duplication length



Non-Mendelian Inheritance



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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.