Optical mapping compendium of structural variants across global cattle breeds

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Abstract

Structural variants (SV) have been linked to several important bovine disease phenotypes, but due to the difficulty of their accurate detection with standard sequencing approaches, their role in shaping economically important traits across global cattle breeds is largely unexplored. Optical mapping is an alternative approach for mapping SVs that has been shown to have higher sensitivity than DNA sequencing approaches. The aim of this project was to use optical mapping to develop a high quality database of structural variation across cattle breeds from different geographical regions and origins, to enable further studies of the important roles of SV in cattle.

To do this we generated 100X Bionano optical mapping data for 18 cattle of nine different ancestries, three continents and covering both major cattle lineages. In total we identified 13,457 SVs, of which 1,200 putatively overlap a coding region. This resource therefore provides a high-quality set of optical mapping-based SV calls that can be used across studies, from validating DNA sequencing based SV calls to prioritising candidate functional variants in genetic association studies and expanding our understanding of the role of SVs in the evolution of cattle.

52 # Background & Summary

53 Structural variants (SV) are a heterogeneous class of genetic variants involving large
54 fragments of the genome (>50bp)[1]. These variants include genomic insertions and deletions
55 (InDels), inversions, duplications, translocations and more complex rearrangements[2]. Single
56 nucleotide polymorphisms (SNPs) have been the primary focus of studies trying to map
57 genetic loci underlying important cattle phenotypes. However, there are multiple lines of
58 evidence suggesting SVs likely underlie many important cattle traits. As many as 25-29% of all
59 protein truncating events are thought to be caused by an SV in humans[1] and notably, despite
60 being less well studied, SVs have already been tied to key livestock phenotypes. For example,
61 a duplication of the CIITA class II major histocompatibility complex transactivator gene in
62 cattle has been tied to resistance to intestinal nematodes[3] and a 12Kb copy number variant
63 has been linked to mastitis in cattle[4]. Chromosomal translocations and duplications have been
64 linked to skin pigmentation, a phenotype closely tied to environmental adaptation, and SVs
65 across livestock species have been linked to phenotypes such as olfaction or resistance to
66 adenocarcinoma-causing viruses[5]. Importantly SVs are responsible for approximately 5-10
67 times as many heritable nucleotide sequence differences between individuals than SNPs[6].
68 Unlike SNPs, that only effect a single basepair, and most often far from coding regions, SVs
69 effect large regions and potentially multiple genes. Consequently, although smaller in
70 number, any given novel SV event is more likely to have a phenotypic consequence.
71 The two most popular methods used to detect SVs are high-throughput sequencing (HTS) and
72 array comparative genomic hybridisation (aCGH), both of which have been applied to
73 European cattle[7–9], but with few studies performed in other cattle breeds[10–12]. Each
74 technology has advantages and limitations. aCGH, for example, involves measuring binding
75 to probes covering the reference genome, and therefore it can only detect relative copy
76 number changes between sample pairs and cannot for example detect novel insertions.
77 Resolution is also limited. A major advantage of HTS approaches is that theoretically they can
78 detect SVs at base-pair resolution. However, accurate calling of SVs from HTS data has proven
79 to be difficult for a number of reasons including poor reference assemblies, chimeric reads,
80 aligners penalising reads that don't match the reference and the difficulties of sequencing
81 and mapping to repetitive regions. This is exemplified by the generally poor agreement
82 between SV callers even when run across the same samples[13,14]. Approaches using long reads

and *de novo* assembly can still have true positive rates as low as 77%, even when using simulated data[15].

Optical mapping (OM), a light microscope-based method that labels and physically locates specific motifs in the genome[16], offers an alternative protocol to accurately detect large SVs. OM molecules can be consistently hundreds of Kb long, allowing for the detection of complex rearrangements undetectable using HTS. Despite the limitation of not being able to detect the actual sequence of the identified SVs, as well as missing smaller SVs, OM has a very high sensitivity and specificity, allowing for the generation of high-quality catalogues of SVs in individuals[17]. A study in humans successfully used OM reads to identify SVs in a total of 26 genomes revealing population-specific patterns of structural variation[18].
In this study, we generated the first catalogue of cattle OM data for 18 animals from 9 different global breeds, and three continents, to better characterise common SVs across the cattle pan-genome. This data is a particularly valuable resource of SVs for the cattle species to intersect with other datasets, for example, for the validation of SV calls from other approaches.

Methods

Sample preparation

We selected a set of 18 cattle across 9 divergent European, African and Indian breeds representative of Indicine, Sanga and Taurine ancestries (Table 1). Blood was collected by jugular venipuncture into EDTA vacutainers. Somatic recombination in B cells and T cells means the Ig and TCR loci in these cell types will be highly heterogenous, confounding accurate reconstruction of germline SVs at these loci from whole blood samples. Consequently, after the erthyrocyte lysis, monocytes were purified from the leukocytes using a MACS positive selection protocol with an anti-bovine SIRPα mono-clonal antibody (ILA-24[19]). Agarose plugs containing 5 $x10^5$ – $1x10^6$ of isolated monocytes were prepared using the Bionano Blood and cell culture DNA isolation kit (Bionano Genomics, San Diego, US) according to the manufacturer's instructions and the extracted DNA used for analysis on the Bionano Saphyr platform to generate ~100X optical mapping coverage of each genome.

Bionano Solve optical mapping processing

OM reads were filtered using the filter_SNR_dynamic.pl script with default parameters included with the Solve workflow, and then processed through the Bionano Solve[20] pipeline (v3.3_10252018) using two different releases of RefAligner to overcome bugs preventing the successful assembly of the reads (version 7915.7989rel and 10330.10436rel). We generated the reference CMAP for the ARS-UCD1.2 genome with the Y chromosome from the 1000 bulls genome project (https://sites.ualberta.ca/~stothard/1000_bull_genomes/) using fa2cmap_multi_color.pl (default options and specifying the DLE1 as enzyme). The resulting data were imported into the Bionano Access (v1.6) software, and single-sample SVs were filtered using the recommended thresholds for SVs generated using Bionano Solve prior to v1.6.0 with the sizes recommended to achieve 90% sensitivity (see https://bionanogenomics.com/wp-content/uploads/2018/04/30110-Bionano-Solve-Theory-of-Operation-Structural-Variant-Calling.pdf): minimum insertion size of 5Kb, minimum deletion size of 5Kb, minimum inversion size of 100Kb, and minimum duplication size of 150kb.

Filtered smap format files were converted to vcf format using smap_to_vcf_v2.py and sorted with bcftools (v1.10.2[21]). The resulting SVs were screened using bcftools and retained if 1) they had successfully been genotyped, 2) their size was >1Kb and 3) their quality was >= 20. The latter filtering largely removed all translocations, duplications, and complex events due to these having either very low (<1) or nil quality values.

We then defined the total amount of non-redundant reference sequence involved in a high-quality deletion. For each deletion, we calculated the central point in the genomic region affected by the SV:

$$Center = \frac{POS + abs(CIPOS) + END - abs(CIEND)}{2}$$

Where POS is the initial position, END is the end position, CIPOS is the confidence interval of POS and CIEND is the confidence intervals of END. Having defined the central point of the region, we defined the initial and final positions of the SV as:

$$BPI = Center - \frac{abs(SVLEN)}{2}; BPE = Center + \frac{abs(SVLEN)}{2}$$

Where BPI and BPE are the limits of the SV and SVLEN is the size of the SV.

141 We then concatenated the regions for all the individuals, sorted them and merged them using
142 bedtools sort and bedtools merge[22] to remove any redundancies among the regions.
143 Following filtering, we merged the resulting variants within samples using SURVIVOR (v1.0.7[2])
144 accounting for the SV type and collapsing those whose break points were within 1kb. We
145 represented the intersection of SVs across individuals by extracting the support vectors
146 generated by SURVIVOR[2] at merging time, and plotted them using the UpSet function from
147 the R[23] package ComplexHeatmap[24] (v2.8.0). We extracted the support value (i.e. how many
148 animals present a specific SV) and SV size for each variant in the combined VCF and tested
149 whether the SVs found in one individual only (support = 1) were significantly larger than those
150 shared among individuals (support > 1) by performing a Wilcoxon signed-rank test in a custom
151 R script.
152 Finally, we defined which of the final set of SVs were found to potentially affect a gene. We
153 ran VEP v105[25] to predict which SVs were likely to disrupt a gene's function, with the options
154 --sift b (both preditions score and term), --nearest symbol (report the gene symbol), and --
155 distance 200 (200 bp up and downstream consequence prediction). Those variants presenting
156 coordinates referring to the negative strand (end position smaller than initial position) were
157 manually fixed through an in-house script. We then investigated which SVs putatively overlap
158 a coding region annotated in the cow genome by intersecting merged SVs with coding
159 sequence intervals. Intersecting genes were investigated with FUMA[26] to identify enriched
160 gene ontologies and gene sets using all 35,142 gene elements with a unique Entrez gene ID
161 as the background list.
162

Data Records

The datasets presented here are stored at ENA under analysis IDs PRJEB47998 and PRJEB52551. The data are uploaded in Bionano BNX format compatible with downstream analyses. The output of the Solve workflows can be downloaded from Zenodo (DOIs: 10.5281/zenodo.6516993 and 10.5281/zenodo.6517172). The raw VCF files, converted using smap_2_vcf_v2.py, can be found on Zenodo with DOI: 10.5281/zenodo.6516933.

Technical Validation

Assembly statistics and SV calling

We aligned the Saphyr optical mapping reads to the ARS-UCD1.2 genome[27], expanded with the BTau5 Y chromosome generated by the 1000 bulls genome project, using Bionano Solve (v3.3 and 3.5) to assemble the genome maps and call SVs.

Workflow metrics are provided in Supplementary Data 1, summarising key metrics for the analysis of these samples in comparison to the recommended values from Bionano (see https://bionanogenomics.com/wp-content/uploads/2017/03/30255-Bionano-Access-Assembly-Report-Guidelines.pdf).

Unfiltered molecules had average read lengths of 131.9-219.8kb (recommended >150kb for both metrics) and molecule N50s ranged from 185.2-361.9kb across the samples (recommended >150Kbp). Following molecule filtering, all samples were within the recommended average length (245.5-383.1, recommended >230Kbp) and molecule N50 (245.0-426.5, recommended >230Kbp), and only 1 sample (Angoni 1) was slightly below the recommended label density (13.1-16.4, recommended 14-17). Importantly all samples passed the recommended values for the effective coverage of the reference (72.5-128.2, recommended >70) and of average confidence (30.1-43.2, recommended >20).

All samples also generated assemblies with high genome map N50s for both the diploid (71.7-85.0, recommended >50) and haploid (71.3-84.5, recommended >50) assemblies. Despite the low proportion of assembled contigs aligning to the reference genome (0.14-0.25, recommended >0.70), the high uniquely aligned length by reference length (0.835-0.906, recommended >0.85) shows the presence of long assembled contigs. The contigs present a

high fraction of molecules aligned (0.77-0.94, recommended >0.80), effective coverage assembly (37.7-66.7, recommended >40) and average confidence (38.5-51.3, recommended >20).

Overall, 1 sample had 11 metrics within the recommended values, 6 had 12 metrics within the recommended values, 9 had 13 metrics within the recommended values and 2 had 14 metrics within the recommended values.

The Bionano Solve workflow identified a number of SV in each sample, ranging from 4,944 to 11,184 for a Hereford and Nelore, respectively (Table 2). This mirrors the evolutionary distance of each sample from the reference genome, with the European taurine possessing fewer SVs (4,944-5,652) than the other samples, and an African taurine N'Dama possessing the least among the non-European individuals (N = 6,254). Relative SV numbers consequently broadly mirror prior expectations.

Variant statistics

SVs were filtered using Bionano Access, excluding SVs with unknown dosages, and retaining those larger than 1Kb and with a quality >20. SVs for each individual were then combined using SURVIVOR[2] if the breakpoints were within 1Kb, i.e. below the effective resolution of the approach[2]. This process allowed us to select a catalogue of 13,457 SVs across the genome, containing 8,262 insertions, 5,191 deletions and 4 inversions (see Supplementary Data 2 and 3 for the details on the type of SV identified). No duplications, inverted duplications and translocations passed the quality filtering. Consistent with results from previous studies[28], most of the post-filtering insertions and deletions identified fell into the smaller classes, though 1,796 SVs (403 deletions, 1,389 insertions and 4 inversions) of over 50kb in length were identified (Figure 1).

These SVs longer than 1Kb and of high quality involve a total of 2,656 unique regions, for an estimated total of over 90Mb of non-redundant bases (Supplementary Data 4). This number is comparable to what has been seen for novel sequences (i.e. insertions) using graph genome approaches, where an extra 70Mb and 116Mb of novel sequence were reported on 5 and 4 cattle reference genomes, respectively[28,29]. After merging the filtered variants from all the samples, most of the SVs were found to be private to an individual (Figure 2), consistent with

222 what has been observed in previous studies[1]. Individuals of indicine ancestry (Nelore and
223 Boran) carry almost twice as many SVs relative to the Hereford reference as taurine
224 individuals, further highlighting that the current reference poorly represents these breeds
225 (Figure 2).
226 Interestingly, we find that SVs only found in one animal (support = 1; n = 7,445, mean SV
227 length = 85,954.23bp) are generally larger (Wilcoxon rank-sum test P-value = $8.99 * 10^{-37}$)
228 than the SVs found in more than one animal (support > 1; n = 6,012, mean SV length =
229 27,747.17bp, Figure 1, Supplementary Figure 1). The list of all SVs, with their position, support
230 and size, are reported in Supplementary Data 5.
231 Finally, we investigated whether any of the high-quality SVs potentially impact annotated
232 genes. VEP successfully processed 12,999 out of 13,457 SVs (see HTML report on GitHub).
233 Some variants were too large to be successfully processed by VEP, and other were called as
234 incomplete by VEP. Of these, 6,946 were intergenic, and the remaining 5,934
235 overlapped5,780 genes and 17,386 transcripts, suggesting the potential for functional
236 variants among the SVs detected. A total of 1,200 SVs putatively overlap a coding sequence.
237 These coding sequences are included in a total of 884 unique gene elements in the cow
238 annotation (Ensembl v105), and of these 483 have an associated gene name (Supplementary
239 Data 6). A total of 292 out of 483 genes had an ID recognized by FUMA[26]. These 292 genes
240 belong to a number of gene sets such as the Hallmark bile acid metabolism and interferon γ
241 and α response sets (Supplementary Figure 2), as well as the olfactory receptor curated gene
242 set (Supplementary Figure 3). All gene set results from FUMA are reported in Supplementary
243 Data 7.

244 Usage Notes

245 Even with the ever-decreasing cost of long read sequencing making it increasingly tractable
246 to call SVs across sets of samples using HTS, validation of the SV calls remain challenging. This
247 compendium of SVs across global cattle breeds provides a validation set called using an
248 independent technology that can be used to assess the quality of cattle SV calls. In fact, optical
249 mapping data has previously been used to validate sequencing based SV calls[28], and we
250 believe this dataset provides the largest set of optical maps to date for a livestock species.

With many SVs shared across the two animals of each breed, the raw molecules in this dataset can also be used to help scaffold and validate novel assemblies of cattle of breeds closely related to the individuals represented here, potentially reducing the cost of future genome assembly projects.

Unlike most cattle studies, this database is not focused just on European cattle breeds, meaning this will be a valuable resource to researchers across the globe. Importantly, it will allow for SVs to inform the interpretation of results from GWAS and population genetics studies by providing candidate functional variants in relevant regions.

Ultimately, we expect the database to enable further insights into SVs, an understudied class of genetic variation in cattle, giving access to a catalogue of thousands of variants present across multiple breeds worldwide.

Code availability

The codes used in this article were deposited in https://github.com/evotools/CattleOManalyses.

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Author contribution

J.G.D.P. and T.C. conceived the study, A.T. and J.G.D.P. designed the analyses and A.T. performed them. J.G.D.P. and A.T. prepared the initial manuscript with all authors contributing to subsequent drafts. J.Po., M.C. and T.C. prepared the DNA and Bionano samples. D.W. and P.T. provided samples and expertise for the study. M.E.Z.M., I.M.S., A.F., B.R.F., C.E., E.T.O., D.M., W.A., I.S., J.B.M., G.M. and R.F.K provided samples for the study.

Figure captions

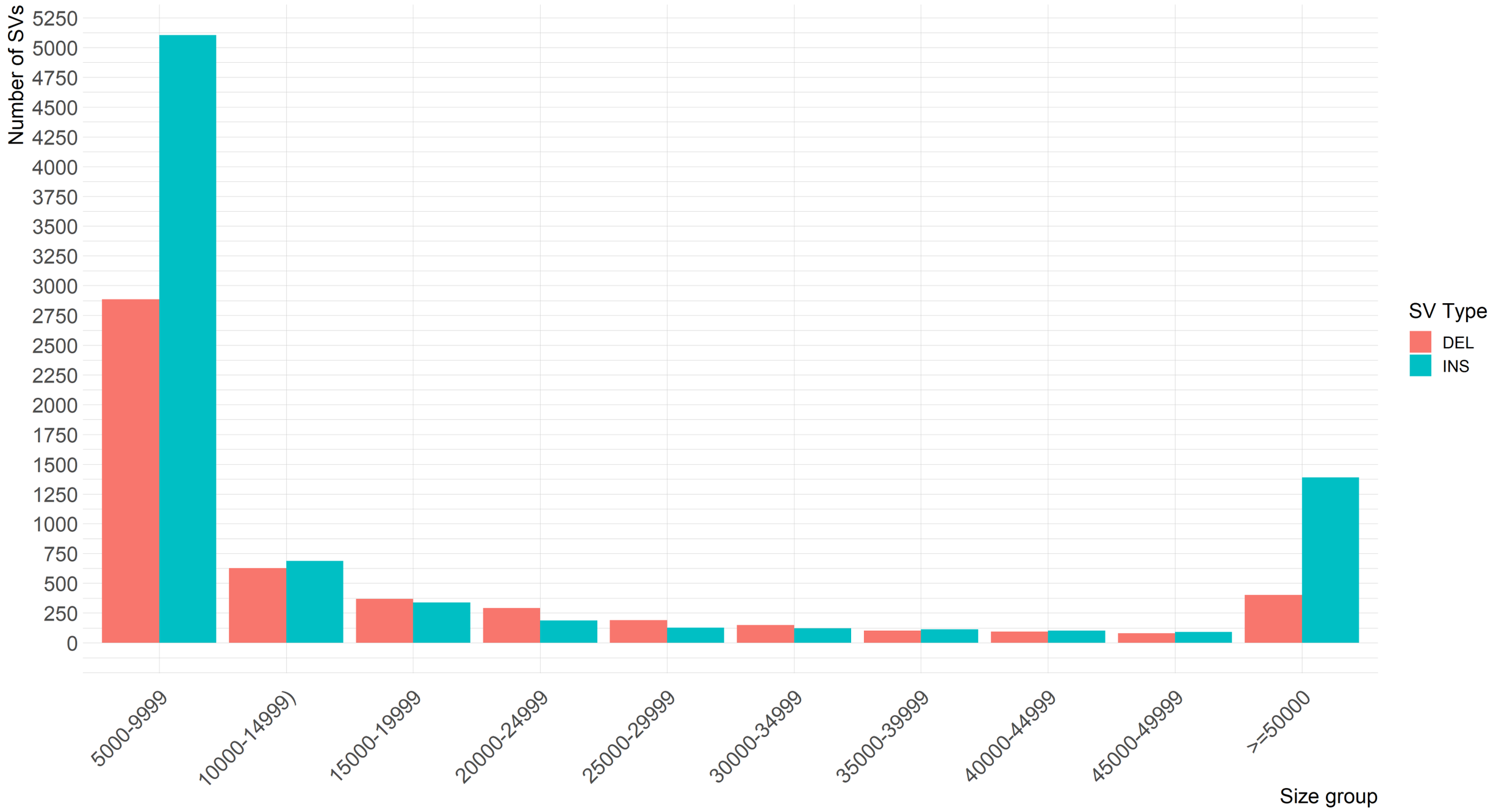
Figure 1: Histogram of the structural variant (SV) sizes. Histogram of the size of the identified SVs in bins of 5Kb.

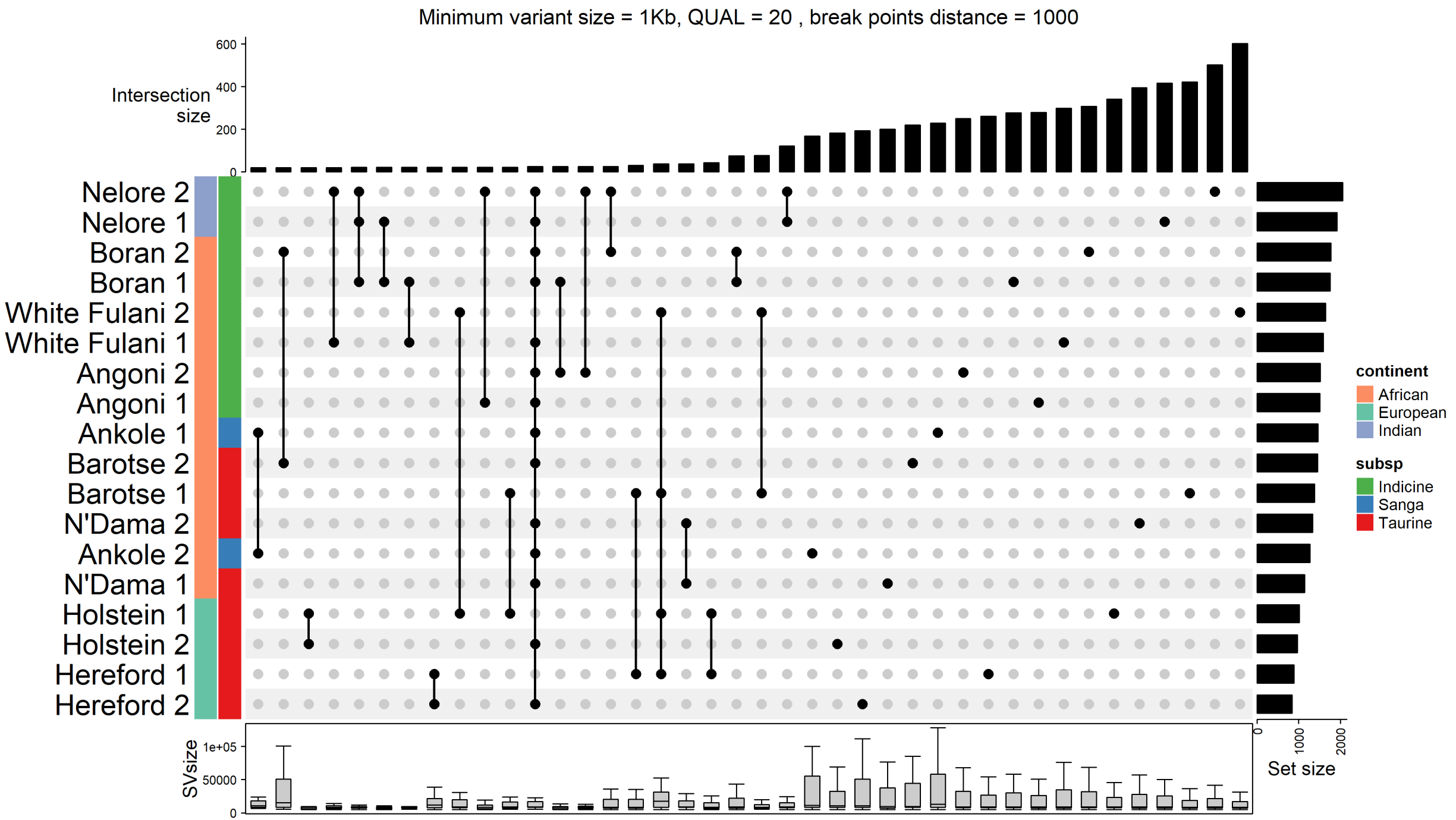
Figure 2: Upset plot of the structural variants. Upset plot of the structural variants by individual for the 40 sets containing the most SVs.

Table captions

Table 1: Description of the samples. Table describing the breeds and ancestry of samples, with the continent and country of origin. The identifiers, as well as the ENA accession codes, for each of the two animals sampled per breed are also reported.

Table 2: Raw number of structural variants (SVs) and type detected in the different samples. This table summarises the number of raw SVs detected in each sample, and their classification (e.g. insertion, deletion, duplication, inversion and inter- and intra-chromosomal translocation).


Number of SVs
5250
5000
4750
4500
4250
4000
3750
3500
3250
3000
2750
2500
2250
2000
1750
1500
1250
1000
750
500
250
0
5000-9999
10000-14999)
15000-19999
20000-24999
25000-29999
30000-34999
35000-39999
40000-44999
45000-49999
>=50000
Size group
SV Type
DEL
INS


Minimum variant size = 1Kb, QUAL = 20 , break points distance = 1000
Intersection size
600
400
200
0
Nelore 2
Nelore 1
Boran 2
Boran 1
White Fulani 2
White Fulani 1
Angoni 2
Angoni 1
Ankole 1
Barotse 2
Barotse 1
N'Dama 2
Ankole 2
N'Dama 1
Holstein 1
Holstein 2
Hereford 1
Hereford 2
SVsize
1e+05
50000
0
0
1000
2000
Set size
continent
African
European
Indian
subsp
Indicine
Sanga
Taurine