Identification of Copy Number Variants in Braque Français type Pyrénées"dog using CanineHD array

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Aim

Copy number variants (CNVs) are an important source of genetic variation complementary to single nucleotide polymorphisms (SNPs). In this study, we used a high-density SNP array (170 K) to detect CNVs in Braque Français, type Pyrénées dogs (BRA) (Fig. 1).

Figure 1: Example of Braque Français, type Pyrénées breed

Materials and Methods

Blood samples were collected from 48 individuals. Genotyping was performed with the Illumina CanineHD BeadChip.

After excluding SNPs which were unmapped or mapped to sex chromosomes, a total of 167,183 markers were used.

CNVs were detected using the algorithms implemented by PennCNV and in the Copy Number Analysis Module (CNAM) of the SVS 8.7.0 software (Golden Helix, Bozeman, MT, USA).

Copy number variation regions (CNVRs) were determined by aggregating the overlapping CNVs identified across all samples.

The gene content of CNVRs was assessed based on CanFam 3.1 in the Genome Data Viewer genome browser from the US NCBI database.

Results

Using PennCNV, a total of 1047 CNVs were detected, with an average length of 107.123 kb and average number of 40.3 CNVs per sample (Fig. 2).

The SVS identified 1638 CNVs with an average length of 110.41 kb and the average number of 63 CNVs per sample (Fig.2).

By aggregating the overlapping CNVs from PennCNV, a total of 181 CNVRs were identified. The CNVs identified with SVS were aggregated into 280 CNVRs.

Intersecting the two CNVR datasets a total of 45 CNVRs were obtained.

The CNVRs from PennCNV that did not fully overlap with SVS results were excluded, and vice versa.

The comparison with canine CNVRs from the available literature (Fig. 3) revealed 16 novel CNVRs for BRA.

Figure 2: Genomic distribution of CNVs in BRA.

The points in black showed the CNVs identified with PennCNV, whereas in red are reported the CNV identified with Copy Number Analysis Module-SVS..

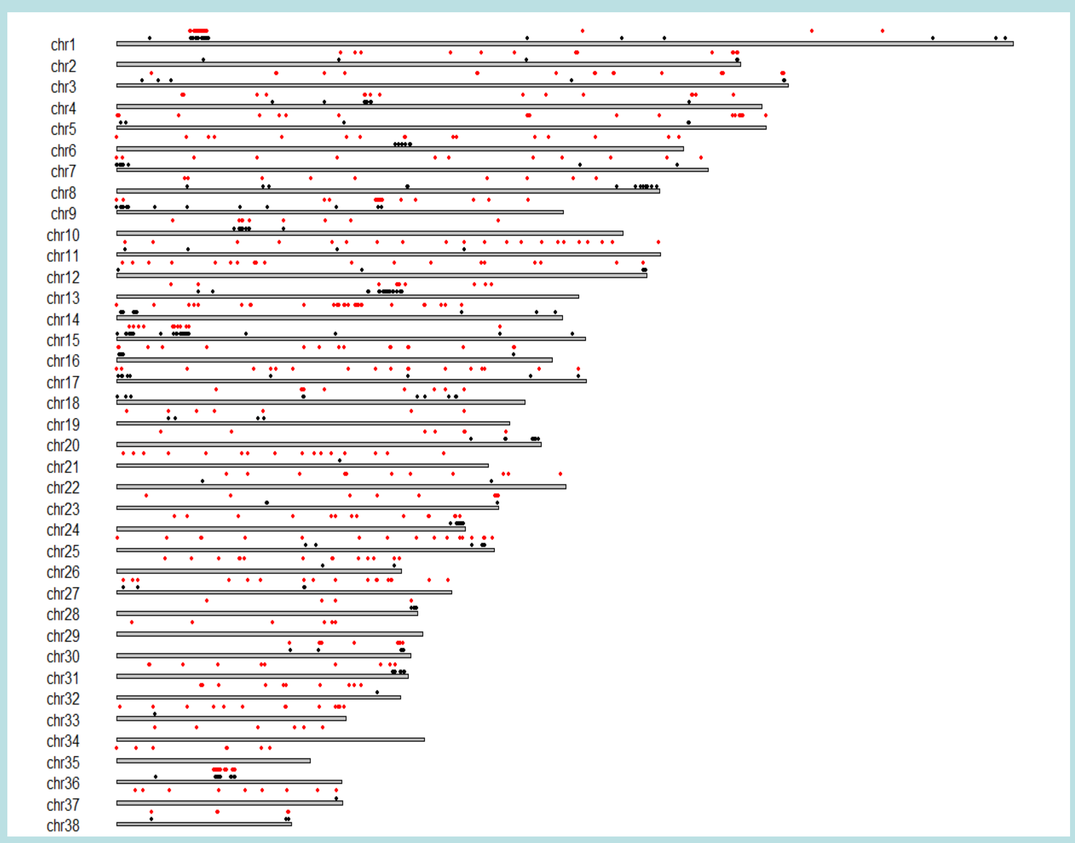
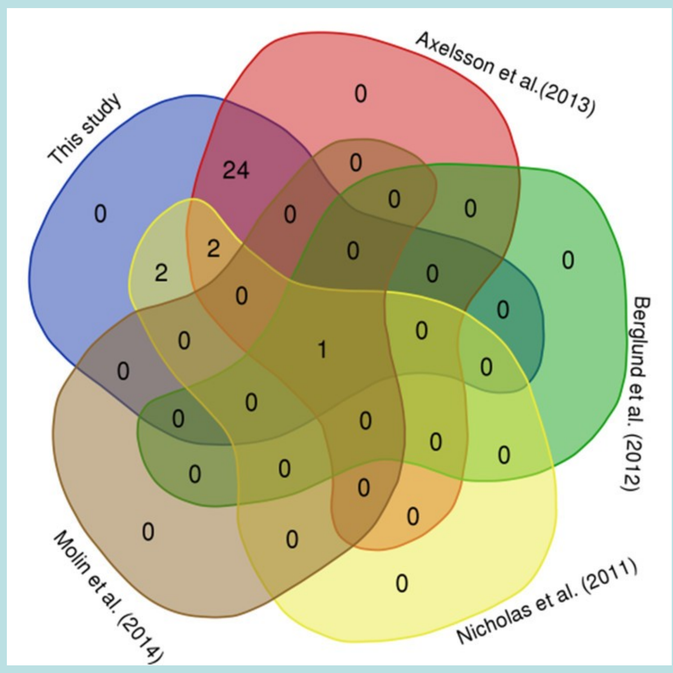


Figure 3. Comparison between identified CNVs in this and other studies



37 CNVRs overlap with 159 annotated genes.

The most significant biological processes were muscle structure development (GO:0061061), muscle cell differentiation (GO:0042692), and striated muscle cell differentiation (GO:0051146).

These are interesting results considering that BRA is a dog breed used for tracking, hunting, pointing and retrieving feathered game.

Conclusion

At present, limited knowledge is available on CNVs detected from HD array in dogs. We can hypothesize that selection for such hunting behavior, for which particular anatomical features are required, could have shaped, at least in part, the genetic background of this breed and, consequently, the frequency/presence of the detected CNVRs in these genes.