

### Keywords

GWAS, Copy Number Variation,  
Brown Swiss

### CORRESPONDING AUTHOR

**Raphaelle.T.M.M. Prinsen**  
raphaelle.prinsen@unimi.it

### JOURNAL HOME PAGE

[riviste.unimi.it/index.php/haf](http://riviste.unimi.it/index.php/haf)



UNIVERSITÀ DEGLI STUDI DI MILANO  
DIPARTIMENTO DI SCIENZE VETERINARIE  
PER LA SALUTE, LA PRODUZIONE ANIMALE  
E LA SICUREZZA ALIMENTARE



## Genome-wide association studies using copy number variants in Brown Swiss Dairy cattle.

R.T.M.M. Prinsen<sup>a\*</sup>, M.G. Strillacci<sup>a</sup>, F. Schiavini<sup>a</sup>, A. Rossoni<sup>b</sup>, B. Gredler<sup>c</sup>,  
M.A. Dolezal<sup>d</sup>, A. Bieber<sup>e</sup>, A. Bagnato<sup>a</sup>

<sup>a</sup>Università degli Studi di Milano, DiMeVet Via Celoria 10, Milan, Italy

<sup>b</sup>A.N.A.R.B., Località Ferlina 204, Bussolegno, Italy

<sup>c</sup>Qualitas AG, Chamerstrasse 56 Zug, Switzerland

<sup>d</sup>University of Veterinary Medicine, Veterinärplatz 1 Wien Austria

<sup>e</sup>Research Institute of Organic Agriculture, Ackerstrasse 113, Frick, Switzerland

### Abstract

Detecting Copy Number Variation (CNV) in cattle provides the opportunity to study their association with quantitative traits (Winchester et al., 2009; Zhang et al., 2009; Hou et al., 2011; Clop et al., 2012; de Almeida et al., 2016;). The aim of this study was to map CNVs in 1,410 Brown Swiss males and females using Illumina BovineHD Genotyping BeadChip data and to perform a genome-wide association analysis for production functional and health traits. After quality control, CNVs were called with the GoldenHelix SVS 8.3.1 and PennCNV software and were summarized to CNV regions (CNVRs) at a population level, using BEDTools. Additionally, common CNVRs between the two software were set as consensus. CNV-association studies were executed with the CNVRuler software using a linear regression model. Genes within significant associated CNVRs for each trait were annotated with a GO analysis using the DAVID Bioinformatics Resources 6.7.

The quality control filtered out 294 samples. The GoldenHelix SVS 8.3.1 software identified 25,030 CNVs summarized to 398 CNVRs while PennCNV identified 62,341 CNVs summarized to 5,578 CNVRs. A total of 127 CNVRs were identified to be significantly associated with one or more of the evaluated traits. The result of this study is a comprehensive genomic analysis of the Brown Swiss breed, which enriches the bovine CNV map in its genome. Finally, the results of the association studies deliver new information for quantitative traits considered in selection programs of the Brown Swiss breed.

### References

- Clop A., Vidal O., Amills M., 2012. Copy number variation in the genomes of domestic animals. *Animal Genetics* 43(5):503-17.
- de Almeida Santana M.H., Junior G.A., Cesar A.S., Freua M.C., da Costa Gomes R., da Luz E Silva S., Leme P.R., Fukumasu H., Carvalho M.E., Ventura R.V., Coutinho L.L., Kadarmideen H.N., Ferraz J.B., 2016. Copy number variations and genome-wide associations reveal putative genes and metabolic pathways involved with the feed conversion ratio in beef cattle. *J Appl. Genet.* 10.1007/s13353-016-0344-7.
- Hou Y., Liu G.E., Bickhart D.M., Cardone M.F., Wang K., Kim ES., Matukumalli L.K., Ventura M., Song J., VanRaden P.M., Sonstegard T.S., Van Tassell C.P., 2011. Genomic characteristics of cattle copy number variations. *BMC Genomics* 12: 127.
- Winchester L., Yau C., Ragoussis J., 2009. Comparing CNV detection methods for SNP arrays. *Brief Funct Genomic Proteomic.* 8(5):353-66.
- Zhang F., Gu W., Hurler M.E., Lupski J.R., 2009. Copy number variation in human health, disease, and evolution. *Annu Rev Genomics Hum Genet.* 10:451-81.