

Italian Journal of Animal Science

ISSN: 1828-051X (Online) Journal homepage: www.tandfonline.com/journals/tjas20

SNP-based diagnosis of robertsonian translocation 1;29 in Italian cattle breeds: frequency estimation and efficient detection

A. Bionda, M. Cortellari, A. Cesarani, F. Sbarra, A. Quaglia, L. Liotta, P. Parma & P. Crepaldi

To cite this article: A. Bionda, M. Cortellari, A. Cesarani, F. Sbarra, A. Quaglia, L. Liotta, P. Parma & P. Crepaldi (2025) SNP-based diagnosis of robertsonian translocation 1;29 in Italian cattle breeds: frequency estimation and efficient detection, Italian Journal of Animal Science, 24:1, 1345-1352, DOI: 10.1080/1828051X.2025.2505209

To link to this article: https://doi.org/10.1080/1828051X.2025.2505209

© 2025 The Author(s). Published by Informa UK Limited, trading as Taylor & Francis Group

Published online: 10 Jun 2025.

Submit your article to this journal

Article views: 754

View related articles

View Crossmark data

Full Terms & Conditions of access and use can be found at https://www.tandfonline.com/action/journalInformation?journalCode=tjas20

ITALIAN JOURNAL OF ANIMAL SCIENCE
2025, VOL. 24, NO. 1, 1345–1352
https://doi.org/10.1080/1828051X.2025.2505209

Taylor & Francis
Taylor & Francis Group

RESEARCH ARTICLE

OPEN ACCESS Check for updates

SNP-based diagnosis of robertsonian translocation 1;29 in Italian cattle breeds: frequency estimation and efficient detection

A. Bionda[a], M. Cortellari[a], A. Cesarani[b,c], F. Sbarra[d], A. Quaglia[d], L. Liotta[e], P. Parma[a] and P. Crepaldi[a]

[a]Dipartimento di Scienze Agrarie e Ambientali—Produzione, Territorio, Agroenergia, Università degli Studi di Milano, Milano, Italy; [b]Dipartimento di Agraria, Università degli Studi di Sassari, Sassari, Italy; [c]Department of Animal and Dairy Science, University of Georgia, Athens, Georgia, USA; [d]Associazione Nazionale Allevatori Bovini Italiani da Carne (ANABIC), San Martino in Colle, Perugia, Italy; [e]Dipartimento di Scienze Veterinarie, Università degli Studi di Messina, Messina, Italy

ABSTRACT

Robertsonian translocation 1;29 (rob(1;29)) is the most widespread chromosomal aberration in cattle, known to cause fertility impairment in heterozygous carriers. While karyological screening programs exist for some breeds, the analysis of SNP data has proven to be a faster and more cost-effective method to identify carriers. In this study, we analysed SNP data from 1938 individuals across 11 Italian cattle breeds, routinely collected by four breeders' associations, to screen for the rob(1;29) translocation. We focused on the first six Mb of chromosome 29, using principal component analysis and partitioning methods to identify the smallest subset of SNPs that could reliably detect carriers. Additionally, we investigated linkage disequilibrium (LD) between chromosomes 1 and 29 in rob(1;29) carriers. The frequency of rob(1;29) carriers in five beef breeds of Podolian origin was consistent with previous reports. No carriers were found in the Piedmontese or Pezzata Rossa Italiana breeds. 2% of Modicana and Bruna Italiana, and 1% of Pezzata Rossa d'Oropa subjects were carriers. Moreover, 40% of Cinisara individuals carried rob(1;29), including 4% who were homozygous. Depending on the array used, we identified one to three SNPs that, alone, could distinguish between carriers and wild type individuals. LD analysis showed no significant hotspots between BTA 1 and 29, but the average linkage was higher in carriers. Our findings confirm that a small SNP set from this region can effectively detect rob(1;29) carriers, supporting its inclusion in routine genotyping panels for broader and more efficient screening across cattle populations.

ARTICLE HISTORY
Received 2 January 2025
Revised 2 April 2025
Accepted 7 May 2025

KEYWORDS
Robertsonian translocation; cattle; SNP; linkage disequilibrium

Introduction

Robertsonian translocation 1;29 (rob(1;29)) is the most widespread chromosomal abnormality in cattle species (Gustavsson and Rockborn 1964; Holečková et al. 2021; Iannuzzi et al. 2021) and corresponds to a centric fusion of the acrocentric chromosomes 1 and 29 followed by the inversion and migration to the q-arm of a fragment 5 to 9 Mb-long pericentromeric segment of BTA 29 (Chaves et al. 2000; Di Meo et al. 2006; De Lorenzi et al. 2012; Escudeiro et al. 2021). As a result of the formation of this anomaly, there is no loss of genetic material, but heterozygous carrier subjects produce genetically imbalanced gametes leading to increased embryonic death and consequently to a fertility reduction (Gustavsson 1969; 1971; Dyrendahl and Gustavsson 1979; Schmutz et al. 1991; Lonergan et al. 1994; Bonnet-Garnier et al. 2006; 2008).

Discovered in 1964 in Swedish Red and White cattle, rob(1;29) has since been extensively studied and identified in over 50 cattle breeds, predominantly intended for meat production, with varying prevalence across the world (Popescu and Pech 1991; Iannuzzi et al. 2021). The absence of reports on *de novo* cases of rob(1;29) and our recent discovery of a shared haplotype among all carriers (Cortellari et al. 2024) supports the theory that this chromosomal aberration has an ancient origin, likely propagated throughout the global cattle population by a common ancestor (Di Meo et al. 2006; De Lorenzi et al. 2012).

CONTACT A. Bionda arianna.bionda@unimi.it

© 2025 The Author(s). Published by Informa UK Limited, trading as Taylor & Francis Group
This is an Open Access article distributed under the terms of the Creative Commons Attribution-NonCommercial License (http://creativecommons.org/licenses/by-nc/4.0/), which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited. The terms on which this article has been published allow the posting of the Accepted Manuscript in a repository by the author(s) or with their consent.

Given the detrimental effects of rob(1;29) on fertility, screening programs have been carried out in several countries, including Italy, for identifying carrier individuals and excluding them from the mating schemes (Ducos et al. 2008; Parma and Zannotti 2020). So far, these screenings were based on cytogenetic technologies, which imply high costs, waiting times, fresh blood samples and require specialised personnel, thus limiting the possibility of a large-scale screening. However, recent literature showed the possibility of identifying rob(1;29) carriers using more economical and effective methods, such as q-PCR (Iannuzzi et al. 2024) and SNP data (Cortellari et al. 2024).

In particular, the widespread adoption of genotyping in cattle population management has resulted in the generation of an extensive amount of SNP data (Holliday et al. 2018; Gutierrez-Reinoso et al. 2021), making them a promising resource for the economical and efficient diagnosis of rob(1;29). For this reason, the aim of the present study is to assess the presence and prevalence of rob(1;29) in eleven Italian cattle populations by analysing SNP data already at the disposal of breeding associations.

Materials and methods

Dataset creation and filtering

Data analysed in the present work were provided from four breeders' associations (Table 1). The first set of data belonged to ANABIC (*Associazione Nazionale Allevatori Bovini Italiani da Carne*), which provided data for five Italian beef breeds, all sharing a Podolian origin –Maremmana, Podolica, Chianina, Romagnola, and Marchigiana–, for a total of 974 individuals genotyped using GGP Bovine 33k SNP chip. For 153 of these individuals, a cytogenetic diagnosis for rob(1;29) was available, since bulls of these breeds are subject to an early screening program. The second set of data consisted of SNP data obtained from 119 Piedmontese beef breed subjects genotyped with GGP Bovine 150K, provided by ANABORAPI (*Associazione Nazionale Allevatori Bovini Razza Piemontese*). The third dataset came from ANAPRI (*Associazione Nazionale Pezzata Rossa Italiana*) and it included 663 individuals belonging to Pezzata Rossa Italiana (Italian Simmental), Cinisara, and Modicana breeds, genotyped using either GGP Bovine 150K or Illumina BovineHD Genotyping BeadChip (including more than 777,000 SNPs). Lastly, 140 individuals of the Pezzata d'Oropa breed genotyped with GGP Bovine 33k SNP chip and 87 individuals of the Bruna Italiana (Italian Brown) breed genotyped with Illumina BovineHD Genotyping BeadChip were provided by ANARB (*Associazione Nazionale Razza Bruna*).

The raw dataset obtained by merging all the data above included 1,993 subjects that were subsequently filtered for genotyping quality of the SNPs (*--geno* 0.05) and subjects (*--mind* 0.05) using PLINK (Purcell et al. 2007). After these operations, a total of 1,938 individuals were retained. Following the indications reported by Cortellari et al. (2024), the analyses, except for those related to linkage disequilibrium and inbreeding, were performed on the 71 SNPs within the first 6 Mb of BTA29 that were correctly genotyped in a reference homozygous subject.

Principal component analysis and partitioning analysis

Principal component analysis (PCA) was performed with PLINK 1.9 software (Purcell et al. 2007). Results were compared with the karyological diagnosis of rob(1;29) provided for a subset of individuals.

Haplotypes of all the individuals were extracted with an in-house script. Using JMP®, Version 16 (SAS Institute Inc., Cary, NC, 1989–2023) software, a partitioning analysis was conducted on the SNPs belonging to the haplotypes. The aim was to identify the smallest possible number of SNPs capable of distinguishing translocated subjects from wild type ones. Sensitivity

Table 1. Analysed data and results.

Breed code	Breed name	N. subjects	N. cytogenetically tested subjects	rob (1;29) SNP diagnosis	SNP-based frequency (standard error)
BRUI	Bruna Italiana	87	0	2 Het, 85 Wild Type	0.02 (0.016)
CHI	Chianina	218	25	3 Het, 215 Wild Type	0.01 (0.008)
CIN	Cinisara	269	0	10 Hom, 97 Het, 162 Wild Type	0.40 (0.030)
MAR	Marchigiana	213	34	17 Het, 196 Wild Type	0.08 (0.019)
MOD	Modicana	179	0	1 Hom, 3 Het, 175 Wild Type	0.02 (0.011)
MRM	Maremmana	122	29	1 Hom, 14 Het, 107 Wild Type	0.12 (0.030)
PIEM	Piedmontese	119	0	119 Wild Type	0.00 (0.00)
POD	Podolica	202	34	28 Het, 174 Wild Type	0.14 (0.024)
PRI	Pezzata Rossa Italiana	182	0	182 Wild Type	0 (0)
PROR	Pezzata Rossa d'Oropa	140	0	2 Het, 138 Wild Type	0.01 (0.010)
RMG	Romagnola	207	31	22 Het, 185 Wild Type	0.11 (0.021)

BRUI: Bruna Italiana; CHI: Chianina; CIN: Cinisara; MAR: Marchigiana; MOD: Modicana; MRM: Maremmana; PIEM: Piedmontese; POD: Podolica; PRI: Pezzata Rossa Italiana; PROR: Pezzata Rossa d'Oropa; RMG: Romagnola.

was measured as $\frac{true\ \ positive}{true\ \ positive+false\ \ negative}$ and specificity as $\frac{true\ \ negative}{true\ \ negative+false\ \ positive}$, where true and false were assigned based on the concordance or discordance between the genotype of the identified SNPs and the complete haplotype or the karyological analysis, where available.

Linkage disequilibrium

For each group (Hom rob(1;29), Het rob (1;29), and Wild type), the linkage disequilibrium (LD), measured as R^2, was calculated between all pairs of markers of BTA1 and BTA29 using gaston 1.5.7 R package (Perdry and Dandine-Roulland 2023). The heatmaps of the values were plotted using ggplot2 (Wickham 2016). A Welch two-sample t-test was performed using stats R package's function *t.test* to compare LD in Het rob(1;29) and Wild type groups.

Inbreeding analysis

To investigate the potential correlation between rob(1;29) frequency and inbreeding, individual genomic inbreeding coefficients were calculated for the subjects and summarised by breed and rob(1;29) diagnosis. The analysis covered all 29 autosomes, with a total of 22,664 SNPs retained after quality control. To identify runs of homozygosity (ROH), PLINK's *--homozyg* function was employed with specific parameters. These included setting the number of SNPs in each window to 20, defining a minimum ROH length of 1000 kb, and ensuring a density of at least one SNP per 500 kb. No heterozygous SNPs were allowed in a window, and up to two missing SNPs were tolerated. The individual genomic inbreeding coefficient (F_{ROH}) was calculated using McQuillan's formula (McQuillan et al. 2008), which measures the ratio of the total length of ROH to the entire autosomal genome covered by SNPs. Due to the low density of the markers, the resulting inbreeding values should be interpreted with caution. However, this analysis can offer insights into the relative inbreeding of the included populations, allowing us to assess whether rob(1;29) carriers or populations with a higher frequency of rob(1;29) exhibit higher levels of homozygosity compared to others.

Results

Principal component analysis

The results of the PCA, considering the SNP included in the first 6 Mb of BTA29, confirm what was reported by Cortellari et al. (2024): the possibility of separating the individuals into three well-distinct groups, i.e. Hom rob(1;29), Het rob(1;29), and Wild type (Figure 1). This allowed us to formulate a SNP-based diagnosis (Table 1), reporting 1% carriers in Chianina, 8% in Marchigiana, 11% in Romagnola, 12% in Maremmana (one of which was homozygous, as already diagnosed with karyological test), and 14% in Podolica breeds. No carriers were observed in Piedmontese or Pezzata Rossa Italiana breeds, whereas 1% of Pezzata Rossa d'Oropa and 2% of Bruna Italiana were translocated. Lastly, we identified 2% of Modicana subjects carrying the translocation, one of which was homozygous, and 40% of Cinisara ones, including 10 (4%) homozygous carriers.

Partitioning analysis

A partitioning analysis on 153 individuals with karyological diagnosis confirmed that a subject presenting at least 66 SNPs or 98.6% of genotyped SNPs in common with the homozygous subject's haplotype can be classified as rob(1;29) carrier with 100% specificity and 100% sensibility. The same number of SNPs shared with the homozygous carrier's haplotype was able to discriminate between carriers and wild types in the entire dataset; however, this corresponded to 95.8% of the genotyped SNPs due to differences in genotyping rates among individuals. Particularly, the discriminant analysis suggested that the correspondence of rs110903659, rs136708960, and rs136182358 was sufficient to obtain the same results in distinguishing between wild type and carrier subjects than analysing the whole haplotype, with no false positives or negatives (sensibility: 100%; specificity: 100%). When only the karyologically tested individuals were considered, the combination of rs110903659 and rs136708960 was sufficient to detect rob(1;29) carriers.

The same analysis was also performed on the publicly available dataset generated within the Bovita project (Mastrangelo et al. 2018), which was analysed with this method for the identification of rob(1;29) carriers in our previous study (Cortellari et al. 2024), finding a total of 762 wild type individuals and 38 carriers of rob(1;29) belonging to 16 breeds. This was done because these samples were genotyped with another SNP chip (GGP Bovine 150K), containing different SNPs on BTA29 which may better identify subjects carrying the translocation. Within the analysed region, all the carriers had 100% of correctly genotyped SNPs (at least 55 out of the 65 analysed ones) in common with the reference rob(1;29) homozygous subject. Furthermore, the correspondence of rs43136718 proved to be a discriminating

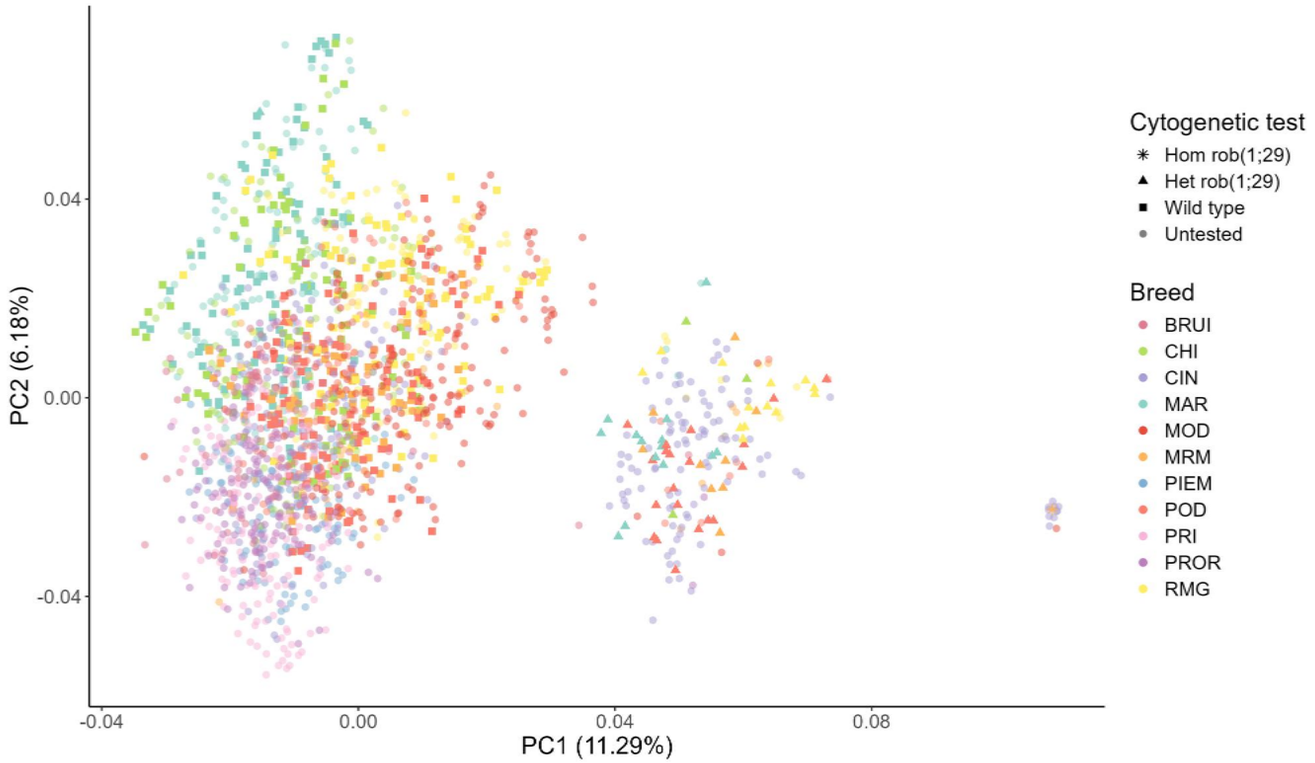


Figure 1. Principal component analysis of the 71 SNPs included in the first 6 Mb of BTA29. Jitter was applied to reduce overlap among points; for istance, all hom rob(1;29) individuals would otherwise be perfectly aligned. Breed codes are reported in Table 1.

factor between the two groups, with only a single false positive among the 762 wild type subjects (sensitivity: 100%; specificity: 99.9%).

Linkage disequilibrium

The LD was obtained between all the pairs of genotyped markers of BTA1 and BTA29 for each group. The results of the LD between markers of the BTA1 and BTA29 are shown in Figure 2. No clear differences were found between the Wild type and the Het rob(1;29) groups (Figures 2A and B, respectively), i.e. no spots of high LD were identified between markers of the two chromosomes in the translocated subjects. Because Hom rob(1;29) individuals present a common haplotype in the first six Mb of BTA29, an LD value was not calculable between pairs of markers located in that region (Figure 2C). It was also observed that, on average, the LD between pairs of markers of BTA1 and BTA29 was significantly higher in Het rob(1;29) (R^2 = 0.0079 ± 0.0114) than in Wild type group (R^2 = 0.0032 ± 0.0051) (p-value of the t-test: 2.2e-16).

Inbreeding

Genomic inbreeding (F_{ROH}) was analysed across all breeds based on subjects' SNP-based diagnosis of rob(1;29) (Figure 3). No statistically significant differences were found between carriers and wild type individuals within the same breed, nor was there a correlation between a breed's rob(1;29) frequency and its mean F_{ROH}. An exception was noted in a single Modicana homozygous carrier, who exhibited a notably high, though not the highest, F_{ROH} within the breed. Given the rarity of the translocation in this population, it is likely that this individual resulted from mating between two closely related heterozygous carriers. The Cinisara breed displayed relatively low but variable inbreeding, with a mean ± SD value of 4.7 ± 5.0%, compared to a global inbreeding rate of 7.8 ± 4.6% across the entire dataset.

Discussion

The analysis of SNP data routinely collected by breeders' associations proved to be a reliable and cost-effective method for pre-screening cattle for the rob(1;29) chromosomal translocation. This approach enables a wider investigation of this translocation within several populations, allowing more individuals to be tested, and across different breeds, including those not routinely screened. This represents a significant advancement in the ability to monitor and manage the spread of rob(1;29) in cattle.

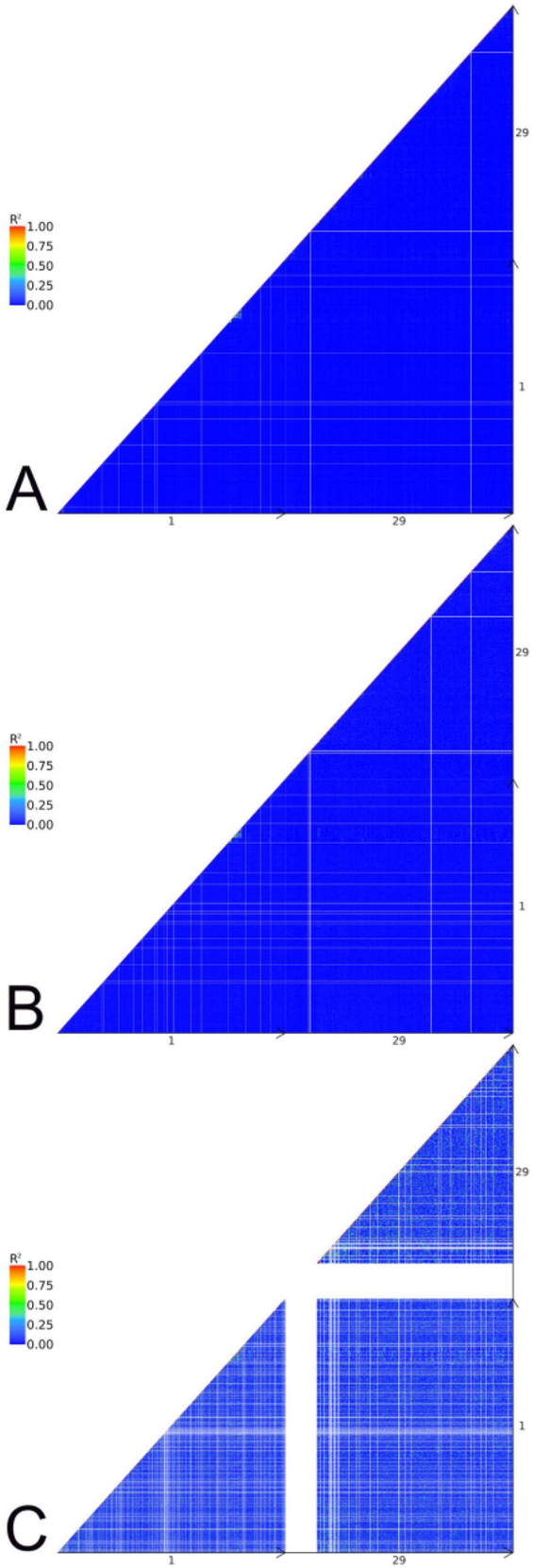


Figure 2. Heatmap of linkage disequilibrium values between markers of BTA1 and BTA29 in Wild type (A), Het rob(1;29) (B), and Hom rob(1;29) (C) groups.

The frequency of rob(1;29) carriers identified in five beef breeds (i.e. CHI, MAR, MRM, POD, and RMG) managed by ANABIC is quite similar to previous reports, such as those by Ducos et al. (2008) and Parma and Zannotti (2020). Minor discrepancies may arise from differences in the periods analysed, as screening programs implemented over time could have altered carrier prevalence. Additionally, while screening programs typically focus on bulls aimed at reproduction, this study evaluated a more heterogeneous population, which may further explain the variation.

Among all the analyses performed, no carriers were detected in either the beef Piedmontese or dual-purpose Pezzata Rossa Italiana breeds. The majority of studies about rob(1;29) prevalence in other breeds date back to the seventies and eighties, as reviewed by Popescu and Pech (1991). In the Simmental breed, variable prevalences were reported in different European countries –from 1–2% in Austria and Switzerland up to 21% in Sweden–. More recently, a prevalence of 0.8% was found in Czech Simmental bulls screened for chromosomal aberrations (Citek et al. 2009). Studies about Brown Swiss breed from the same years ('70-'80) indicated a 0.4% prevalence in Switzerland and 6–10% in the USA (Popescu and Pech 1991). A more recent survey of cytogenetic screenings reported 3.5% of carriers in Bruna Italiana cattle and 0.7% in the Brown from Romania (Ducos et al. 2008). In the present study, we found 2% of the analysed Bruna Italiana individuals to be rob(1;29) carriers.

To the best of our knowledge, only two studies investigated the karyological status of small samples of Modicana, reporting a rob(1;29) frequency of 6.4% and 0%, respectively (Succi et al. 1980; Ciotola et al. 2009). Ciotola et al. (2009) also found 23% of rob(1;29) carriers out of 30 Cinisara subjects analysed. In our Modicana dataset, 2% of individuals were identified as carriers, including one homozygous individual. The highest frequency of rob(1;29) carriers was found in the Cinisara breed, where 40% of individuals were carriers. Among these, 17 individuals (4% of the tested population) were homozygous carriers. This high prevalence of carriers and homozygous individuals aligns with the findings of Cortellari et al. (2024), further validating the significant presence of rob(1;29) in this breed. This high prevalence cannot be justified by a high inbreeding level within the population, which instead shows relatively low F_{ROH} coefficients. Although Cinisara breeders reported no fertility issues, further research is necessary to confirm whether the translocation affects reproductive performance. Indeed, it is also possible that the traditional semi-extensive farming system of this breed makes it

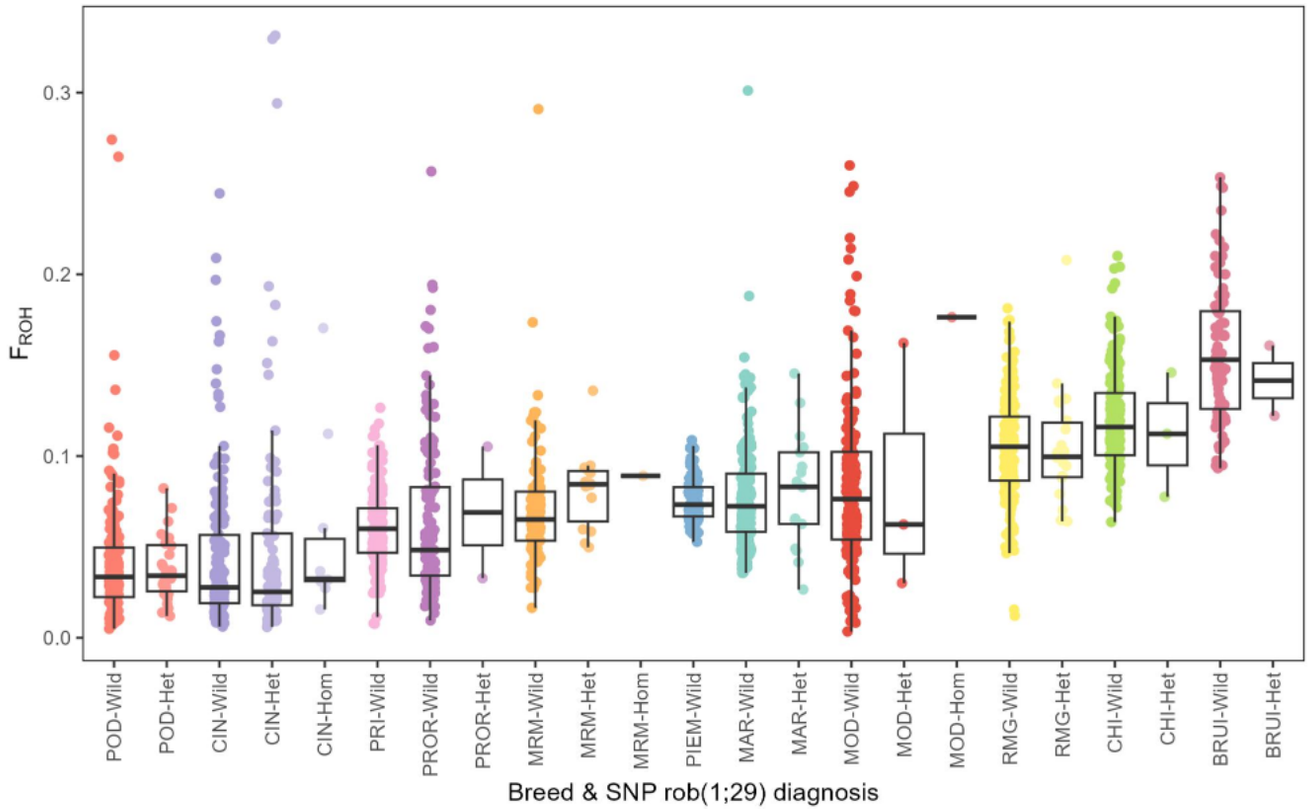


Figure 3. Boxplot of genomic inbreeding (F_{ROH}) by breed and rob(1;29) SNP-based diagnosis.

difficult to detect the rob(1;29)-induced decline in fertility. The presence of a substantial number of homozygous carriers may make the Cinisara breed a valuable genetic reservoir for studying the effects of rob(1;29) and its long-term impact.

Additionally, this study confirms that all rob(1;29) carriers, either homozygous or heterozygous, shared a common haplotype within the first 6 Mb of BTA29. Moreover, the partitioning analysis revealed that a small subset of specifically selected SNPs—three from the GGP bovine 33K panel and one from Illumina arrays—was sufficient to correctly identify carriers, as defined by karyological analysis (where available) or by SNP-based diagnosis using the entire haplotype. This finding highlights the potential for these SNPs to be incorporated into commonly used cattle genotyping panels, such as the Bovine ISAG SNP Parentage Panel, which currently lacks markers from this region. Including these SNPs in routine screenings would improve the accuracy and efficiency of detecting rob(1;29) carriers, contributing to more effective genetic management practices across cattle populations.

Linkage disequilibrium (LD) analyses did not reveal any significant hotspots of high linkage between the two chromosomes involved in the translocation, BTA1 and BTA29, among rob(1;29) carriers. This could explain why Jourdain et al. (2023), despite analysing a large dataset and including breeds known to carry the translocation, did not identify any rob(1;29) through their LD investigation. However, it is important to note that, for a more accurate comparison, our analysis should ideally be performed on large family groups, as was done in their study. Additionally, our results showed that the average linkage between BTA1 and BTA29 in rob(1;29) carriers was higher than in wild type individuals, suggesting that, despite not being detectable from classical graphical analyses, the translocation may still influence chromosomal interaction.

Conclusions

The findings of the present study pointed out breed-specific differences in rob(1;29) prevalence, with notably high frequencies of heterozygous and homozygous carriers in the Cinisara breed, which could represent a valuable resource for studying the translocation's genetic and phenotypic impacts. This study demonstrates the practical feasibility of using SNP data routinely collected by breeders' associations for cost-effective pre-screening of the rob(1;29) chromosomal translocation in cattle. Because this approach relies on a simple and rapid examination of existing genotyping data, with no need for additional costs or time-consuming analyses, it would enable the

resurgence of pre-screening for rob(1;29) in all cattle breeds under genomic selection and/or evaluation.

The shared haplotype identified among rob(1;29) carriers within the first 6 Mb of BTA29 highlights the potential for targeted genotyping, with a small subset of SNPs proving sufficient for accurate carrier detection. Integrating these SNPs into standard cattle genotyping panels could significantly enhance the efficiency of screening programs. This study provides an efficient tool for broader population and breed monitoring, thereby enhancing efforts to manage and mitigate the spread of this chromosomal aberration. Further validation in non-Italian breeds could provide insights into the potential for implementing this tool in other countries and populations.

Acknowledgements

The authors are grateful to ANABIC, ANABORAPI, ANAPRI, and ANARB associations.The authors acknowledge support from the University of Milan through the APC initiative.

Disclosure statement

No potential conflict of interest was reported by the author(s).

Funding

Data collection was partially supported by the Italian Ministry of Agriculture through the projects DUALBREEDING (grant no. J21J18000010005, and DUALBREEDING Fase 2, grant no. J22C21000670005), I-BEEF (grant no. J99H18000000005) and I-BEEF2 (grant no. J92C21002110005). This study was carried out within the Agritech National Research Center and received funding from the European Union Next-GenerationEU (PIANO NAZIONALE DI RIPRESA E RESILIENZA (PNRR) – MISSIONE 4 COMPONENTE 2, INVESTIMENTO 1.4 – D.D. 1032 17/06/2022, CN00000022). This manuscript reflects only the authors' views and opinions, neither the European Union nor the European Commission can be considered responsible for them.

ORCID

A. Bionda http://orcid.org/0000-0001-9116-3208
M. Cortellari http://orcid.org/0000-0002-5161-0648
A. Cesarani http://orcid.org/0000-0003-4637-8669
L. Liotta http://orcid.org/0000-0002-3242-1817
P. Parma http://orcid.org/0000-0003-1544-6673
P. Crepaldi http://orcid.org/0000-0002-6526-2162

Data availability statement

The genomic data analysed in this study were generated by breeders' associations and remain their property. For access, reasonable requests can be directed to the corresponding author, AB, who will facilitate contact with the appropriate representatives of the associations.

References

Bonnet-Garnier A, Lacaze S, Beckers JF, Berland HM, Pinton A, Yerle M, Ducos A. 2008. Meiotic segregation analysis in cows carrying the t(1;29) Robertsonian translocation. Cytogenet Genome Res. 120(1–2):91–96. http://www.ncbi.nlm.nih.gov/pubmed/18467829. doi:10.1159/000118744.

Bonnet-Garnier A, Pinton A, Berland HM, Khireddine B, Eggen A, Yerle M, Darré R, Ducos A. 2006. Sperm nuclei analysis of 1/29 Robertsonian translocation carrier bulls using fluorescence in situ hybridization. Cytogenet Genome Res. 112(3–4):241–247. http://www.ncbi.nlm.nih.gov/pubmed/16484779. doi:10.1159/000089877.

Chaves R, Heslop-Harrsion JS, Guedes-Pinto H. 2000. Centromeric heterochromatin in the cattle rob(1;29) translocation: alpha-satellite I sequences, in-situ MspI digestion patterns, chromomycin staining and C-bands. Chromosome Res. 8(7):621–626. [accessed 2024 Sep 25]. https://pubmed.ncbi.nlm.nih.gov/11117358/. doi:10.1023/a:1009290125305.

Ciotola F, Albarella S, Liotta L, Contessa A, di Meo GP, Barbieri V, Peretti V. 2009. Native cattle breeds of Southern Italy: karyological profile. Ital J Anim Sci [Internet]. 8(Suppl. 2):54–56. [accessed 2024 Sep 25]. https://www.tandfonline.com/doi/abs/10.4081/ijas.2009.s2.54. doi:10.4081/ijas.2009.s2.54.

Citek J, Rubes J, Hajkova J. 2009. Short communication: robertsonian translocations, chimerism, and aneuploidy in cattle. J Dairy Sci. 92(7):3481–3483. [accessed 2024 Sep 26]. http://www.journalofdairyscience.org/article/S0022030209706666/fulltext. doi:10.3168/jds.2009-2099.

Cortellari M, Bionda A, Liotta L, Sbarra F, Parma P, Crepaldi P. 2024. Identification of a common haplotype in carriers of rob (1;29) in 32 Italian cattle breeds. Sci Rep. 14(1):2057. doi:10.1038/s41598-023-46341-3.

De Lorenzi L, Genualdo V, Gimelli S, Rossi E, Perucatti A, Iannuzzi A, Zannotti M, Malagutti L, Molteni L, Iannuzzi L, et al. 2012. Genomic analysis of cattle rob(1;29). Chromosome Res. 20(7):815–823. http://www.ncbi.nlm.nih.gov/pubmed/23053571. doi:10.1007/s10577-012-9315-y.

Di Meo GP, Perucatti A, Chaves R, Adega F, De Lorenzi L, Molteni L, De Giovanni A, Incarnato D, Guedes-Pinto H, Eggen A, et al. 2006. Cattle rob(1;29) originating from complex chromosome rearrangements as revealed by both banding and FISH-mapping techniques. Chromosome Res. 14(6):649–655. http://www.ncbi.nlm.nih.gov/pubmed/16964571. doi:10.1007/s10577-006-1074-1.

Ducos A, Revay T, Kovacs A, Hidas A, Pinton A, Bonnet-Garnier A, Molteni L, Slota E, Switonski M, Arruga MV, et al. 2008. Cytogenetic screening of livestock populations in Europe: an overview. Cytogenet Genome Res. 120(1–2):26–41. http://www.ncbi.nlm.nih.gov/pubmed/18467823. doi:10.1159/000118738.

Dyrendahl I, Gustavsson I. 1979. Sexual functions, semen characteristics and fertility of bulls carrying the 1/29 chromosome translocation. Hereditas [Internet]. 90(2):281–289. http://www.ncbi.nlm.nih.gov/pubmed/437991. doi:10.1111/j.1601-5223.1979.tb01315.x.

Escudeiro A, Adega F, Robinson TJ, Heslop-Harrison JS, Chaves R. 2021. Analysis of the Robertsonian (1;29) fusion in Bovinae reveals a common mechanism: insights into its clinical occurrence and chromosomal evolution. Chromosome Res. 29(3–4):301–312. [accessed 2024 Sep 25]. https://link.springer.com/article/10.1007/s10577-021-09667-0. doi:10.1007/s10577-021-09667-0.

Gustavsson I. 1969. Cytogenetics, distribution and phenotypic effects of a translocation in Swedish cattle. Hereditas [Internet]. 63(1):68–169. http://www.ncbi.nlm.nih.gov/pubmed/5399228. doi:10.1111/j.1601-5223.1969.tb02259.x.

Gustavsson I. 1971. Distribution of the 1/29 translocation in the A.I. Bull population of Swedish Red and White cattle. Hereditas [Internet]. 69(1):101–106. http://www.ncbi.nlm.nih.gov/pubmed/5173414. doi:10.1111/j.1601-5223.1971.tb02422.x.

Gustavsson I, Rockborn G. 1964. Chromosome abnormality in three cases of lymphatic leukæmia in cattle. Nature [Internet]. 203(4948):990–990. https://www.nature.com/articles/203990a0. doi:10.1038/203990a0.

Gutierrez-Reinoso MA, Aponte PM, Garcia-Herreros M. 2021. genomic analysis, progress and future perspectives in dairy cattle selection: a review. Anim [Internet]. 11(3):599. [accessed 2024 Sep 25]. https://www.mdpi.com/2076-2615/11/3/599/htm.

Holečková B, Schwarzbacherová V, Galdíková M, Koleničová S, Halušková J, Staničová J, Verebová V, Jutková A. 2021. Chromosomal aberrations in cattle. Genes (Basel). 12(9):1330. [accessed 2024 Sep 25]/pmc/articles/PMC8468509/ doi:10.3390/genes12091330.

Holliday JA, Hallerman EM, Haak DC. 2018. Genotyping and sequencing technologies in population genetics and genomics. In: Rajora OP, editor. Popul genomics concepts, approch appl [Internet]. Cham: Springer. p. 83–125. [accessed 2024 Sep 25]. https://link.springer.com/chapter/10.1007/13836_2017_5.

Iannuzzi A, Demyda-Peyrás S, Pistucci R, Morales R, Zannotti M, Sbarra F, Quaglia A, Parma P. 2024. A genomic biomarker for the rapid identification of the rob(1;29) translocation in beef cattle breeds. Sci Rep [Internet]. 14(1):2951. [accessed 2024 Sep 25]. https://www.nature.com/articles/s41598-024-53232-8. doi:10.1038/s41598-024-53232-8.

Iannuzzi A, Parma P, Iannuzzi L. 2021. Chromosome abnormalities and fertility in domestic bovids: a review. Animals [Internet]. 11(3):802. https://www.mdpi.com/2076-2615/11/3/802. doi:10.3390/ani11030802.

Jourdain J, Barasc H, Faraut T, Calgaro A, Bonnet N, Marcuzzo C, Suin A, Barbat A, Hozé C, Besnard F, et al. 2023. Large-scale detection and characterization of interchromosomal rearrangements in normozoospermic bulls using massive genotype and phenotype data sets. Genome Res. 33(6):957–971. http://www.ncbi.nlm.nih.gov/pubmed/37414574. doi:10.1101/gr.277787.123.

Lonergan P, Kommisrud E, Andresen O, Refsdal AO, Farstad W. 1994. Use of semen from a bull heterozygous for the 1 29 translocation in an IVF program. Theriogenology [Internet]. 41(7):1379–1384. http://www.ncbi.nlm.nih.gov/pubmed/16727492. doi:10.1016/0093-691x(94)90189-p.

Mastrangelo S, Ciani E, Ajmone Marsan P, Bagnato A, Battaglini L, Bozzi R, Carta A, Catillo G, Cassandro M, Casu S, et al. 2018. Conservation status and historical relatedness of Italian cattle breeds. Genet Sel Evol. 50(1):35. [accessed 2024 Sep 24]. https://gsejournal.biomedcentral.com/articles/10.1186/s12711-018-0406-x. doi:10.1186/s12711-018-0406-x.

McQuillan R, Leutenegger A-L, Abdel-Rahman R, Franklin CS, Pericic M, Barac-Lauc L, Smolej-Narancic N, Janicijevic B, Polasek O, Tenesa A, et al. 2008. Runs of homozygosity in European populations. Am J Hum Genet. 83(3):359–372. [accessed 2020 Jun 16]. http://www.ncbi.nlm.nih.gov/pubmed/18760389. doi:10.1016/j.ajhg.2008.08.007.

Parma P, Zannotti M. 2020. Le analisi cromosomiche nelle razze Chianina, Marchigiana e Romagnola [Chromosomic analyses in Chianina, Marchigiana, and Romagnola breeds]. Taurus [Internet]. 4:4–9. https://www.anabic.it/stampa/pdf/taurus/2020/04/Taurus_2020_n4_web.pdf.

Perdry H, Dandine-Roulland C. 2023. gaston: genetic data handling (QC, GRM, LD, PCA) & linear mixed models [Internet]. https://cran.r-project.org/package=gaston.

Popescu C, Pech A. 1991. Une bibliographie sur la translocation 1/29 de bovins dans le monde (1964-1990). Ann Zootech. 40(4):271–305. doi:10.1051/animres:19910405.

Purcell S, Neale B, Todd-Brown K, Thomas L, Ferreira MAR, Bender D, Maller J, Sklar P, de Bakker PIW, Daly MJ, et al. 2007. PLINK: a toolset for whole-genome association and population-based linkage analysis. Am J Hum Genet. 81(3):559–575. doi:10.1086/519795.

Schmutz SM, Moker JS, Barth AD, Mapletoft RJ. 1991. Embryonic loss in superovulated cattle caused by the 1;29 Robertsonian translocation. Theriogenology [Internet]. 35(4):705–714. http://www.ncbi.nlm.nih.gov/pubmed/16726939. doi:10.1016/0093-691x(91)90411-6.

Succi G, Molteni L, de Giovanni AM. 1980. Cytogenetic study of some Italian cattle breeds on the decrease or in the process of extinction. In: Proceeding 4th Eur Colloq Cytogenet Domest Anim. [place unknown]. p. 136–140.

Wickham H. 2016. ggplot2: elegant graphics for data analysis. New York, USA: Springer-Verlag.