



# SNP-based identification of Robertsonian translocation 1;29 in 32 Italian cattle breeds

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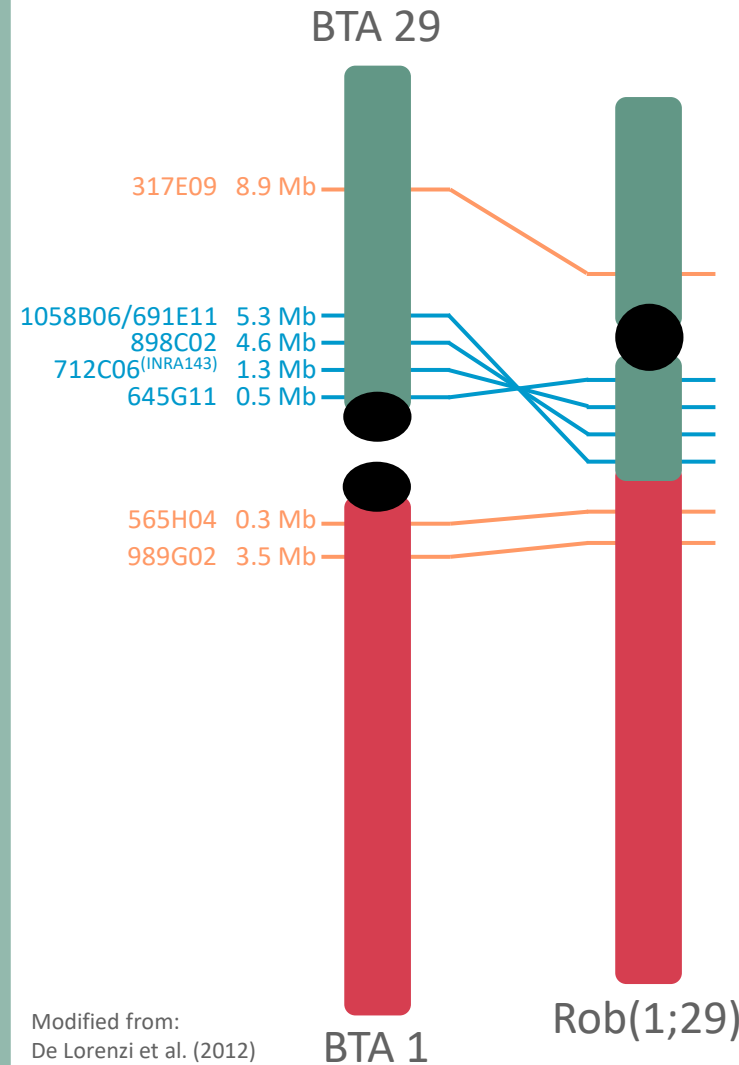
01

# Introduction

# Robertsonian translocation 1;29

Chromosomal rearrangement involving the fusion of two acrocentric chromosomes (BTA 1 and 29) across their pericentromeric regions. No lack or gain of genetic material.

No meiotic recombination in the first centromeric region of BTA 29 due to the **inversion of a cosmid** around 5.4 Mb and its **migration** onto the q arm of rob(1;29).



# Frequency in cattle

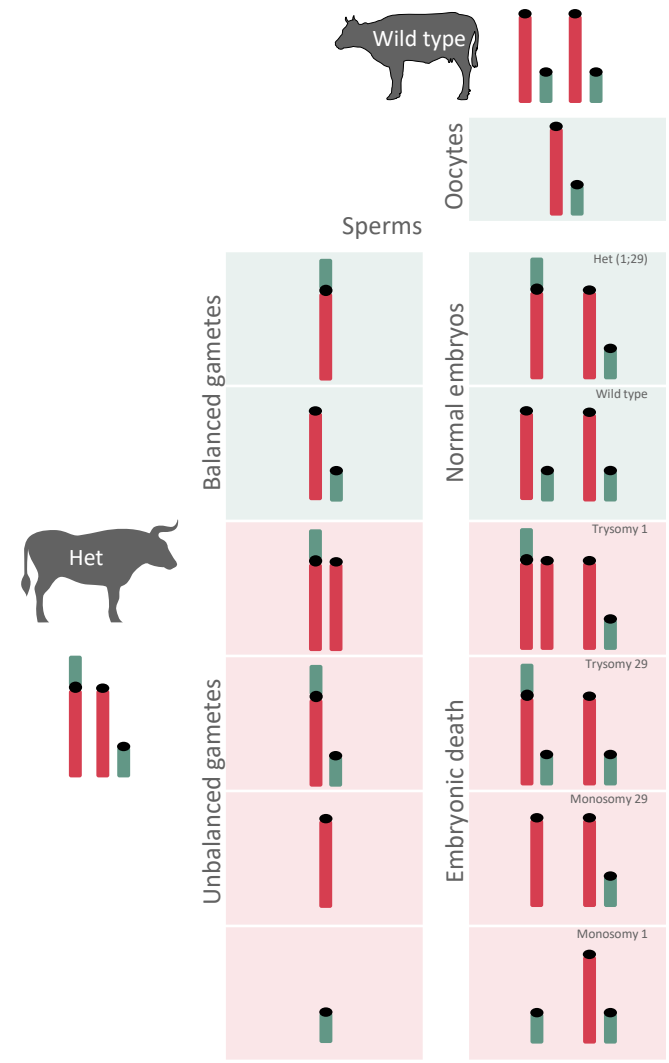
Rob(1;29) is known since 1964 and is the **most common** chromosomal aberration in cattle.

Mainly found in breeds aimed at **meat** production.

| Chromosomes Involved in Rob Translocation (Centric Fusion) | Breed of Cattle                   | Frequency   | Year      | Reference |
|------------------------------------------------------------|-----------------------------------|-------------|-----------|-----------|
| rob(1;29)                                                  | Swedish Red and White cattle      | 13–14%      | 1964      | [35]      |
|                                                            | Over 50 breeds of cattle          |             | 1964–2014 | [31,32]   |
|                                                            | British White                     | Up to 60%   | 1975      | [33]      |
|                                                            | Corsican                          | Up to 60%   | 1984      | [34]      |
|                                                            | local Portuguese cattle           | above 50%   | 2008      | [42]      |
|                                                            | Maremmana                         | 18.8%       | 2008      | [26]      |
|                                                            | Romagnola                         | 13.0%       |           |           |
|                                                            | Podolian cattle                   | 11.7%       |           |           |
|                                                            | Marchigiana                       | 11.7%       |           |           |
|                                                            | Chianina                          | 1.4%        |           |           |
|                                                            | Limousine                         | 12.3%       | 2008      | [26]      |
|                                                            | blonde d'Aquitaine                | 7.9%        |           |           |
|                                                            | Charolaise                        | 1.2%        |           |           |
|                                                            | Rubia Gallega                     | 21.9%       | 2008      | [26]      |
|                                                            | Retinta                           | 16.1%       |           |           |
|                                                            | Czech Simmental                   | 27.08%      | 2009      | [27]      |
|                                                            | Andalusian breeds: Negra Andaluza | 19.45%      | 2013      | [43]      |
|                                                            | Berrenda en Negro                 | 28.9%–32.6% |           |           |
|                                                            | Criollo                           | 12.3%       | 2015      | [44]      |
|                                                            | Swiss American                    | 7.5%        |           |           |
|                                                            | Braunvieh (Swiss Brown)           | 1.4%        |           |           |
|                                                            | Holstein                          | 0.4%        |           |           |

# Effects on fertility

Het rob(1;29) carriers show **impaired infertility** due to the production of genetically **unbalanced gametes** and, consequently, unbalanced embryos that cannot survive.

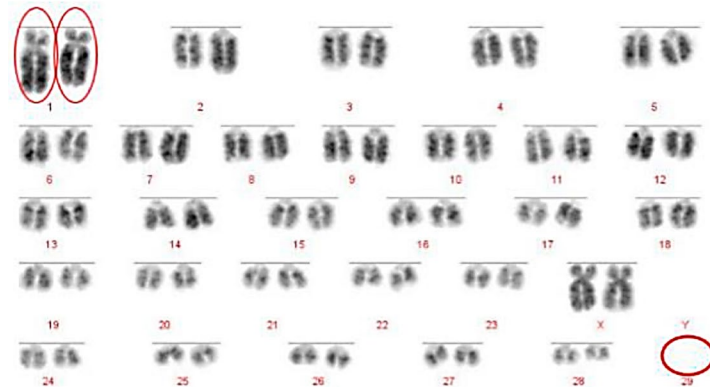


# Screening

Screening programs are in place in many European countries, based on **karyological tests**.

However, only a limited part of the population (e.g., bulls for breeding) is screened due to:

- High costs and prolonged time
- Need for fresh blood samples
- Specialized personnel requirements



Can **SNP data** routinely  
collected by breeder  
associations be used to  
**identify rob(1;29) carriers?**

02

# Methods and Results



# DATASETS

**Medium-density SNP data** were retrieved from public data, newly genotyped animals or breeder association datasets.

For a subset of individuals a **cytogenetic test** was available.



|                    | Dataset 1                    | Dataset 2                | Dataset 3                                     |
|--------------------|------------------------------|--------------------------|-----------------------------------------------|
| Data origin        | ANABIC screened subjects     | BOVITA public dataset    | Breeder associations                          |
| N. Breeds          | 5 Italian beef cattle breeds | 32 Italian cattle breeds | 9 Italian cattle breeds                       |
| N. Subjects        | 151                          | 800                      | 1711                                          |
| N. Cytogetic tests | 1 hom, 47 het, 103 wild type | -                        | 1 hom, 47 het, 105 wild type                  |
| SNPchip            | GGP Bovine 33k               | Illumina 50K             | GGP Bovine 33k + Illumina 150k + Illumina 50K |
| N. Subset SNPs     | 75                           | 59                       | 72                                            |





# FST

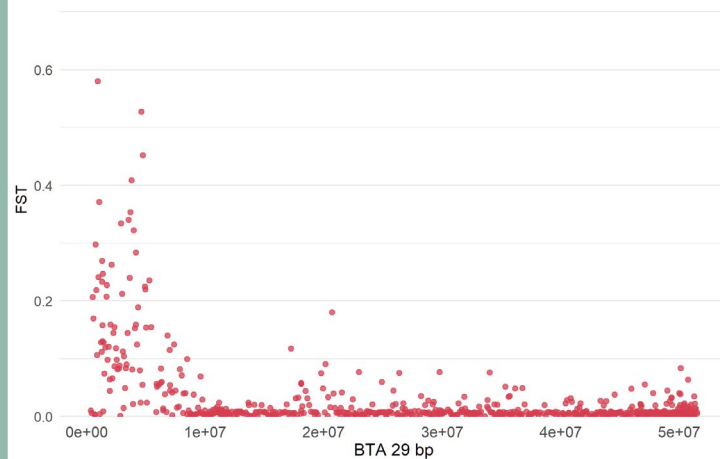
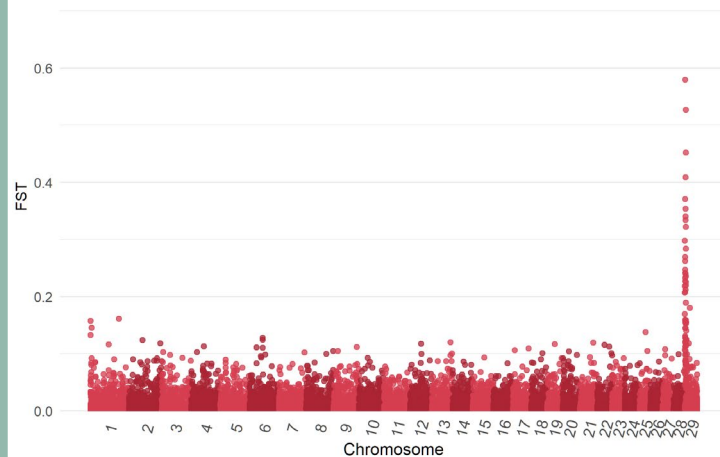
Subjects of Dataset 1 with a cytogenetic diagnosis of being wild type or rob(1;29) carriers were compared through FST analysis.



# FST

## wild type vs carriers

- No peaks on BTA 1
- Clear peak on the **first 6 Mb of BTA 29**
- This regions corresponds to the one involved in the inversion of translocated chromosomes
- For further analyses, we only used this highlighted region





# HAPLOTYPE ANALYSIS

The **haplotype** of the region with peaking FST values, i.e., the first 6 Mb of BTA 29, was compared between the hom rob(1;29) carrier and the het carriers or the wild type subjects.



Dataset

1

Wild type had **max 64 SNPs (85%)** in common with the hom subject.

Het individuals had **at least 71 SNPs (99%)** in common with the hom subject.

Dataset

2

Wild type had **max 50 SNPs (85%)** in common with the hom subject.

Het individuals had **all the genotyped SNPs (100%)** in common with the hom.

Dataset

3

Wild type had **max 61 SNPs (85%)** in common with the hom subject.

Het individuals had **at least 66 SNPs (96%)** in common with the hom subject.



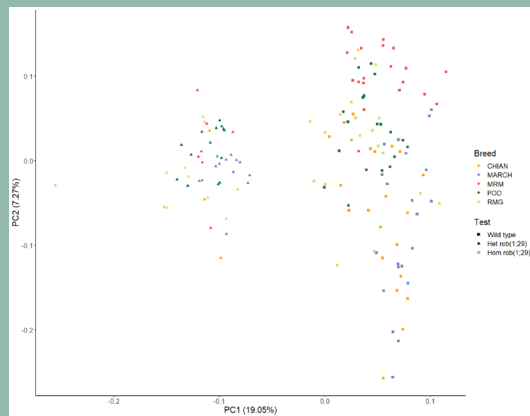


# PCA

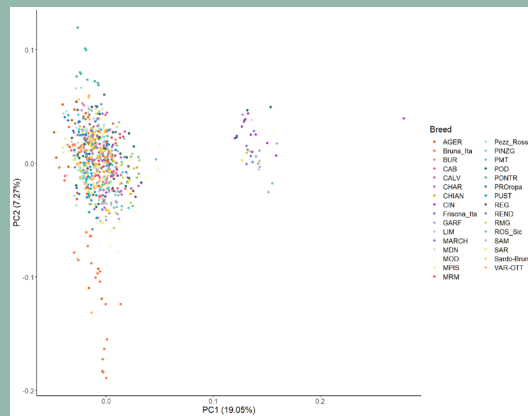
A **principal component analysis (PCA)** was performed on all the subjects using only the highlighted region, to assess if a **separation among the three groups** was visible.



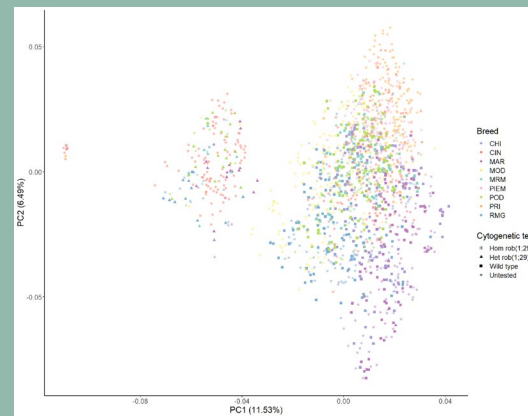
# Dataset 1



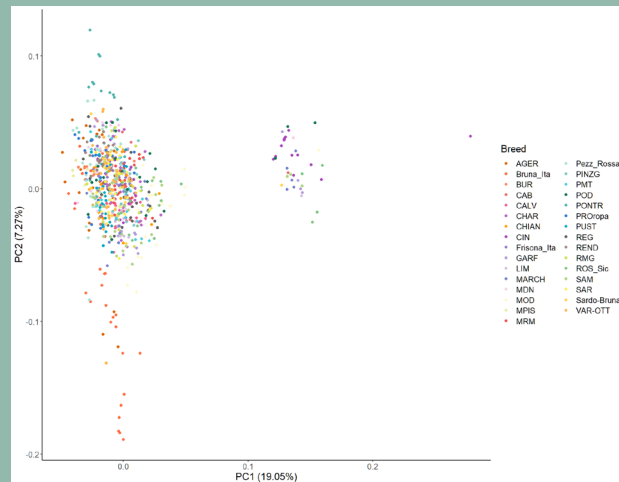
# Dataset 2



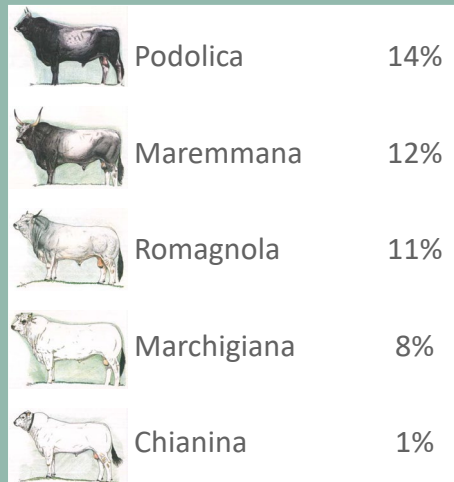
# Dataset 3



## Dataset 2

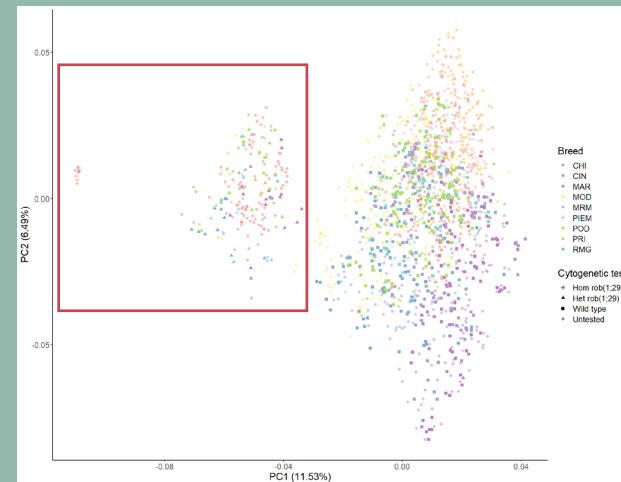


## Rob(1;29) frequency in population for screened breeds

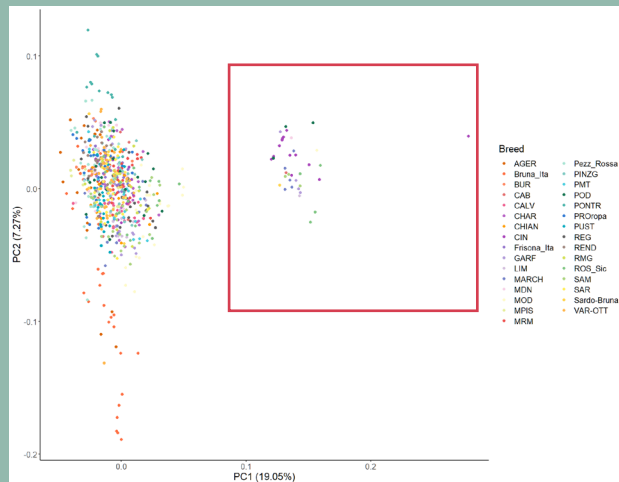


Thanks to ANABIC association for data and support.

## Dataset 3



## Dataset 2



Breeds with no carriers  
according to both  
datasetes

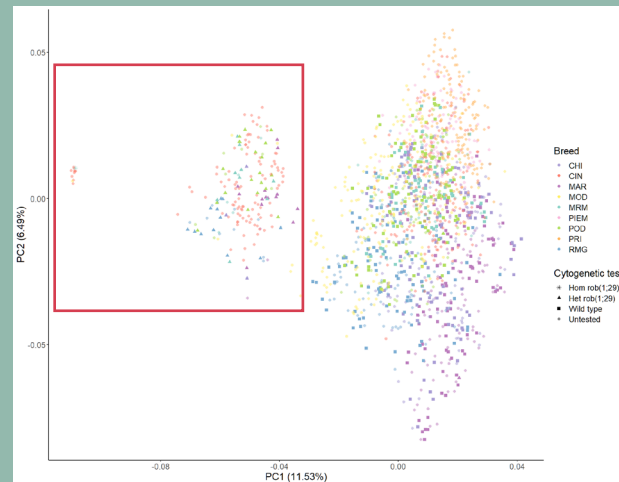
Pezzata Rossa Italiana  
(Italian Red Simmenthal)



Piedmontese



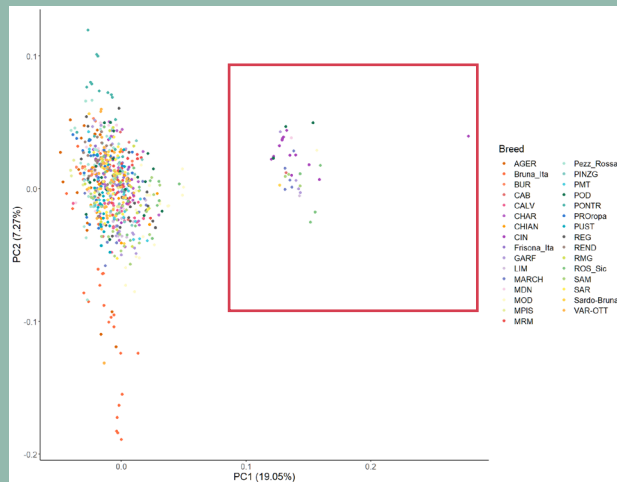
## Dataset 3



Thanks to ANAPRI and ANABORAPI associations for  
data and support.



## Dataset 2

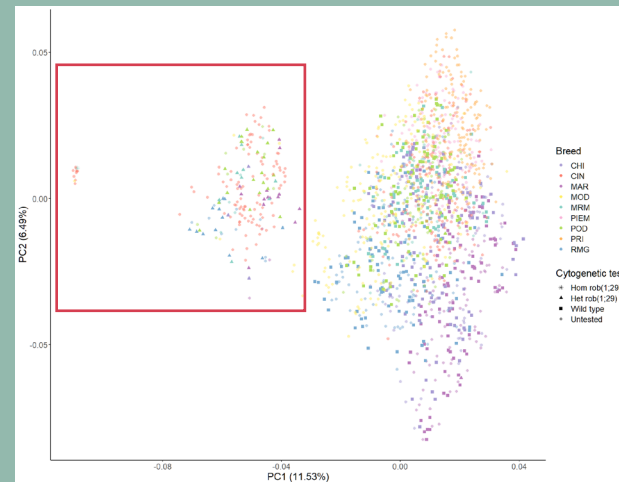


## Unscreened breeds with high rob(1;29) frequency

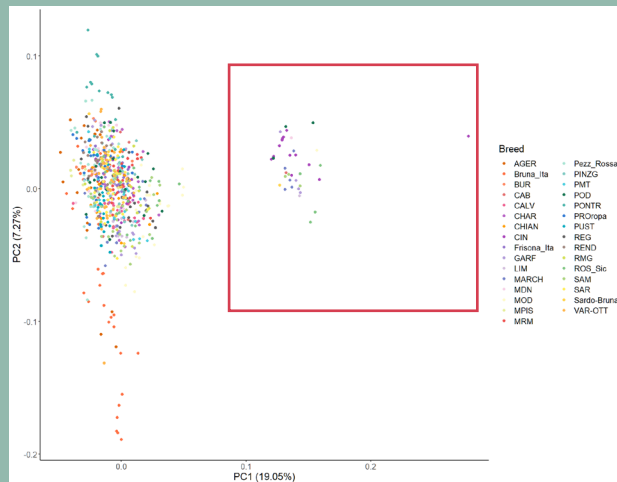
|                                                                                   |                 |      |
|-----------------------------------------------------------------------------------|-----------------|------|
|  | <u>Cinisara</u> | ~40% |
|  | Rossa Siciliana | 21%  |
|  | Garfagnina      | 13%  |
|  | Sardo-Modicana  | 11%  |
|  | Sardo-Bruna     | 10%  |
|  | Limousine       | 5%   |
|  | Modenese        | 4%   |
|  | <u>Modicana</u> | ~3%  |

Thanks to ANAPRI association and Dual Purpose project for data and support.

## Dataset 3



## Dataset 2

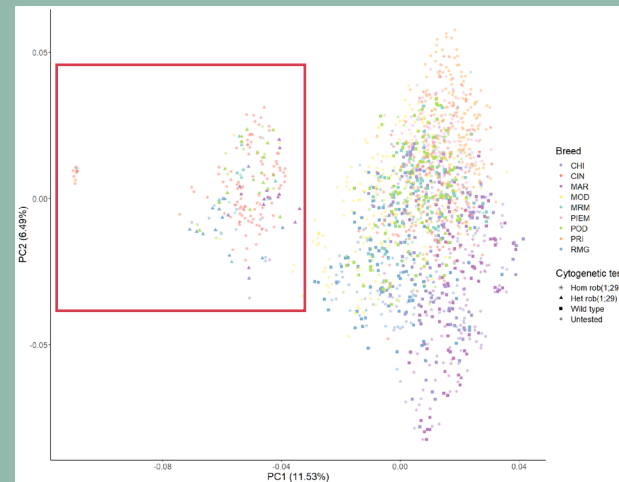


## Cinisara breed



- Dual-purpose Sicilian breed
- **Podolian** origin
- High frequency found both in Dataset 2 and 3
- Presence of **homozygous** carriers
- **Confirmed with cytogenetic tests**
- No fertility problems?

## Dataset 3





# PARTITIONING ANALYSIS

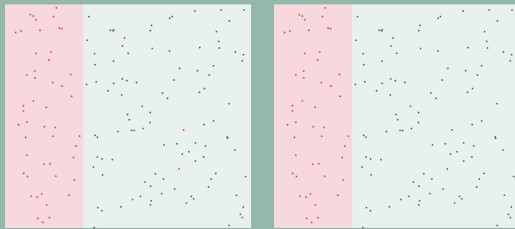
Using JMP 16, a partitioning analysis was conducted on the SNPs belonging to the haplotype to identify **the smallest possible number of SNPs capable of distinguishing translocated subjects from wild type ones.**



## Dataset 1

We identified a **single SNP** that, alone, identified all the carriers, but also 4 false positives (96.2% specificity).

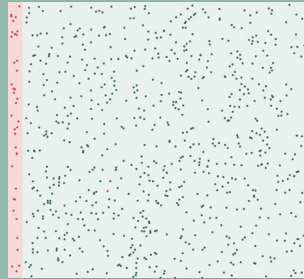
When **adding also a second one**, we obtained 100% specificity and 100% sensitivity.



N = 151, GGP Bovine 33k

## Dataset 2

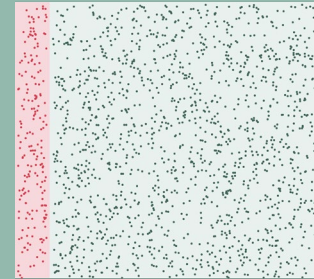
A **single SNP** that was not present in other arrays discriminated carriers and wild type with a single false positive (99.9% specificity and 100% sensitivity).



N = 800, Illumina 50k

## Dataset 3

**Three specific SNPs** (two of which were those found in Dataset 1) were enough to correctly identify all the individuals with 100% specificity and 100% sensitivity.



N = 1711, GGP Bovine 33k + Illumina 150k + Illumina 800k



03

# Conclusions

01

## SNP-based screening

**SNP data** represent a practical and cost-effective tool for **widespread rob(1;29) pre-screening** and prompt identification of carriers.

02

## Integration into panels

We recommend **adding SNPs** from this region **to SNP panels** (e.g., ISAG SNP Parentage Panel).

03

## Common haplotype

All carriers shared the same haplotype supporting the theory of a **common ancestor**.

04

## Breed frequencies

**High frequency in unscreened populations**, such as the Cinisara breed.

# Thanks!



**scientific** reports

## Identification of a common haplotype in carriers of rob(1;29) in 32 Italian cattle breeds

Matteo Cortellari, Arianna Bionda , Luigi Liotta, Fiorella Sparra, Pietro Parma & Paola Crepaldi

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