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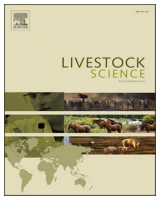

Invited review: the application of runs of homozygosity and heterozygosity-rich regions to infer population history in livestock species

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HIGHLIGHTS

- ROH are valuable genomic tools for quantifying individual inbreeding and identifying regions under selection in livestock populations.
- HRR are potentially maintained by balancing selection and linked to fitness-related traits, such as immunity and fertility.
- Joint analysis of ROH and HRR enhances our understanding of genetic diversity patterns and supports the development of more sustainable breeding programs.
- ROH and HRR have practical implications for managing inbreeding depression, maintaining adaptive genetic variation, and designing conservation strategies for local or endangered breeds.

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ABSTRACT

Advancements in high-throughput and cost-effective genotyping techniques, coupled with robust statistical methods, have greatly facilitated the investigation of the genomic architecture of farm animals. Runs of Homozygosity (ROH) and Heterozygosity-Rich Regions (HRR) are among the most informative features of animal genomes. ROH reflects inbreeding levels and historical population dynamics, whereas HRR may indicate loci under balancing selection that contribute to important functional traits. In this review, these two genetic parameters are described and discussed. We present an updated summary of ROH studies and applications in livestock species, as well as a comprehensive overview of the status of knowledge on HRR in livestock populations. Additionally, we discuss methods for the identification of ROH and HRR, and the insights they offer on the demographic history of animal species. We investigated the link between the presence, distribution, and specific characteristics of ROH and HRR, and the breeding and selection trajectories of farm animals. ROH- and HRR-abundant regions often harbour genes associated with economically important traits and local adaptation, underscoring the significant role of artificial and natural selection in shaping the genomic architecture of livestock. The increasing availability of high-density genotyping and whole-genome sequencing data allows for the finer-scale detection of ROH and HRR, enabling a more accurate identification of functionally relevant genomic regions. The future application of ROH and HRR in livestock genetics will likely move toward more integrated, data-driven strategies aimed at improving genetic resilience, productivity and sustainability.

1. Historical perspective on ROH and HRR

Homozygosity and heterozygosity are two fundamental parameters of farm-animal population genetics. Homozygosity typically arises when parents share a common ancestor and transmit identical chromosomal segments to their offspring. According to Wright (1922), the probability that two alleles are identical by descent (IBD) increases with inbreeding. In modern terms, this can be interpreted as leading to homozygous segments in the offspring's genome. When homozygous segments arise for reasons other than direct and traceable parent-offspring

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transmission, such as convergent mutations, genetic drift, positive or negative directional selective pressure, or technology (e.g., cloning, genome-editing), we talk about identity by state (IBS). Therefore, IBS and IBD are related concepts, but they differ in their underlying mechanisms. IBS refers to alleles that are the same regardless of their origin, while IBD refers to alleles that are the same because they are inherited from a common ancestor. While a genomic region that is IBD must also be IBS, the opposite is not necessarily true (Henden et al., 2018).

Heterozygosity can arise or be maintained through several processes, including interbreeding between genetically distinct individuals or populations (e.g., crossbreeding, introgression and admixture), the introduction of novel alleles through mutation, and evolutionary and demographic processes such as balancing selection, gene flow, migration, and large effective population size, which reduces the loss of genetic variation due to drift. Technology can also play a role when novel mutations are introduced artificially (e.g., genome editing, recombinant DNA). Simply put, heterozygosity increases genetic diversity, while homozygosity reduces it. When the objective is to investigate properties of the entire genome or animal population under study, rather than the status of a single locus of interest (Mulim et al., 2022), segments or regions of homozygosity or heterozygosity are generally more informative than single genomic sites, for two main reasons: i) they reduce the noise associated with single loci, which may be homozygous or heterozygous by chance or as a result of genotyping errors; ii) they may reflect true demographic processes, like autozygosity, defined as homozygosity resulting from IBD, or introgression.

The first study to identify long homozygous chromosomal segments in humans was conducted by Broman and Weber (1999). They proposed that extended homozygous regions likely reflect autozygosity and could have potential implications for human health. Gibson et al. (2006) were the first to fully recognize the significance of this finding by examining the patterns and distribution of runs of homozygosity (ROH) in outbred HapMap populations. However, ROH were first defined formally by Lencz et al. (2007), in a study on genetic loci associated with the risk of schizophrenia, as any window of 100 or more consecutive SNP on a single chromosome without any intervening heterozygous SNP. Soon after, McQuillan et al. (2008) used SNP data to estimate individual autozygosity from ROH, thus introducing the genomic inbreeding coefficient F_{ROH}: the sum of the lengths of all ROH segments in an individual as a percentage of the total length of the autosomal genome. This approach has since been used extensively in human genetics to study population history, disease genetics, and inbreeding (e.g., Nothnagel et al., 2010; McQuillan et al., 2012; Pemberton et al., 2012; Ceballos et al., 2018).

With the increasing availability of high-throughput genomic data beyond humans, ROH started to be studied also in farm animals, initially in cattle (Sölkner et al. 2010; Ferenčaković et al. 2011; Purfield et al. 2012; Ferenčaković et al. 2013a; Kim et al. 2013; Mastrangelo et al., 2016), and subsequently in other livestock species, such as sheep (e.g. Mastrangelo et al., 2017; Purfield et al., 2017; Luigi-Sierra et al., 2019), goat (e.g. Brito et al., 2017; Bertolini et al., 2018; Onzima et al., 2018), pig (e.g. Bosse et al., 2012; Herrero-Medrano et al., 2013; Schiavo et al., 2021), horse (e.g. Metzger et al., 2015; Grilz-Seger et al., 2018), buffalo (e.g. Ghoreishifar et al., 2020; Macciotta et al., 2021), rabbit (e.g. Ballan et al., 2022; Ping et al., 2025), and chicken (e.g. Marchesi et al., 2018; Zhang et al., 2020; Talebi et al., 2020). Similarly, F_{ROH} has been extensively applied in livestock populations as a measure of genomic inbreeding.

Interest in genomic regions characterised by high heterozygosity emerged more recently and, in contrast to ROH, initially in animals, with the seminal work by Williams et al. (2016) on Chillingham white cattle. These genomic regions were initially named "runs of heterozygosity", in analogy with ROH. However, in most cases they cannot be considered as true runs, since SNPs arise from the process of variant calling, i.e. looking for polymorphisms in the genome, and what is not polymorphic, is by definition monomorphic, hence homozygous (inter-variant regions). Consequently, the base pairs between genotyped heterozygous SNPs are not necessarily all heterozygous, even with whole-genome sequence data. In contrast, in ROH, all positions between consecutive variants can generally be considered homozygous, except for differences due to variant density and genotyping technology. Therefore, Ferenčaković et al. (2016) proposed the term heterozygosity-rich regions (HRR), which better reflects their nature and is now the accepted terminology in the scientific community. HRR are defined as genomic regions characterised by continuous high heterozygosity in diploid organisms (Li et al., 2022). Accumulated over time, this increased heterozygosity may in some cases be attributed to balancing selection, which maintains allelic diversity within populations through mechanisms such as heterozygote advantage (Fijarczyk and Babik, 2015). Irrespective of their origin, HRR plays a pivotal role in preserving adequate genetic diversity in animal populations and may harbor loci associated with key functional traits such as immune response (Mulim et al., 2022) and other fitness traits (Selli et al., 2021; Chessari et al., 2024). Given the relatively recent introduction, the literature about HRR is not as abundant as for ROH. Few recent studies have detected HRR in cattle (e.g., Ferenčaković et al., 2016; Biscarini et al., 2020; Mulim et al., 2022), sheep (Selli et al., 2021; Tsartsianidou et al. 2021; Szmatoła et al., 2025; Carta et al., 2026), goats (Li et al., 2022; Chessari et al., 2024), horses (Bizarria dos Santos et al., 2021), and pigs (Chen et al., 2022; Bordonaro et al., 2023; Fabbri et al., 2025). However, there are still substantial knowledge gaps and the need for more systematic research that encompasses a broader range of livestock species, methodological approaches and that elucidates the causal mechanisms (e.g., balancing selection). A deeper comprehension of HRR regions might offer valuable insights into population diversity, evolutionary history, and the genomic underpinnings of complex traits in domestic animals.

Besides individual articles on specific topics, ROH have been the subject of a few review articles. Curik et al. (2014) reviewed methods to estimate inbreeding using genomic data, highlighting the importance of ROH in livestock, humans, and (diploid) plants. Peripolli et al. (2017) focused on the identification, characterization, and practical implications of ROH in animal populations. Additional reviews have been published on ROH in farm (Nosrati, 2017; Rebelato and Caetano, 2018) and wild (Shafer and Kardos, 2025) animals. In contrast, HRR are far less described in literature and have not been the subject of any review work, in farm animals, humans, or any other species. In this review, we therefore provide an updated overview of methods, studies, and applications of ROH in livestock species, along with a comprehensive summary of current knowledge on HRR in animals. Additionally, we discuss how ROH and HRR have been used to investigate patterns of genetic diversity and demographic history.

2. Measuring the genome: methods for the detection of ROH and HRR

"*Metior, ergo sum*" (paraphrasing the famous motto of rationalism): the ability to measure natural phenomena is at the root of the process of advancing our knowledge from a scarcely organised heap of thoughts to the stage of science (Kelvin, 1891). This also applies to the extent of homozygosity and heterozygosity in animal genomes. These two parameters are central to population genetics: compared to the use of single-site genotypes, with or without genealogical data, ROH and HRR offer a more informative and structured way to measure homozygosity and heterozygosity at the genomic level. They allow us to get more robust and less noisy estimates of these two genetic parameters, to investigate their distribution across the genome, and to develop a wider range of applications (e.g., metrics to measure inbreeding, tools to identify signatures of selection, etc.).

Two main approaches have emerged for detecting ROH and HRR across the genome: i) a method based on overlapping sliding windows that move sequentially along the genome; ii) a method that does not

make use of any windows and just scans the genome locus by locus. Given that ROH and HRR have so far been defined only for diploid genomes, and that all the livestock species considered in this review are diploid, we restrict our discussion to diploid genomes only.

The sliding-window approach was the first to be developed for the detection of ROH (and later on, *mutatis mutandis*, of HRR) in the genome (McQuillan et al., 2008; Howrigan et al., 2011). Briefly, a fixed width sliding window is used to scan the genome moving by one SNP at a time (stride= 1), and the characteristics of consecutive partially overlapping windows are used to determine whether a SNP is or not in a run (either ROH/HRR). Fig. 1 presents a simplified illustration: a series of 7-SNP sliding windows, one SNP apart from each other, is built along the genome; at each SNP, we count the total number of overlapping windows and the number of homozygous windows (where all 7 SNP are homozygous). The SNP for which the ratio between these two numbers (n. of homozygous windows / total. n. of windows) is larger than a threshold (10% in this example) is considered to be part of a ROH (the same holds, *mutatis mutandis*, for HRR (see Fig. 2)). Several parameters need to be specified to detect ROH/HRR in the genome, and these will affect the calculations outlined above. In the sliding-window approach, two sets of parameters are needed: one for the sliding windows and one for the run (be it ROH or HRR). The sliding window parameters are: the size of the window in n. of SNP (7 SNP in the example in Fig. 1), the threshold of homozygous (heterozygous for HRR) windows as fraction of the total windows above each SNP, and the maximum number of missing SNP or opposite-genotype SNP allowed in a window to call it homozygous/heterozygous. The parameters that control the genomic runs are: the minimum number of SNP, the minimum length (bps: base pairs), the minimum density (SNP/kbps) to call a run, the maximum gap (bps) allowed between adjacent SNP not to break (interrupt) the run, and the maximum number of missing SNP or opposite-genotype SNP allowed not to discard a run.

The "consecutive runs" method stemmed as a simplification of the sliding-window approach: it is window-free, and it directly scans the genome, SNP by SNP (Marras et al., 2015). The genotype of each SNP is recorded until, given the specified run parameters, a ROH/HRR is called. If the minimum requirements specified by the parameters are not met, that stretch of genome is discarded (not considered as a run). For this method, obviously, there is only one set of parameters, the ones about the runs (which are the same parameters described for the sliding window approach). Figs. 1 and 2 illustrate, with simplified numerical examples, the sliding-window and consecutive methods for the detection of ROH and HRR. While we did not allow for any heterozygous SNP in the example ROH, we chose to allow for a maximum of two homozygous SNP (opposite genotype) in the detection of HRR. We did this on purpose, to show that, unlike ROH, HRR are not "runs" (i.e. continuous stretches of heterozygous genome), but rather regions with excess heterozygosity.

The choice of whether to use a sliding-window or contiguous-SNP approach has significant implications. Window-based methods smooth local fluctuations and are computationally efficient, but they may mask fine-scale recombination patterns and introduce dependency on window size and step length. Conversely, contiguous-SNP approaches can capture true homozygous tracts with higher precision but are more sensitive to genotyping errors and SNP density variation. Although only few studies have focused on identifying HRR islands with both methods of

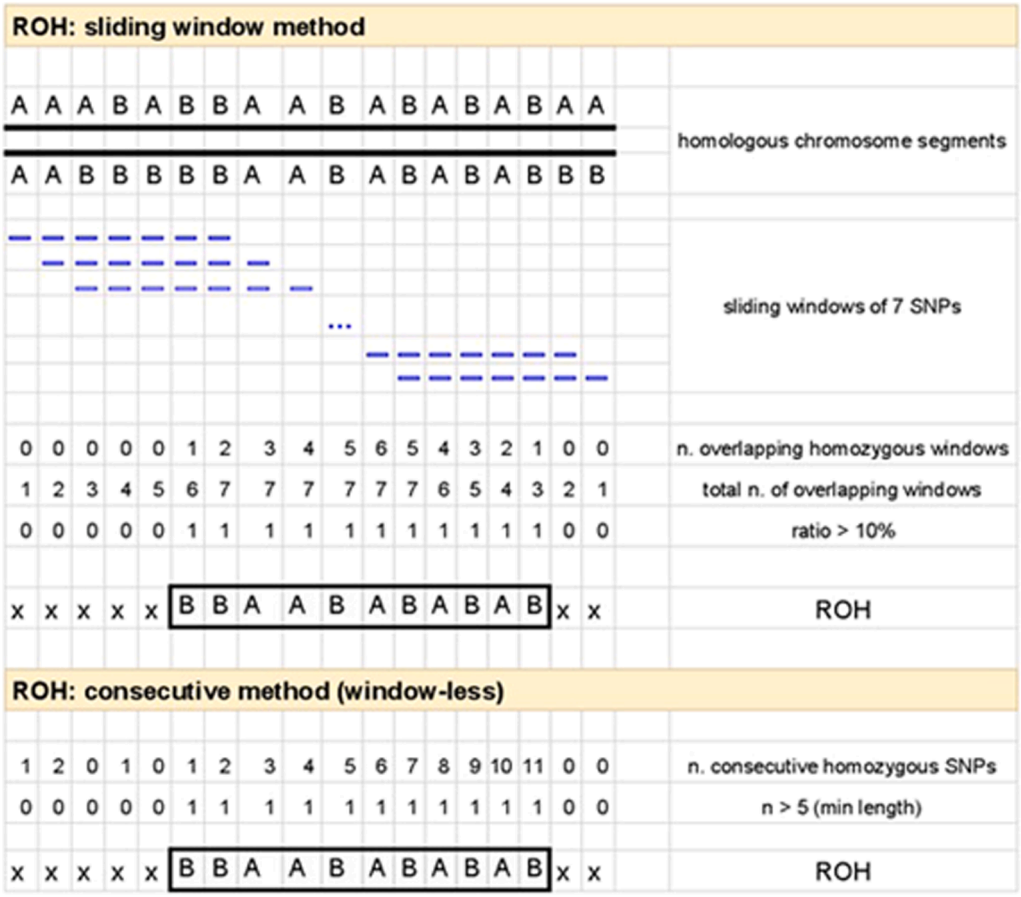

Fig. 1. Graphical illustration of the sliding-window approach to the detection of Runs of homozygosity (ROH). The sliding-window parameters are: size= 7 SNP; fraction of homozygous overlapping windows= 10%; max missing and heterozygous SNP in the homozygous window= 0. The ROH parameters are minimum ROH size as n. of SNP= 5; max missing and heterozygous SNP in ROH= 0 (minimum length in bps, minimum density (SNP/kbps), max gap (bps) between SNP were not considered). Adapted from Bjelland et al. (2013).

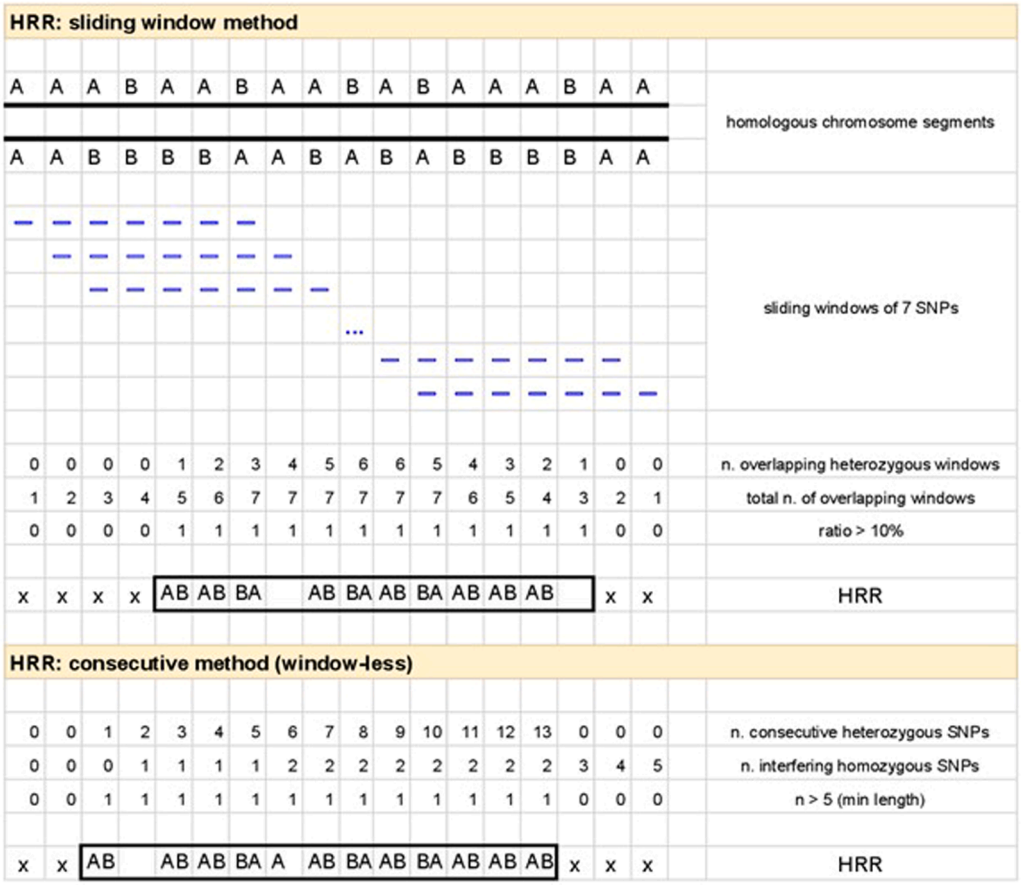

Fig. 2. Graphical illustration of the sliding-window approach to the detection of heterozygosity rich regions (HRR). The sliding-window parameters are: size= 7 SNP; fraction of heterozygous overlapping windows= 10%; max missing SNP in the heterozygous window= 0; max homozygous SNP in the heterozygous window= 1. The HRR parameters are: minimum HRR size as n. of SNP= 5; max missing SNP in HRR= 0; max homozygous SNP in HRR= 2 (minimum length in bps, minimum density (SNP/kbps), max gap (bps) between SNP were not considered).

detection, it seems that the use of the "consecutive runs approach" is preferred (Bizarria dos Santos et al., 2021; Mulim et al., 2022). Previous studies (Bordonaro et al., 2023; Chessari et al., 2024), showed overlapping results for HRR identified from both approaches. A recent study reported that results obtained using the sliding window approach implemented in PLINK and in the detectRUNS package are very similar, as expected, since these are two software implementations of the same statistical method (Nisa et al., 2025). Larger discrepancies are observed in the results from the consecutive run approach, given the fundamentally different statistical algorithm used to detect ROH/HRR. It should be noted that these differences mainly relate to locally detected ROH/HRR, whereas average properties like the inbreeding coefficient are less affected. These methodological differences can influence the detection and interpretation of genomic regions, highlighting the importance of tool selection and parameter tuning in ROH/HRR studies. Which statistical approach and which parameters to use remain unresolved and may depend on a case-by-case basis.

2.1. Main software for the detection of ROH and HRR

After the formal definition of ROH, specialised modules in more general software packages for the analysis of genomic data began to be developed. There are basically two software packages that implement the methods for the detection of ROH and HRR described above (the "sliding window" and "consecutive runs" approaches): Plink and detectRUNS. Plink (C/C++) was the first package to provide geneticists with tools for the detection of ROH in diploid genomes, already since version 1.07 (Purcell et al., 2007), based on the sliding-window method described in Bjelland et al. (2013). Plink takes in input the native ped/map file format but also accepts the standard vcf file format. DetectRUNS (Biscarini et al., 2019) is an R package (with some functions written in C++ for computational efficiency) that implements both the "sliding window" and "consecutive runs" methods and scans diploid genomes for both ROH and HRR. DetectRUNS is currently the only available software to detect HRR and to implement the consecutive runs method. As of now, detectRUNS only accepts non-binary ped/map files as input. Additional software packages for the detection of ROH (but not HRR) are the C program bcftools (Narasimhan et al., 2016) and the R package RZooRoH (Bertrand et al., 2019), both based on probabilistic approaches that implement Hidden Markov Models (HMM) for the identification of autozygous segments of diploid genomes. More software exists that scans diploid genomes for something similar, but not exactly equal, to ROH and HRR. GERMLINE (Genetic Error-tolerant Regional Matching with Linear-time Extension; *C++*; Gusev et al., 2009) and BEAGLE (Java; module Refined-IBD) (Browning and Browning, 2010) focus on the detection of IBD (identical by descent) segments, including HBD (homozygous by descent) segments. ROHan (*C++*) uses whole-genome sequence data to estimate local and global rates of heterozygosity, and to infer ROH (Renaud et al., 2019). When taking the special case of IBD within individuals, GERMLINE detects ROH based on non-overlapping sliding windows. BEAGLE and ROHan are based on HMM approaches to estimate IBD haplotypes and heterozygosity rates (ROH are a byproduct of this latter in ROHan), respectively. Table 1 summarises the most salient characteristics of the mentioned packages. The relative performance of Plink, Beagle, and GERMLINE has been compared in previous works (Howrigan et al.,

Table 1
List of software packages for the detection of Runs of homozygosity (ROH) and heterozygosity rich regions (HRR) in diploid genomes.

Software	Language	Released	Version	ROH	HRR	Method	Input format data	link(s)
Plink	C/C++	2007	1.9/2.0	yes	no	sliding windows	ped/map; vcf; gz files	https://zzz.bwh.harvard.edu/plink/contact.shtml#cite https://www.cog-genomics.org/plink/1.9/; https://www.cog-genomics.org/plink/2.0/
detectRUNS	R/C++	2018	0.9.6.9004	yes	yes	sliding windows, consecutive loci	ped/map	https://cran.r-project.org/web/packages/detectRUNS; https://github.com/bioinformatics-ptp/detectRUNS
GERMLINE	C++	2009	GERMLINE2	yes	no	non-overlapping sliding windows	ped/map; haps/samples/map	https://github.com/gusevlab/germline https://github.com/gusevlab/germline2
ROHan	C++	2019	v1.0.1	yes	no	HMM	bam / fasta	https://grenaud.github.io/ROHan/
bcftools	C	2016	v1.22	yes	no	HMM	vcf	https://samtools.github.io/bcftools/bcftools.html#roh
RZooRoH	R/ Fortran	2018	v0.4.1	yes	no	HMM	Oxford/GEN format	https://cran.r-project.org/web/packages/RZooRoH/
Beagle	Java	2013	Beagle 4.0/ 4.1	yes (IBD)	no	Refined IBD/HMM	vcf	http://faculty.washington.edu/browning/refined-ibd.html

2011).

2.2. Data sources and preprocessing

When scanning the genome for the detection of ROH or HRR, special attention must be paid to the input data source and preprocessing. Detection methods were originally developed on SNP-array data, i.e., genotyping data. Subsequent technological advances in genome sequencing and genotyping led to the widespread availability of SNP arrays with different marker densities and, more recently, to the increasing use of whole-genome sequence (WGS) data.

SNP density strongly influences the efficiency of ROH and HRR detection, albeit to different degrees. When low density SNP arrays are used, there is an inflation of long ROH, due to a failure to identify possible intercurring heterozygous SNP, while few short ROH are detected, given the average large distances between adjacent SNP. Ferenčaković et al. (2013b), in a study on three cattle populations, showed that the 50 K panel overestimated the numbers of ROH segments 1–4 Mb long, suggesting insufficient sensitivity for the accurate detection of short ROH. Similarly, as SNP array density increases, more ROH are detected, although their average length tends to decrease (Purfield et al., 2012; Falchi et al., 2024). Interestingly, very long ROH segments (e.g., >8 Mb) may still be underestimated even with high-density panels, likely due to methodological limitations in identifying contiguous homozygous stretches (Ferenčaković et al., 2013b). In contrast, the number of HRR appears to decrease, while their average length increases, with increasing SNP density (Falchi et al., 2024). These observations suggest that SNP density clearly influences both the length and number of detectable ROH/HRR, potentially leading to false discoveries (i.e., spurious ROH/HRR). This issue is particularly relevant when focusing on individual ROH/HRR segments or population-specific islands/regions. This potential local bias tends to average out when looking together at the ensemble of ROH/HRR in one animal or one population. As a matter of fact, unlike the detection of single ROH segments, genome-wide metrics such as F_{ROH} are generally consistent across different SNP densities (e.g., F_{ROH}= 0.73 vs 0.70, Purfield et al., 2012). This holds well within reasonable SNP density boundaries, as shown by Meyermans et al. (2020): when the SNP density is too low, nothing can be detected (the gap is too wide, the number of SNPs is too low etc.); when SNP density increases, F_{ROH} suddenly (and very rapidly) increases, and quickly plateaus to a stable value.

When using WGS data, results on the detection of ROH and HRR apparently do not agree with those from SNP array data (Mulim et al., 2022). Minn et al. (2025) demonstrated that a substantial number of ROH, particularly short ones that are often missed with SNP-array data, can be reliably detected using WGS. By filtering sequencing errors and incorporating pedigree information, longer ROHs are also more accurately identified in WGS than in SNP-array datasets. Such disagreements have been sometimes dealt with by tweaking the detection parameters values so as to get similar results from both data sources (Bosse et al. 2012; Zhang et al. 2015). It's worth mentioning that WGS data can help circumvent problems with ascertainment bias, which may be present when using SNP arrays on local breeds. All this may call for a rethinking of the definition and detection method for genomic runs with WGS data, where SNP density and distribution, occasional sequencing errors and new individual mutations may influence the results. Further studies will be required to clarify how to use sequencing data, from both long- and short-read technologies, to obtain the most reliable SNP genotyping and, consequently, more robust inference of ROH and HRR (Smaragdov, 2025).

Typical preprocessing of genomic data includes: (i) filtering out poor-quality or non-informative samples and variants; (ii) imputation of missing SNP genotype data. Variants and samples can be filtered for missing rate, since a high missing rate can, on one hand, be indicative of low-quality input data (something went amiss during genotyping, sequencing, alignment), and on the other, be challenging for the imputation algorithms, thereby reducing the accuracy of imputing missing data points. In both cases, too much noise might be left or introduced in the data, increasing the probability of getting unreliable results. Whether or not to filter for minor allele frequency (MAF) or minor allele count (MAC) must be carefully considered. First, are we looking for ROH or HRR? If it's ROH we are after, removing monomorphic SNP is likely to be counterproductive, since you would be removing exactly those highly homozygous stretches of the genome that you intend to detect. Meyermans et al. (2020) reported that MAF pruning can severely impact ROH analyses and suggested skipping MAF pruning prior to ROH analysis. However, when analysing multi-breed datasets, the primary interest may be the comparison of relative homozygosity levels rather than the precise characterisation of ROH within each breed. In such cases, MAF filtering leads to uniformly shorter ROH across individuals, while preserving relative differences in length and distribution (Cesarani et al., 2021; Yang et al., 2017; Dzomba et al., 2021). Notwithstanding, the removal of the SNP may break a longer ROH into shorter ROH, which may then fall below the parameters used for detection (e.g., minimum length or minimum number of SNP), thereby potentially altering significantly the results by finding fewer regions (Ceballos et al., 2018; Meyermans et al., 2020). If your objective is to detect HRR instead, removing low-frequency SNP would be beneficial, as it will help with getting rid of noise (non-informative data) and will allow better focus on the signal of interest (i.e., heterozygosity).

After filtering, a small fraction of missing genotypes typically remains in the dataset. Or, you might need to combine data with different SNP densities (e.g., from low- and high-density SNP arrays). In such cases, it is very common to impute the missing genotype data, which is usually required for genetic analysis based on matrix algebra (e.g., GWAS, genomic predictions). For ROH/HRR, on the other hand,

imputing missing SNP data is not strictly necessary, as their detection can be carried out with or without missing data points. Therefore, you may choose whether or not to impute missing genotype data, although in some cases you are forced to go down the imputation route, e.g. when datasets with different densities are integrated. In any case, you may be reassured by knowing that the effect of imputation on the detected runs seems negligible (e.g., Biscarini et al., 2022, for HRR).

2.3. Criteria, parameter values and sampling strategy

The identification of ROH and HRR depends critically on the criteria applied for their detection, which strongly influence downstream biological interpretation. When discussing methods to detect ROH and HRR, we mentioned the several parameters that are involved and that can be tweaked to obtain results. The choice of specific values for these detection parameters will affect the results, especially in terms of the number and length of detected regions, giving vent to the important question of which parameter values to use. In general, all parameters are likely to affect the detection of ROH and HRR (Ferenčaković et al., 2013b; Biscarini et al., 2020; Meyermans et al., 2020; Chessari et al., 2024; Mulim et al., 2024). For example, in individuals from the same Soay sheep population, two studies reported different results: Nosrati et al. (2021) detected on average 50.38 ROH per animal, whereas Selli et al. (2021) reported an average of 188.4, which is roughly four times as many. As discussed by Selli et al. (2021), this divergence could be attributed to differences in ROH detection parameters, including the minimum number of SNPs per ROH (40 vs. 20), the maximal gap allowed between adjacent SNPs (1 Mb vs. 250 kb), and the lower SNP density (100 kb/SNP vs 70 kb/SNP). The authors showed that using less stringent parameter settings, in combination with a low-density SNP chip, can fragment ROH in regions with sparse marker coverage, resulting in an overestimation of the total number of runs and an underestimation of the proportion of long runs. Minimum SNP density (kb/SNP) can strongly affect the results of ROH analysis: in Maremmana cattle, Biscarini et al. (2020) found that by increasing minimum SNP density from 50 to 100 SNP/kbps, the number of ROH detected and their length, as well as the average F_{ROH}, increased.

For HRR detection, Selli et al. (2021) observed that changing the minimum number of SNP and window size from 10 to 5 did not increase the number of HRR detected. Biscarini et al. (2020) reported that allowing only one homozygous SNP reduced the number of detected HRR and their average size in cattle, compared with allowing more homozygous SNP (up to five). Similar results were found in goats, while more -but shorter- HRR were identified in sheep when allowing for a larger number of homozygous SNP (Biscarini et al., 2023). Using high-density SNP arrays, Ferenčaković et al. (2016) investigated HRR in cattle and identified 11 regions when up to four homozygous SNPs were permitted within a region. In contrast, analyses performed on Simmental cattle with high-density arrays did not detect any HRR segments when a stricter threshold of three homozygous SNPs was applied (Falchi et al., 2024). Selli et al. (2021) tested different numbers of homozygous (1 to 3) and missing (1 or 2) SNP within an HRR and observed a similar effect: when reducing the number of homozygous allowed from three to two, the number of HRR detected was reduced and the length increased, while reducing it further to one caused both metrics to decrease. Fabbri et al. (2025), in a study on pig breeds, observed that modifying the number of allowed homozygous and missing SNP influenced the total number of HRR detected but did not alter the trend among breeds. Minimum number of SNP, minimum length and maximum number of homozygous SNP appear to be the factors that mainly affect the detection of HRR (Mulim et al., 2024). From Chessari et al. (2024), the minimum number of SNP per segment and the maximum number of SNP with opposite genotype were the parameters that most influenced the number of HRR from the sliding window method; for the consecutive runs, the number of opposite genotypes was the most relevant. Selli et al. (2021) reported that a reduction in the minimum number of SNP in a run and window, as well as the number of homozygous and missing SNP allowed, caused a reduction of detected HRR, whereas reducing the minimum length of the run increased the number of HRR.

In literature, several studies used mathematical approaches to determine the minimum number of markers to call a ROH (e.g., Purfield et al., 2012; Ghoreishifar et al., 2020; Meyermans et al. 2020; Falchi et al., 2024). The optimal balance between stringency and detection sensitivity has been mathematically described by Purfield et al. (2012), who proposed parameter settings to minimize false positives while ensuring the detection of true autozygous segments. The equation, originally proposed by Lencz et al. (2007) considers both the number of SNP and individuals in the analyses, the percentage of false positive ROH allowed (that is usually set to 0.05), and the average heterozygosity across all markers.

Tweaking the parameters used for detection can mask or counteract the effects of low SNP array density discussed above. For instance, the excess of long ROH can be minimised by not allowing any heterozygous or missing SNP in a run, or by decreasing the gap between adjacent SNP, while the underestimation of short ROH can be alleviated by changing the requirements in terms of minimum length and minimum number of SNP in a ROH (Ferenčaković et al., 2013b). However, one must be aware of potential biases that may be introduced in the analysis when tweaking the parameter values. For example, allowing for heterozygous SNP in ROH can generate false positives (Hillestad et al., 2017). When allowing a small number of heterozygous calls, it is difficult to differentiate between situations where these heterozygous SNP are adjacent to each other, and therefore indicative of an actual break in the run or are scattered along the length of the ROH (Ferenčaković et al., 2013b). Testing different scenarios by tweaking parameter values is a good strategy to observe the consistency and accuracy of results, or at least to control potential bias (Biscarini et al., 2020; Selli et al., 2021).

From the above, it emerges that one of the main limitations in studies on ROH and HRR is the lack of consensus on standardized criteria for their definition and detection, which complicates comparisons across studies and species (Peripolli et al., 2017; Biscarini et al., 2020). Therefore, careful parameter optimization is essential to ensure consistent and biologically meaningful detection of ROH and HRR across studies.

Finally, we briefly address two key aspects that should be carefully considered when designing experiments on ROH and HRR: sampling strategies and the effect of population structure. Sample size, relatedness among individuals and breed composition can introduce bias in the estimation of the length and frequency distributions of ROH (Peripolli et al., 2017). The required sample size depends on the study objective. When the goal is to estimate recent/current inbreeding (or heterozygosity), a few tens of individuals per population is enough; for example, F_{ROH} is largely based on long ROH, which are robust to small sample sizes. Conversely, if the objective is to infer demographic history, shorter ROH and HRR are required, hence relatively larger sample sizes will be needed (e.g., ~100 per population). Further yet, using ROH to detect associations with binary or continuous traits (e.g. diseases) calls for much larger sample sizes, in the order of the thousands (Ceballos et al. 2018). Once size is established, sampling can be: (i) random, if the population is homogeneous; (ii) stratified, if structure is present in the data (geography, time, breed, age); (iii) proportional, if there are large differences in population sizes. Besides the implications for the sampling strategy, mentioned above, a strong population structure will affect data preprocessing steps like filtering and pruning based on LD, potentially leading to biased ROH/HRR analyses (Meyermans et al. 2020). In humans, Naslavsky et al. (2022) observed that admixture and ancestry composition strongly affect ROH profiles in elderly Brazilians, underscoring the importance of including diverse populations in genomic research. Using simulations and data on wild animals (*Tasmanian devils*), Silva et al. (2024) provided critical methodological evidence that the detectability and interpretation of ROH are highly sensitive to analysis parameters (e.g., SNP density, minimum ROH length) and

population-specific demographic histories, reinforcing the need for parameter optimization tailored to each dataset.

3. Applications of ROH and HRR in farm animals

To quantify the amount of work on ROH and HRR, a literature search was conducted in the SCOPUS (www.scopus.com) and Web of Science (WOS, www.webofscience.com) databases (accessed on the 27th of October 2025). The following keywords were used: "runs of homozygosity", "runs of heterozygosity", "heterozygosity rich regions". Since ROH are considerably more studied, a second search step included additional keywords: "autozygosity", "inbreeding", "selection signatures", "demographic history", and "genome-wide association". Ten different species (cattle, sheep, goat, pig, horse, chicken, rabbit, deer, donkey, and dogs) were considered. When searching only for the primary keywords in SCOPUS, 1228, 14, and 16 documents were retrieved for "runs of homozygosity", "runs of heterozygosity", and "heterozygosity-rich regions", respectively. Similar numbers of documents were found in WOS: 1172 for "runs of homozygosity", 14 for "runs of heterozygosity", and 15 for "heterozygosity rich regions". Tables 2 and 3 report the number of documents found in the two databases based on the keyword combinations in the different species. Supplementary Table 1 reports the year, DOI (digital object identifier), and link for the documents retrieved from the SCOPUS and WOS repositories. It clearly appears that ROH are more widely investigated than HRR. Among ROH studies, the largest number of documents was found in cattle (264 in SCOPUS and 280 in WOS); about 77% of these documents were found when searching also for "inbreeding". For all considered species, the largest number of documents was found using the keyword "inbreeding". The lowest number of documents on ROH was found for rabbits (5) and donkeys (4–5 depending on the database). Documents on HRR were found only for cattle, pig, sheep, goat, horse, and dogs. Also, for HRR, the largest number of documents was found in cattle: 3 documents using "runs of heterozygosity" and 7 documents using "heterozygosity rich regions". In farm animals, the first study on "runs of homozygosity" was published by MacLeod et al. (2009) in cattle, whereas the first document on "runs of heterozygosity"/"heterozygosity rich regions" was published only in 2016 (Williams et al., 2016), again in cattle.

3.1. ROH and estimates of inbreeding and inbreeding depression

Traditionally, inbreeding (F) is estimated based on pedigree information (F_{PED}), but in most cases this is unavailable or inaccurate (e.g., Banos et al., 2001). It is not uncommon for pedigree data to contain errors. Rates of pedigree errors in different dairy cattle populations worldwide have been estimated to reach >20% (Banos et al., 2001). Visscher et al. (2002) estimated a 10% overall pedigree error rate in United Kingdom dairy populations. Moreover, the probabilistic approach of pedigree analysis does not take into account the stochastic nature of recombination (McQuillan et al., 2008). The availability of genomic information has facilitated the quantification of inbreeding in livestock species and has led to significant advances in the estimation of inbreeding coefficients from ROH (F_{ROH}). F_{ROH} captures the total inbreeding coefficient of the individual, irrespective of pedigree accuracy or depth (or absence), within the resolution of the data set available (and hence the size of ROH that can be called) (Ceballos et al., 2018). The possibility of estimating the level of consanguinity in livestock without using pedigree records is a notable advantage, especially in breeding systems in which genealogies are absent or unreliable, as is the case of local breeds (Cortes-Hernández et al., 2021). Moreover, it allows us to distinguish between recent and ancient inbreeding (McQuillan et al., 2008). The "age of inbreeding" refers to the distance to the common ancestor that shared a specific haplotype and is approximately correlated with ROH length (Howrigan et al., 2011). Under the assumption that 1 cM equals 1 Mb, estimated F_{ROH} is expected to

Table 2
Number of documents on Runs of homozygosity retrieved in SCOPUS and WOS databases according to the used keywords.

	SCOPUS						WOS					
Species	Total	Autozygosity	Inbreeding	Selection signatures	Demographic history	Genome wide association	Total	Autozygosity	Inbreeding	Selection signatures	Demographic history	Genome wide association
Cattle	264	57	204	50	3	40	280	82	213	59	5	28
Pig	118	13	93	22	5	22	93	14	70	24	4	9
Sheep	121	14	88	30	4	28	126	20	88	31	4	15
Goat	74	5	56	24	7	15	70	9	52	24	8	4
Horse	59	9	44	16	0	11	47	10	34	12	0	4
Chicken	57	4	38	18	2	5	44	7	28	10	1	2
Dog	47	1	30	6	1	10	30	3	20	3	0	2
Donkey	5	1	1	1	0	1	4	1	1	1	1	0
Deer	9	1	7	0	2	0	7	1	5	0	2	0
Rabbit	5	0	5	0	1	0	5	0	5	0	1	0
Total	759	105	566	167	25	132	706	147	516	164	26	64

Table 3
Number of documents on Runs of heterozygosity and Heterozygosity rich regions retrieved in SCOPUS and WOS databases.

Species	SCOPUS		WOS	
	Runs of heterozygosity	Heterozygosity rich regions	Runs of heterozygosity	Heterozygosity rich regions
Cattle	3	7	3	6
Pig	5	4	4	3
Sheep	3	1	4	1
Goat	3	1	3	1
Horse	1	2	1	2
Chicken	0	0	0	0
Dog	0	1	0	1
Donkey	0	0	0	0
Deer	0	0	0	0
Rabbit	0	0	0	0
Total	15	16	15	14

correspond to the reference ancestral population dating 50 (F_{ROH} 1–2 Mb), 20 (F_{ROH} 2–4 Mb), 12.5 (F_{ROH} 4–8 Mb), 6 (F_{ROH} 8–16 Mb), and 3 (F_{ROH} > 16 Mb) generations ago. Because recombination events interrupt long chromosome segments, long ROH (~10 Mb) indicate recent inbreeding (up to five generations ago), while shorter ROH (~1 Mb) are the results of more distant ancestral effects (up to 50 generations ago), such as breed founder effects (Howrigan et al., 2011). Therefore, estimating *F* from ROH is particularly appealing as the number of generations of inbreeding and the history of recent selection events can be inferred from the extent and frequency of ROH regions.

Over the last 15 years, starting from the first report on autozygosity in cattle (Sölkner et al., 2010), the number of scientific works that have derived the F_{ROH} index in livestock has grown exponentially, increasingly refining the correlation between the molecular data and the actual genealogical consanguinity through the use of genome-wide SNP markers at various densities (e.g., Gurgul et al., 2016; Peripolli et al., 2018; Dadousis et al., 2022; Falchi et al., 2024) and whole genome sequences (e.g., Alemu et al., 2021; Mulim et al., 2022). In animal breeding, the correlation between pedigree- and genomic- inbreeding coefficients, particularly estimated from ROH (F_{ROH}), has received significant attention. Purfield et al. (2012) reported a strong correlation ($r = 0.75$, $P < 0.0001$) between the pedigree-based inbreeding coefficient and a statistic based on the sum of ROH longer than 0.5 kb. The authors concluded that, in the absence of pedigree information, the proportion of the genome covered by ROH can be used to infer aspects of recent population history, even with relatively small sample sizes. Similarly high correlations (up to 0.86) have been reported in recent studies (Nishio et al., 2023). However, literature reports correlations between F_{PED} and F_{ROH} ranging from low to high (Curik et al., 2014; Peripolli et al., 2017). Such variability can be attributed to differences in population structure, the parameters used to define ROH, the accuracy and depth of the pedigrees or the effect of ROH length (Peripolli et al., 2017). Indeed, ROH capture both ancient and recent autozygosity, reflected by shorter and longer IBD segments, respectively. In contrast, F_{PED} estimates inbreeding solely from recorded pedigree information, which often extends back only to a limited number of generations. Zavarez et al. (2015) observed that incomplete pedigrees fail to capture remote inbreeding and that F_{PED} estimates are only comparable with F_{ROH} calculated using long ROH segments. Consequently, the correlation between F_{PED} and F_{ROH} is influenced by both the depth of the pedigree and the size threshold of ROH used to capture ancestral inbreeding. The increasing correlation between F_{PED} and F_{ROH} with ROH length may be explained by considering that ROH reflect both past and recent relatedness and that F_{PED} estimates are based on pedigree records, which may not extend back many generations (Ferenčaković et al., 2013a; Marras et al., 2015). When only longer ROH reflecting recent relatedness are considered, the F_{PED}-F_{ROH} correlation tends to be higher. Several authors have shown that deeper pedigrees increase this correlation (e.g., Purfield et al., 2012; Ferenčaković et al., 2013a; Marras et al., 2015). Reversing the perspective, a high correlation between F_{PED} and F_{ROH} suggests that most inbreeding is recent and can be attributed to the relatively complete pedigree (Forutan et al., 2018). As an example, in Italian goat populations, a moderate correlation (0.30) between F_{PED} and F_{ROH} has been reported: since pedigree depth is related to the correlation between F_{ROH} and F_{PED}, to accurately estimate F_{PED} between four and six generations of known ancestors are needed (Cortellari et al., 2022).

This result is important for breeders, who should target for genotyping animals with few known ancestors. This relationship underscores the importance of integrating genomic data into breeding programs, as it allows for a more accurate evaluation of genetic diversity and inbreeding levels, ultimately aiding in the selection of healthier and more productive livestock.

Increased inbreeding can negatively affect the fitness of populations by reducing genetic variability, leading to allele fixation, long-term decrease in genetic variance, and slower response to selection (Howard et al., 2017). Moreover, incremented levels of inbreeding may reduce the mean phenotypic performance of livestock populations, a phenomenon known as inbreeding depression (Leroy, 2014). Inbreeding depression is primarily attributed to homozygosity for deleterious recessive mutations, which typically occur at low frequencies in a population. ROH could be enriched for these deleterious recessive alleles and can, therefore, be linked to inbreeding depression (Szpiech et al., 2013). Different studies showed that genomic inbreeding can be used instead of pedigree estimates to infer the impact of inbreeding depression on dairy (e.g. Pryce et al., 2014; Luigi-Serra et al., 2022; Cesarani et al., 2023), reproductive (e.g. Saura et al., 2015; Martikainen et al., 2020) and growth (e.g. Silió et al., 2013; Cortes et al., 2024) traits. All these studies confirmed that the use of inbreeding coefficient estimates from ROH, rather than pedigree estimates, can be used to monitor inbreeding, manage intra-breed diversity, and calculate the effects of inbreeding on phenotypic traits.

3.2. ROH islands

Selection signatures are genomic regions containing functionally important variants that have been, or continue to be, subjected to natural or artificial selection, leaving characteristic patterns in the DNA sequence (Qanbari and Simianer, 2014). ROH reflect such selection events and offer insights into both historical and recent demographic processes that have shaped a population (Purfield et al., 2012; Kim et al., 2013). Regions that are highly frequent and shared across individuals, first described as ROH islands by Nothnagel et al. (2010), represent potential targets of selection. ROH islands are not randomly distributed across the genome and are shared among individuals within a breed (Zhang et al., 2015; Gorssen et al., 2021). As such, they represent potential targets of positive selection and provide important insights into the demographic and selective history of the population under study. Accordingly, in livestock species, ROH-based analyses can contribute to the identification of genomic regions underlying phenotypic differences among breeds, particularly for traits of economic relevance (Mastrangelo et al., 2017). Genomic regions under selection identified using haplotype-based approaches (e.g., XP-EHH, Rsb, iHS, CLR) frequently overlap with ROH islands, both within and across breeds. For instance, Kim et al. (2013) reported that approximately two-thirds of the selection signatures detected in a German Holstein population overlapped with regions of high ROH frequency in U.S. Holsteins. Similarly, Purfield et al. (2017), analysing six commercial meat sheep breeds, found that 9 out of 11 selection signatures identified using global F_{ST} contained SNPs located within ROH. Further support comes from a WGS study on layer breeding population (Li et al., 2024), in which 12 of 23 detected ROH islands coincided with candidate regions identified using the de-correlated composite of multiple signals (DCMS) approach. Similar patterns have also been reported in 13 different chicken breeds

(Tan et al., 2024). Collectively, these findings suggest that recurrent ROH patterns are not solely shaped by demographic processes but can also harbour targets of positive selection. In this context, ROH-based analyses provide a complementary and robust framework for detecting selection signatures, particularly those resulting from recent artificial selection or local adaptation. The distribution of ROH across the autosomes may therefore help to validate candidate regions identified by other approaches and reduce the number of false positives from single-method analyses. Because ROH islands can represent signatures of selection, their overlap across populations and species provides a valuable framework for comparative genomic studies and may highlight functionally important genomic regions. A publicly available repository of ROH islands compiled by Gorssen et al. (2021) offers a valuable resource for comparative studies, providing access to ROH-based selection signatures across 442 populations from eight livestock and companion animal species, and can serve as a reference for validating newly identified ROH regions.

The genes found in these regions are generally related to several functions of productive and adaptive interest, such as immunity, growth, and production (e.g., meat, milk, egg, and wool) (Peripolli et al., 2018; Mulim et al., 2022; Mastrangelo et al., 2023). For example, previously known ROH islands were observed around the myostatin (*MSTN*) gene (Fariello et al., 2013; Purfield et al., 2017; Gorssen et al., 2021). *MSTN* is known to regulate muscle growth and development (Aiello et al., 2018), and its recurrent presence in ROH suggests that this locus has been under strong selection, likely associated with economically important traits such as increased muscle mass and meat production. In several livestock species, ROH islands have also been reported to harbor other candidate genes related to body weight and milk production, including *LAP3, NCAPG, LCORL*, and*ABCG2* (e.g., Cesarani et al., 2018; Mastrangelo et al., 2018; Xu et al., 2018; Grilz-Seger et al., 2019; Luigi-Sierra et al., 2019; Ablondi et al., 2020; Selli et al., 2021; Signer-Hasler et al., 2023; Becker et al., 2024; Senczuk et al., 2024). Several studies also identified the *KIT* gene, a well-characterized locus involved in coat colour determination, as overlapping with ROH islands in goats (Bertolini et al., 2018; Sun et al., 2022), pigs (Gorssen et al., 2020), and cattle (Cesarani et al., 2018). The recurrent presence of *KIT* within ROH islands suggests that this gene has been under selection for visual and breed-specific traits.

Although ROH contain valuable information for identifying signatures of selection, Nandolo et al. (2018) reported that some of the ROH islands found in the bovine genome may be artefacts due to copy number variants (CNV) and/or coverage gaps, and that CNV and coverage gaps need to be considered with great care when assessing signatures of selection via ROH patterns.

It is important to emphasize that the presence of genes within ROH does not imply a direct causal relationship with the inferred biological processes. In particular, ROH often occur in genomic regions characterized by reduced recombination and extended linkage disequilibrium (LD), where multiple genes may be encompassed within long homozygous segments, and the observed signal may reflect demographic history or hitchhiking effects rather than selection acting on the annotated genes themselves (Ceballos et al., 2018).

3.3. ROH analysis for production and functional traits

A specialised application of ROH analysis in livestock science is the association with productive and functional traits, which allows researchers to assess the impact of autozygosity on animal performance. Several studies have used ROH to identify candidate genes or quantitative trait loci (QTL) associated with important traits. For example, Modiba et al. (2022) analyzed the ROH pattern in beef cattle and identified 13 regions associated with semen traits, including known genes such as *CDF9* and *MARCH1*. A study on pigs identified several SNPs associated with the total number born and the number born alive (Mekonnen et al., 2024). ROH-based genomic scans have also been proposed as a complementary approach to traditional GWAS for exploring the genetic architecture of complex traits (Biscarini et al., 2015). In some studies, the presence or absence of specific ROH segments has been used as a tool to identify candidate genes via GWAS. These investigations applied formal statistical tests to associate ROH islands with economically important traits and reported *p*-values to assess significance. For example, Kim et al. (2015) explored potential links between ROH patterns and fertility and health-related traits in Jersey cattle, suggesting negative associations with daughter pregnancy rate and somatic cell score. In European Simmental cattle, ROH segments were significantly associated with milk yield, fat, and protein contents, with several regions influencing multiple traits simultaneously (Cesarani et al., 2021). Similarly, in sheep, ROH hotspots were linked to litter size, highlighting the potential of ROH analysis to reveal breed-specific genetic determinants of fertility (Tao et al., 2021). In American mink, 13 ROH regions were identified as significantly affecting growth and feed efficiency traits, four of which influenced multiple traits simultaneously (Davoudi et al., 2024). Across studies, a few consistent patterns emerge. ROH often overlap with known QTL or candidate genes for production and functional traits. Some ROH affect multiple traits, suggesting pleiotropy, and are particularly useful for detecting rare or population-specific variants that standard GWAS may miss. Overall, these findings highlight that ROH analysis complements existing genomic tools and can capture different aspects of the genetic architecture of complex traits.

3.4. HRR: balancing selection and diversity index

HRR can be analysed to search for signals of balancing selection, the evolutionary mechanism by which a multiplicity of alleles is maintained at specific *loci*, thereby preserving genetic diversity within populations (Charlesworth, 2013). In livestock subjected to high anthropogenic selective pressure, this process is crucial for maintaining traits related to disease resistance, reproductive fitness and adaptability to highly diversified environments (Hammer et al., 2020).

The mechanisms underpinning balancing selection are not yet fully understood, but they are certainly dynamically associated with the genomic structure of populations (Charlesworth, 2006; Derks and Steensma, 2021). A widely documented example in livestock is overdominance, which determines the conservation of both alleles in a population due to the improved performance, although not necessarily the fitness, of heterozygotes over homozygotes (Hedrick, 2015). In the scenario of frequency-dependent selection processes, rare alleles (which determine a rare phenotype) can confer a selective advantage, allowing them to persist within the population's gene pool. This evolutionary mechanism is reported for the variability of the immune response, where rare alleles can provide an advantage against pathogens that have adapted to more common alleles (Christie and McNickle, 2023). Furthermore, environmental factors can modulate the selective pressures that act on a population, leading to the conservation of multiple alleles over time and expanding their adaptive capabilities. Alleles that confer an advantage in one environment may be disadvantageous in others. For example, some coat colours in livestock may be favoured in specific climates or landscapes, leading to the maintenance of the associated genetic variability (Bizarria dos Santos et al., 2021). The Major Histocompatibility Complex (MHC) plays a critical role in the immune response, and the high levels of polymorphism in MHC genes, maintained through balancing selection (Jeffery and Bangham, 2000), enhance the ability of populations to respond to a diverse array of pathogens (Behl et al., 2012). In some cases, alleles that negatively affect fertility can be maintained in the population due to the heterozygote advantage, such as those that increase litter size or reproductive efficiency in heterozygous individuals but lead to reduced fitness or viability in homozygous individuals (Sironen et al., 2012). De Kort et al. (2022) highlighted high levels of heterozygosity downstream of transposable elements (TEs) in *Arabidopsis lyrata* inbred populations,

probably associated with genomic housekeeping functions, thus admitting the possibility of species evolution even with high inbreeding rates. Chessari et al. (2024) reported the case of the Montecristo goat that, despite being a highly inbred population due to its history of isolation and reduced number of mating animals, shows the highest number of HRR hotspots within the analyzed meta-population. Balancing selection has been observed in cattle (Kadri et al., 2014) and swine with the involvement of TEs (Sironen et al., 2012; Derks et al., 2018).

While F_{ROH} is well established in literature, a similar coefficient for heterozygosity based on HRR (a.k.a. HER, ROHet) is not always reported in studies on livestock. Mulim et al. (2024) highlighted Tajima's D as a useful test to detect deviations from neutrality, including potential signatures of balancing selection (Tajima, 1989). Recent works on the distribution of HRR in dairy goat breeds (Chessari et al., 2024), dual-purpose Simmental cattle (Falchi et al., 2024), and in the Duroc pig breed (Ruan et al., 2022) used a diversity index similar to F_{ROH} (named D_{ROHet} or F_{ROHet}). Taken together, these results indicate that HRR-derived heterozygosity indices provide information orthogonal to F_{ROH} by capturing localized retention of allelic diversity, potentially reflecting signatures compatible with balancing selection and heterozygosity hotspots, thereby refining the characterization of genome-wide diversity structure beyond what homozygosity-based metrics alone can resolve.

3.5. HRR islands genes

Unlike ROH islands, HRR regions are less characterized in livestock. Before providing an overview of specific findings, it should be emphasised that HRR islands are putative signals of balancing selection rather than established functional targets. The functional interpretation of HRR should therefore account for the possibility that high heterozygosity may arise from structural and demographic processes rather than from direct selective pressure acting on the genes within these regions. In fact, several phenomena may contribute to the occurrence of HRR islands. They could be the result of chromosomal properties such as high recombination rates, inversions, the concentration of structural elements such as SINE and LINE, local balancing selection for gene alleles, local reduced inbreeding effects, or random events (Smaragdov, 2025).

Cattle are the most studied species, with Taurine, Indicine, and Composite breeds analyzed using SNP data (Ferencakovic et al., 2016; Williams et al., 2016; Biscarini et al., 2020; Lashmar et al., 2022; Mulim et al., 2022; Carrara et al., 2024; Falchi et al., 2024; Mulim et al., 2024; Szmatoła et al., 2024; Maxman et al., 2025). Identified HRR are associated with immune function, reproduction and fertility, disease resistance, climate adaptation, and longevity. Some regions are conserved across breeds, whereas others appear breed-specific, reflecting distinct demographic histories and selection regimes. For example, common BTA21 regions across multiple breeds (Biscarini et al., 2020; Falchi et al., 2024; Mulim et al., 2024) include genes linked to reproduction (*SERPINA6*), anaemia response (*PCSK6*), and temperament (*GABRB3*).

In swine, HRR studies are limited but highlight regions associated with immunity, reproduction, and litter traits, with some overlap across breeds (Sanglard et al., 2021; Chen et al., 2022; Bordonaro et al., 2023). Overlapping HRR on SSC14 across different breeds (Ruan et al., 2022; Bordonaro et al., 2023) includes *LIF* (ovulation, implantation) and *EWSR1* (meiotic regulation). Recently, Fabbri et al. (2025) identified SSC1, 2, 6, and 14 as HRR-rich, with genes on chromosome 6 particularly linked to immune response.

A considerable overlap in HRR hotspots has been observed across studies on caprine species, highlighting several genes associated with the survival and adaptation of breeds worldwide. The regions that, among the reviewed scientific studies, have shown almost perfect matching are in CHI1 (Li et al., 2022; Chessari et al., 2024; Tsartsianidou et al., 2025), CHI12 and CHI18 (Chessari et al., 2024; Pegolo et al., 2025; Tsartsianidou et al., 2025). In sheep, the relatively few available studies have identified HRR across multiple chromosomes; however, concordance among studies is generally limited, and overlap at specific loci is rare, with only OAR13 reported in more than one study, albeit involving distinct sub-regions (Tsartsianidou et al., 2021; Selli et al., 2021). Candidate genes within these regions have been linked to reproduction (*SPAG4*), immune response (*ROMO1*), and traits potentially related to domestication (*SUMF1*). Some HRR coincide with previously reported sweeps in arid-adapted breeds (Kim et al., 2016), while others encompass genes influencing sperm function, neurodevelopment, and metabolism. More recent high-density SNP studies in Mediterranean breeds (Carta et al., 2026) confirmed the presence of HRR islands composed of loci associated with reproduction and climate adaptation (e.g., *CAPSPERB, TC2N*, and *VPS13B*), supporting their potential relevance to phenotypic diversity and local adaptation.

In horses, shared HRR regions across breeds (Bizzaria dos Santos et al., 2021; Santos et al., 2023; Moravčíková et al., 2024) suggest selection for performance, muscle repair and sensory functions.

These findings reflect the history of adaptation and selection and provide insights on heterozygosity patterns. However, despite the growing number of studies, the statistical reliability of HRR islands detection remains insufficiently explored. Many studies rely on empirical thresholds without proper cross-validation, and the biological significance of several identified regions is still speculative. Moreover, variability in SNP density array and filtering criteria complicates direct comparison across studies. Future research should therefore aim to integrate robust statistical frameworks and functional validation to distinguish true signals of heterozygosity enrichment from methodological artifacts.

Finally, several studies have reported that genomic regions exhibiting high heterozygosity in one breed can be strictly homozygous in another (Selli et al., 2021; Chessari et al., 2024; Pegolo et al., 2025). Such contrasting patterns across breeds, largely influenced by their demographic histories, breeding strategies, and adaptive trajectories (Bordonaro et al., 2023), reinforcing the idea that ROH and HRR islands are shaped by breed-specific selective pressures (Tsartsianidou et al., 2021). Interestingly, this contrast is not limited to comparisons among breeds. Emerging evidence indicates that ROH and HRR hotspots may also occur within the same genomic region in different individuals of the same breed (Carta et al., 2026). This intra-breed variability suggests that homozygosity and heterozygosity represent dynamic genomic states influenced by the interplay of selection, recombination, and demographic processes. In such regions, balancing selection may maintain alternative haplotypes in part of the population, while directional or purifying selection may promote fixation in others. Overall, these findings underscore the complexity of genome architecture, suggesting that homozygosity or heterozygosity islands may reflect the combined effects of multiple evolutionary processes rather than a single selective regime.

4. Concluding remarks and future perspectives

In this review, we provide an updated overview of the methods, studies, and applications of ROH and HRR in livestock species. Studies on ROH and HRR are rapidly expanding, with applications across a wide range of species, populations and research topics. ROH can be used to identify autozygosity, provide insights into the timing of inbreeding, and inform the design of breeding programs aimed at minimizing the risk of inbreeding depression. HRR may help identify genomic regions under balancing selection, where heterozygosity is maintained due to a fitness advantage (e.g., disease resistance or fertility).

Many indigenous breeds have unique ROH and HRR patterns that can guide programs for the preservation of adaptive traits in local populations threatened by cosmopolitan breeds. ROH and HRR analyses are promising tools to enhance sustainable breeding, genetic health, and adaptability in livestock. However, several limitations must be considered when interpreting results, including sample size, marker density and parameter choice. Different studies define ROH and HRR using heterogeneous criteria, and this variability can strongly affect

reproducibility and comparability across studies. ROH/HRR analyses are also prone to false positives, especially in populations with complex demographic histories. Because drift and inbreeding may lead to increased homozygosity (ROH) or reduced heterozygosity (and thus fewer HRRs), interpreting the patterns purely from the perspective of artificial selection (productive traits) can be misleading. Common mitigation strategies include applying stringent thresholds on segment length and SNP count, validating results across independent populations, and integrating genomic signals with functional annotation or QTL information. Replicating ROH/HRR findings in independent datasets provides stronger evidence that a region reflects selection rather than population-specific drift.

Comparative ROH/HRR analysis across livestock species (cattle, sheep, goats, pigs, poultry, rabbits, and horses) can highlight common regions of interest, thereby accelerating the functional annotation of livestock genomes. The increasing availability of large genomic datasets offer a valuable opportunity to refine the identification of ROH and HRR, and to deepen our understanding of the underlying biological mechanisms. Coupling ROH/HRR analyses with multi-omics data (e.g., transcriptomics, epigenomics, metabolomics) will enhance our understanding of the biological processes underlying complex traits and adaptive responses. The future application of ROH and HRR in livestock genetics is likely to shift toward more integrated, data-driven strategies aimed at enhancing genetic resilience, productivity, and sustainability. As computational methods and functional annotation improve, ROH and HRR will play an increasingly central role in precision breeding and the long-term sustainability of livestock populations.

Finally, with the advent of pangenome assemblies (e.g. Gong et al., 2023), future analyses of ROH and HRR are expected to gain both accuracy and resolution (Smaragdov, 2025). By moving beyond a single linear reference genome, pangenomic frameworks provide a more comprehensive representation of structural variation, alternative haplotypes, and population-specific genomic segments. This shift is expected to mitigate reference bias in read alignment and genotype calling, thereby enhancing the reliability of detecting autozygous and heterozygous regions across diverse populations.

Declaration of generative AI in scientific writing

We affirm that artificial intelligence (AI) tools were solely employed to enhance the readability and language of this article.

CRediT authorship contribution statement

F. Biscarini: Writing – review & editing, Writing – original draft. **A. Cesarani:** Writing – review & editing, Writing – original draft. **A. Criscione:** Writing – review & editing, Writing – original draft. **S. Mastrangelo:** Writing – review & editing, Writing – original draft, Supervision, Conceptualization.

Declaration of competing interest

The authors declare that they have no conflict of interest regarding the topics approached within this study.

Supplementary materials

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