



Background

- Mastitis is a complex polygenic disease that is regulated by several genes with small effects.
- We performed a systematic review, meta- and gene prioritization analysis of genome-wide association study (GWAS) studies to identify key genetic markers and genes associated with mastitis-related traits and somatic cell score (SCS) in dairy cattle.

Materials and Methods

- Systematic review question followed the PPO framework (**population: dairy cattle; prognostic tool: GWAS; outcome: mastitis and SCS**).
- Search was conducted using electronic databases, conference proceedings and industry meeting reports.
- Gene prioritization analysis: GUILDify and ToppGene.
- Meta-analysis: Han and Eskin's random effects model in METASOFT.

Preliminary results

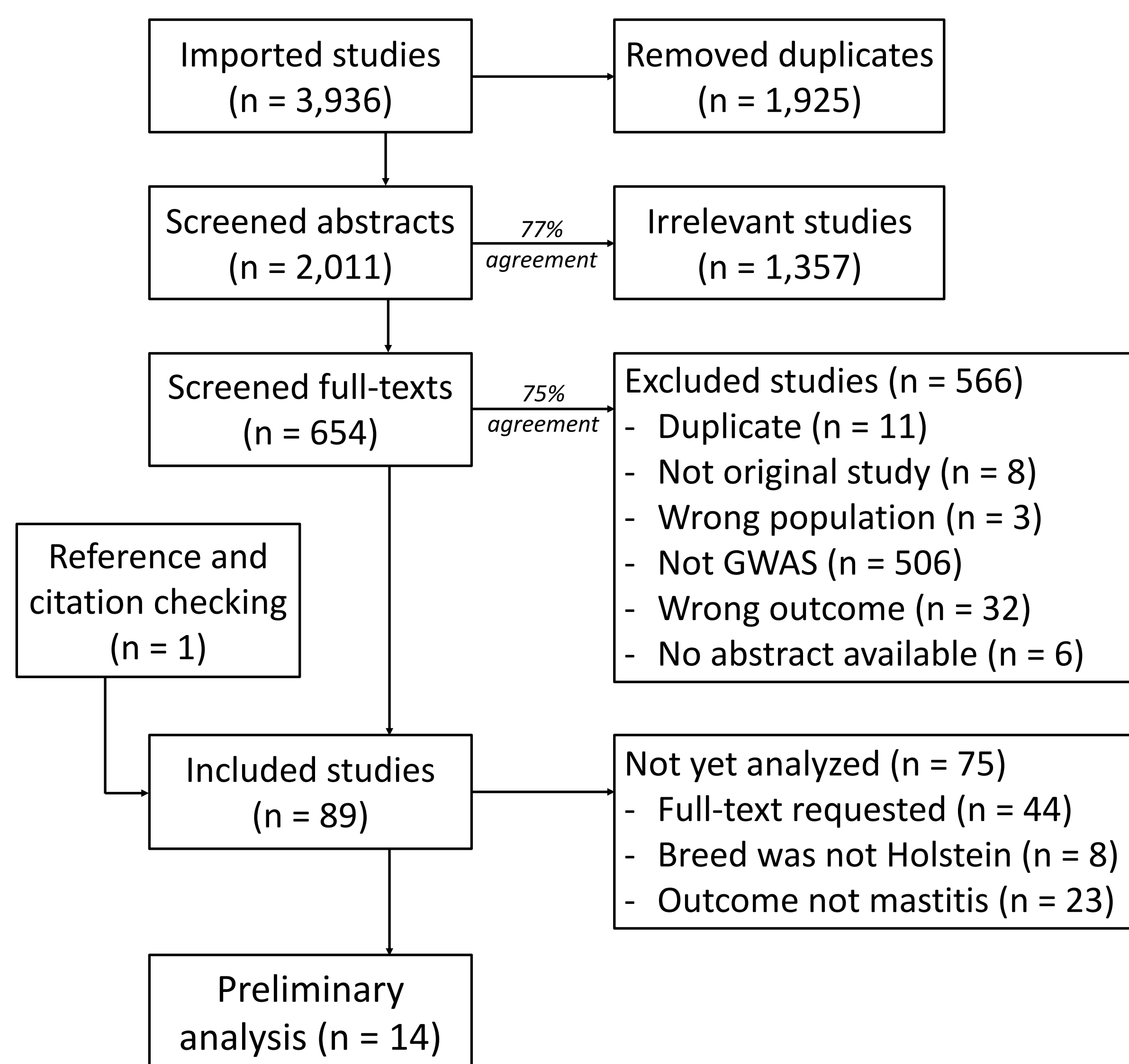


Figure 1: PRISMA flow diagram

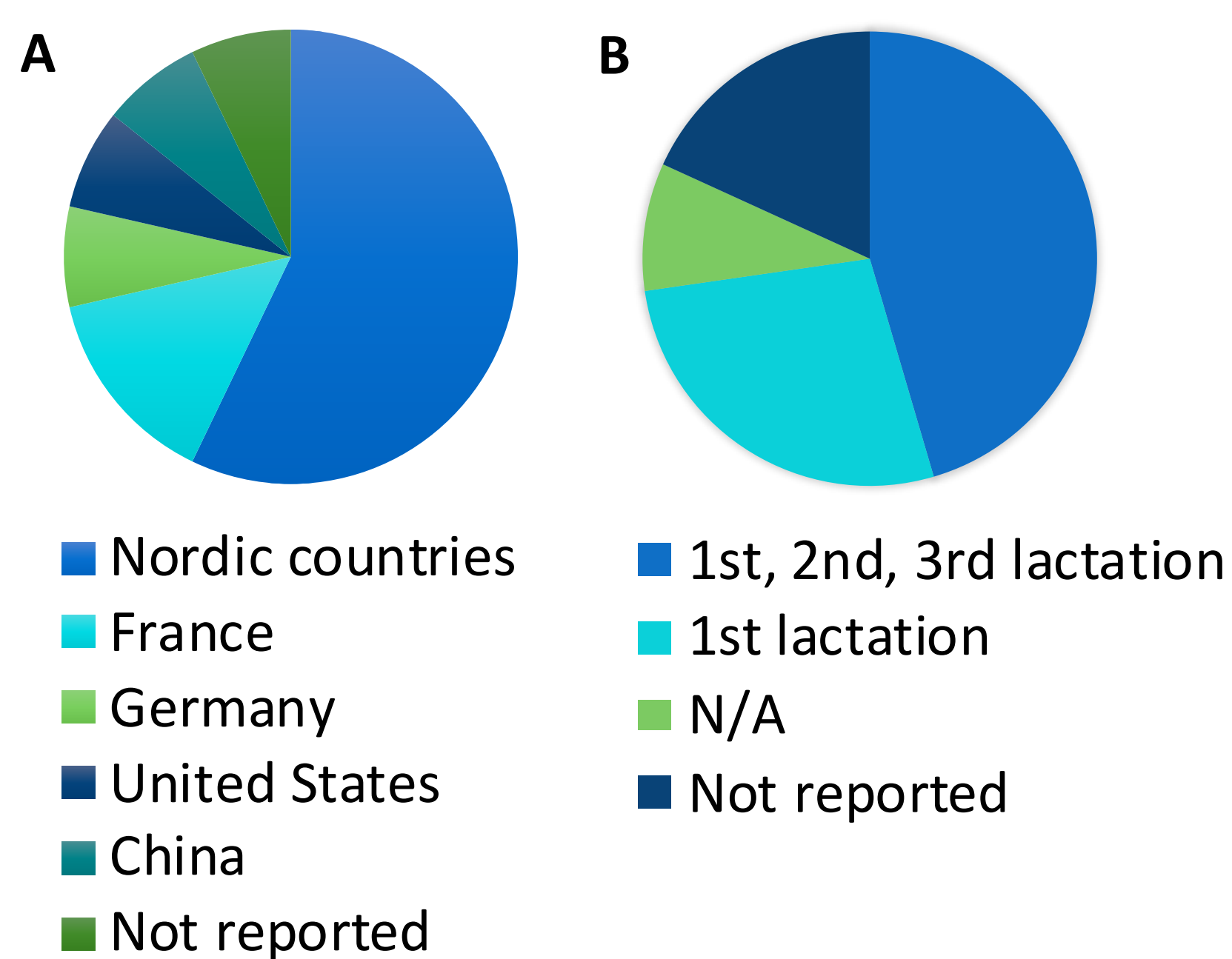


Figure 2: Origin of data (A) and lactation considered (B), 14 studies

Table 1: Characteristics of included 14 studies

Study	Number of genotyped animals	# SNPs used for the analysis	# variants associated with mastitis
Cai 2018	5,147 bulls	15,552,968	22 SNPs
Fang 2017a	5,056 cattle	15,355,382	5 GO
Fang 2017b	5,056 cattle	15,355,382	5 SNPs
Fang 2018	5,056 bulls	15,355,382	5 QTL
Kurz 2019	43 cows	585,949	116 SNPs, 27 QTLs
Ma 2019	3,114 bulls	15,388,916	405 markers
Marete 2018a	46,732 cattle	40,810	11 SNPs
Marete 2018b	32,491 cows	49,835	28 SNPs
Naderi 2018	6,744 cows	43,939	4 SNPs
Sahana 2013	5,035 bulls	648,219	23 SNPs
Sahana 2014	2,098 bulls	36,387	143 SNPs
Su 2014	5,643 bulls	44,919	
Tiezzi 2015	1,361 bulls	39,004	10 SNP windows
Yang 2019	40 cows	10,058	27 SNPs

- A clear definition of mastitis was absent in most of the studies.
- Cases in either the first, second or third lactation were often considered within studies.
- Quality assessment predominately focused on the handling of genomic data and the selection of SNPs for the GWAS.
- Various methods were used to conduct the GWAS.
- Reported outcomes varied across 14 studies.
- Identified genes were mostly associated with immune-related processes.

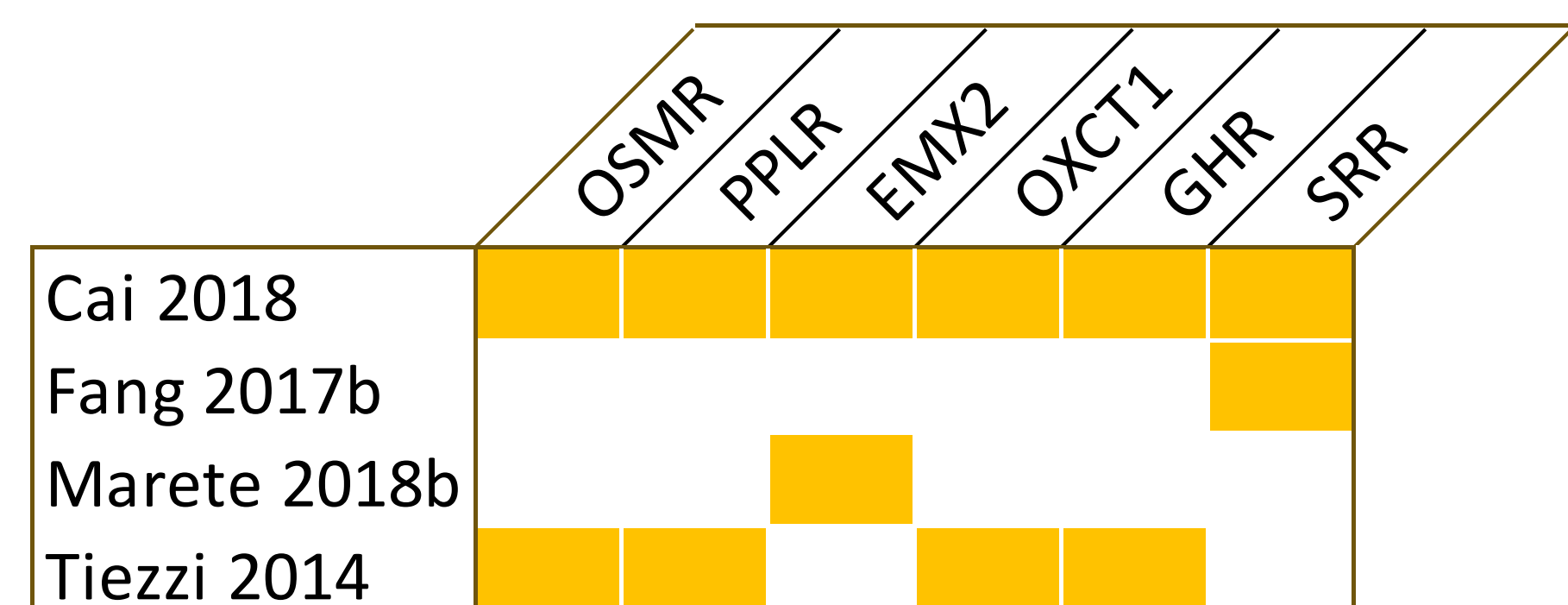


Figure 4: Commonly reported genes among the 14 pre-analysed studies

Conclusion

- 89 articles were identified.
- Among 14 studies immune-related 6 common genes were identified.

Significance

This systematic review will add value in summarizing GWAS findings to better understand the genetic architecture of mastitis in dairy cattle.