

## INTRODUCTION

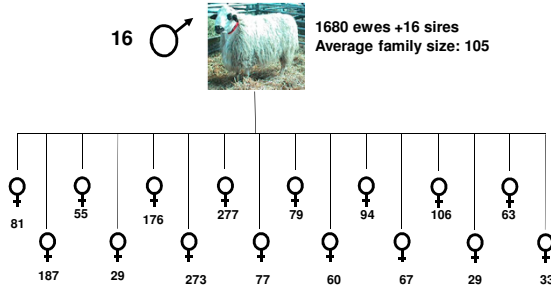
- **Subclinical mastitis:** a major problem for the dairy sheep industry.
- **Somatic cell score (SCS) for milk:** a good indicator of this complex disease.
- **QTL for SCS in Churra sheep:** A previous genome scan identified a single significant QTL influencing SCS on sheep chromosome (OAR) 20<sup>1</sup>.
- **Aim of current work:** performing a medium-high density mapping analysis to detect and fine-map QTL controlling SCS in Churra sheep by using the *Illumina OvineSNP50 BeadChip* to study.



## METHODS

### 1-Resource Population and Experimental Design

- **Daughter design:** 1680 ewes from 16 half-sib families and 29 flocks of the Selection Nucleus of the Spanish Churra Breeders' Association (ANCHE).



1680 ewes +16 sires  
Average family size: 105

### 3-Phenotypes

- Raw data of SCS (Official Milk Recording, ANCHE)
- Yield deviations (YD<sub>SCS</sub>) estimated using a multivariate animal repeatability model

$$YD_{ijklmn} = Y_{ijklm} - \mu - HTD_i - BO_j - Age_k(BO) - BL_l - WL_m - Ep_n$$

HTD: Herd-Test-Day  
BO: birth order  
Age<sub>k</sub>: age of the ewe at parturition (covariate within birth order)

BL<sub>l</sub>: number of born lambs  
WL<sub>m</sub>: number of weeks of milk production of the ewe and  
Ep<sub>n</sub>: ewe's permanent environmental effect

### 2-Genotypes

**2.1- Raw genotypes** for the OvineSNP50 BeadChip for the whole population

**2.2- Quality Control (QC)**

- **QC1 (per animal):** call rate > 90%
- **QC2 (per SNP):**
  - call rate > 95%
  - minor allele frequency (MAF) > 0.05
  - correspondence HWE, p-value > 0.00001

**2.3- Markers for analyses:**

43,784 autosomal SNPs (1,665 ewes +16 rams)

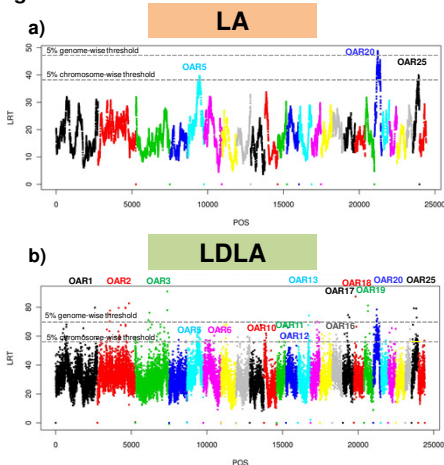
### 4- Genetic analyses

- Linkage map used based on the Ovine Assembly v2.0 (conversion ratio: 1 cM ~ 1 Mb)
- **Linkage (LA)** and **combined linkage and linkage disequilibrium (LDLA)** analyses performed with *QTLMap software*<sup>2</sup>
- **Positional candidate genes** extracted from the bovine orthologous region (UMD3.1) of the estimated QTL confidence intervals (CI) using *BioMart*<sup>3</sup>
- *BioGraph*<sup>4</sup> used to assess the possible relationship between the positional candidates and the target term "immune response"

## RESULTS

Nine genome-wise significant QTL were identified by LDLA, with the one located on OAR20 also identified by LA (Table 1, Figure 1),

**Figure 1**



**Table 1**

	Chr.	Peak position (Morgans)	Localization (Mb); Flanking interval	CI= 2*log(test) drop off (Mb)	Average effect (SD of trait) in significant families (number of significant families)
LA	OAR20 [910]	0.217	OAR20_22851804.1 s39376.1 259.65	21.2-23.8	0.233 [10]
	OAR1 [4987]	2.597	OAR1_280315444.1 OAR1_280355916.1 208.66	259.6-259.8	0.211 [6]
LDLA	OAR2 [4676]	2.087	OAR2_220142031.1 s12784.1 212.34	208.6-208.8	0.262 [4]
	OAR3 [4164]	2.123	OAR3_27933.1 s42120.1 71.73	212.2-212.4	0.184 [6]
	OAR13 [1402]	0.717	DU360920_246.1 s64654.1 - 33.95	71.6-71.8	0.283 [7]
	OAR1 [1178]	0.340	OAR17_36829676.1 s46426.1 12.58	33.8-34.0	0.249 [1]
	OAR18 [1192]	0.126	OAR18_1250969.1 OAR18_12576454.1 26.30	12.5-12.7	0.263 [3]
	OAR19 [1032]	0.263	OAR19_27611685.1 s14176.1 23.72	26.1-26.4	0.228 [7]
	OAR20 [910]	0.237	OAR20_24966073.1 s18014.1 15.38	22.6-23.8	0.245 [10]
	OAR25 [846]	0.154	OAR25_17043093_X1 s37560.1 15.3-15.5		0.245 [6]

- **29 positional candidate genes** extracted from the identified QTL regions
- The ranked list of 24 genes included in *BioGraph*<sup>4</sup> based on the "ranking score (RS)" related to the target term will be considered to select functional candidate genes to further study.
- The genes showing the higher RS have previous reports in relation to the immune response and are related to the QTL detected on
  - OAR3: Interleukin 17A receptor (*IL17RA*)
  - OAR20: - Beta-Defensin 12 (*DEFB12*)  
- Cysteine-rich secretory proteins 1, 2, 3 (*CRISP1*, *CRISP2* and *CRISP3*)

## CONCLUSIONS

- Results on OAR20: replication of the previously reported QTL on OAR20 in Churra sheep.<sup>1</sup>
- Promising functional candidate genes have been detected for the QTL detected on OAR3 and OAR20.
- Future research may undertake the possible relationship of these candidate genes with the SCS phenotype in Churra sheep