



Genome-wide association analysis reveals the common genetic locus for both the typical and atypical polycerate phenotype in Tibetan sheep

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Source description: Through a recent survey of Chinese native sheep breeds, the polycerate phenotype, an ancient and rare trait,¹ was observed in Tibetan sheep (Fig. 1). In this study, we sampled 72 polycerate vs. 24 two-horned Tibetan sheep in the Qinghai-Tibet plateau (5200 meters above sea level) and genotyped them with the Illumina OvineSNP50 BeadChip array to map the genomic regions of typical (four horns) and atypical polycerate (three, five and six horns) phenotype. We removed all full sibs from the samples.

Genome-wide association study (GWAS) analysis for typical and atypical polycerate phenotype: A total of 6258 SNPs and three individuals (1 three-horned and 2 two-horned) were removed due to failure to meet the following conditions in PLINK v1.90b3y²: a SNP call rate > 95%, minor allele frequency > 0.05 and call rate per animal > 0.9. Finally, 47 983 SNPs were left for the analysis. The genotypic dataset has been submitted to Animal Genome

Database (<https://www.animalgenome.org/repository/pub/CAAS2017.0809/>).

Three case-control GWAS analyses were performed to locate the genomic regions for the typical polycerate (four horns; $n = 39$), atypical polycerate (14 three-horned, 15 five-horned and 4 six-horned; $n = 32$) and all polycerate (more than two horns; $n = 71$) phenotypes, with two horned sheep ($n = 22$) used as a control group. The GWAS was performed through the use of EMMAX software.³ The kinship matrix was constructed using the EMMAX-KIN program.

Significant SNPs and gene annotation: All three GWAS analyses for typical, atypical (Fig. 2a) and all polycerate phenotypes revealed the same genome-wide significant SNP marker (rs399639314) at the position of 132.814 Mb on ovine chromosome 2 with $-\log_{10}(P) = 11.46$, 12.91 and 15.26 respectively. This SNP is 65 kb upstream of the *metaxin 2* (*MTX2*) gene and 16 kb downstream of the *HOXD* gene cluster according to the gene annotation in Oar_v4.0, the updated version of *Ovis aries* assembly (<http://www.ncbi.nlm.nih.gov/genome/?term=ovis±aries/>). The SNP was located within a merged genomic region (OAR2:131.990–133.525 Mb) from previous studies (Fig. 2b). This suggests that the same SNP affects both the typical and atypical polycerate phenotype.

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Figure 1 Examples of the polycerate trait. (a) Four-horned Tibetan ram with the forward upper horns. (b) Tibetan ram with four big horns. (c) Four-horned ewe with smaller horns.

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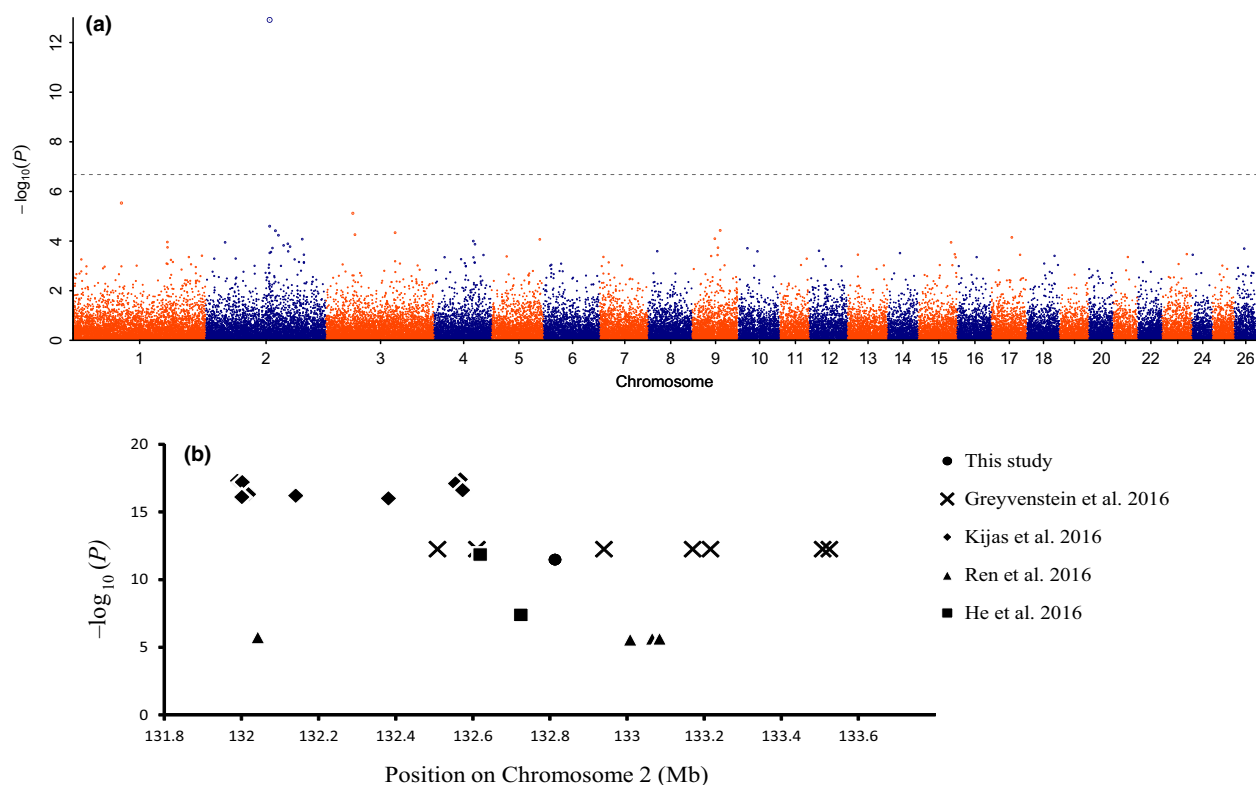


Figure 2 Genome-wide association study results and summary of all significant SNPs for the polycerate locus. (a) Manhattan plot of genome-wide association analysis for atypical polycerate phenotype (three, five and six horns). (b) Summary of all significant SNPs for the polycerate locus in previous papers and this study.

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References

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