# A large-scale genome-wide association study in U.S. Holstein cattle

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## Abstract

#### Background

Genome-wide association study (GWAS) is a powerful approach to identify genetic variants associated with phenotypes. However, only limited mutual confirmation from different studies was avaiable. The rapidly growth in single nucleotide polymorphism (SNP) data and phenotypic observations in U.S. Holstein cattle provides an opportunity of large-scale GWAS for the discovery of new effects and genetic mechanisms towards building consensus of genetic variants associated with dairy traits.

#### Results

The GWAS using 294,079 first lactation Holstein cows identified new additive and dominance effects on three yield traits, two percentage traits, three fertility traits and somatic cell score. This study revealed *DGAT1* had extreme antagonistic pleiotropy effects on milk production with the largest positive effect on fat yield and the largest negative effects on milk and protein yields, and these extreme positive and negative effects were responsible for *DGAT1*'s most significant effects on milk production. The *SLC4A4-GC-NPFFR2-ADAMTS3* region of Chr06 had multiple highly significant effects for milk production, fertility and somatic cell score. The *GHR-PRLR* region of Chr20 had significant effects on milk yield and the largest positive allelic effects on milk yield. A Chr05 region had significant effects on fat yields. *COX17* of Chr01, *GC* of Chr06 and *SIPA1L3* of Chr18 had the most significant effects on daughter pregnancy rate and cow conception rate, *AFF1* of Chr06 had the most significant dominance effect on heifer conception rate. These fertility effects were primarily due to their negative allelic effects. For somatic cell score, *GC* of Chr06 and *PRLR* of Chr20 had the most significant additive effects, and *CEP97* of Chr01 had the lowest somatic cell score.

#### Conclusions

The large-scale GWAS identified new additive and dominance effects associated with dairy traits and new genetic mechanism of SNP effects, and provided a large-sample prospective of dairy SNP effects. Results from this large-scale GWAS are a significant contribution towards building consensus on genetic variants associated with dairy phenotypes.

### Keywords

GWAS, dairy cattle, milk production, fertility, somatic cell score