

26. Farm-GTEx project and Allele-Specific Expression in Pig

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PigGTEx is part of the FarmGTEx project (Farm animal Genotype-Tissue Expression, <https://www.farmgtex.org/>). FarmGTEx was established to develop a comprehensive public resource of genetic regulatory variants in domestic animal species, which is essential for linking genetic polymorphisms to variations in phenotypes, helping fundamental biology discovery and exploitation in animal breeding and human biomedicine. The aim of this project is to study tissue-specific regulation of gene expression and other molecular phenotypes, in relation to genotype in distinct biological contexts (e.g., breeds, cell type, and environmental exposures), and to elucidate the underlying molecular mechanisms of action using multi-omics data.

Pervasive allelic variation at the gene and single nucleotide (SNV) levels between individuals is commonly associated with complex traits in livestock. As a complementary analysis of expression quantitative trait loci (eQTL) mapping, allele-specific expression (ASE) analyses can provide a detailed annotation of allelic imbalance and infer the existence of cis-acting transcriptional regulation. We developed an unbiased standardized computational pipeline for ASE analysis by using gene expression data from the PigGTEx project. This was used to generate a vast allelic imbalance resource, consisting of 6,864 samples spanning 23 pig breeds and 42 pig tissues. We evaluated tissue- and breed-sharing of the identified ASE and compared them with eQTL. We find a large amount of considerable variations of ASE across tissues and breeds, suggesting complex genomic regulation of allelic expression. These tissue-specific ASE profiles may underline economically important traits and could be utilized as weighted SNVs. The identified ASE profiles are a valuable foundation for identifying the molecular regulatory codes that are driving complex traits in livestock. In future analyses, we will continue to explore the association between ASE and phenotypic diversity, and how it works.