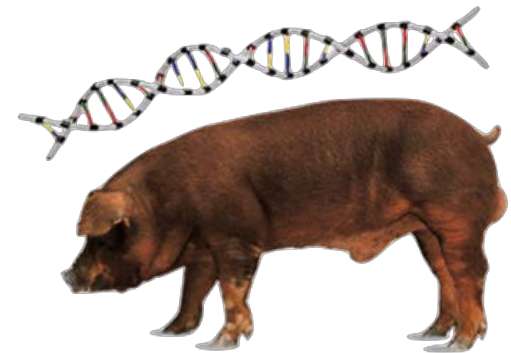


Skeletal Muscle eQTL and Allele-Specific Expression Associated with Phenotypic Traits in pigs

Catherine W. Ernst

Department of Animal Science
Michigan State University
East Lansing, MI



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Graduate Students & Co-PIs



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Kaitlyn Daza



Juan Steibel



Ron Bates

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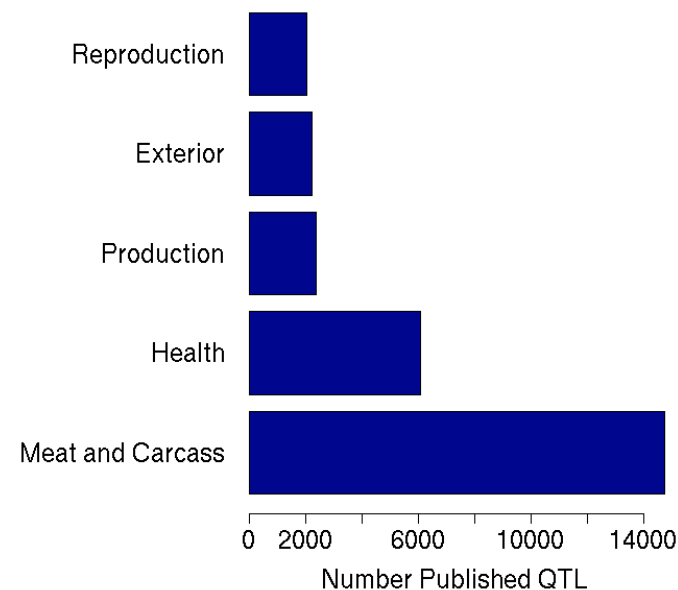
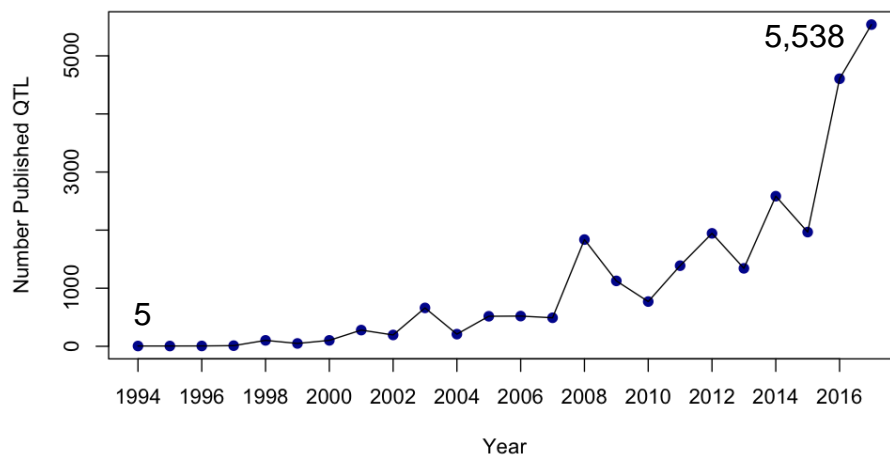
Animal Molecular Genetics Lab

- Dr. Cathy Ernst
- Nancy Raney
- Dr. Deborah Velez-Irizarry
- Kaitlyn Daza
- Ryan Corbett
- Scott Funkhouser

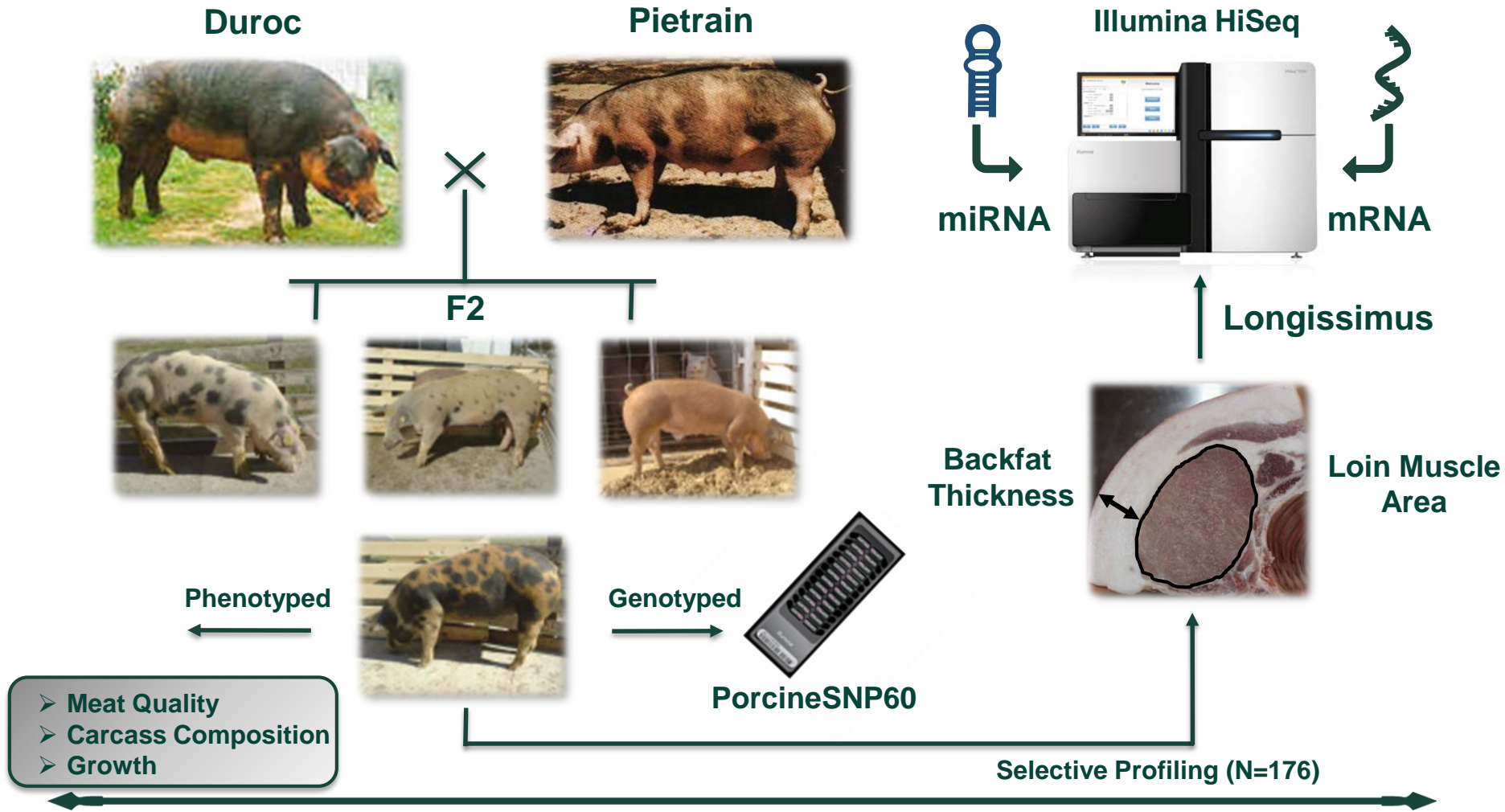


Introduction

- Numerous QTL identified for pork production traits
 - Functional relevance unknown
- Functional genomics
 - Genetic architecture of a trait

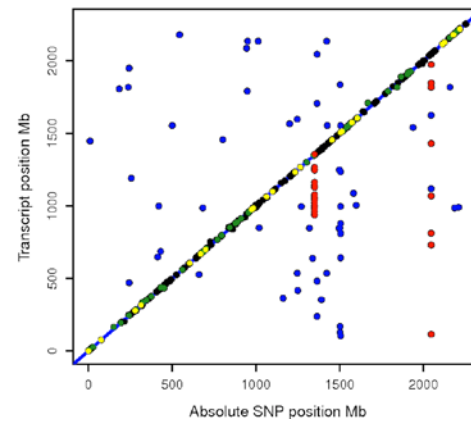
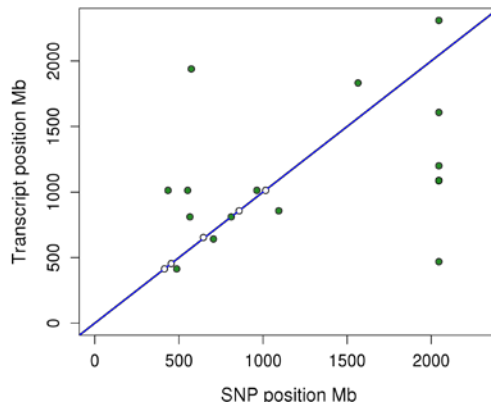


Methods Overview

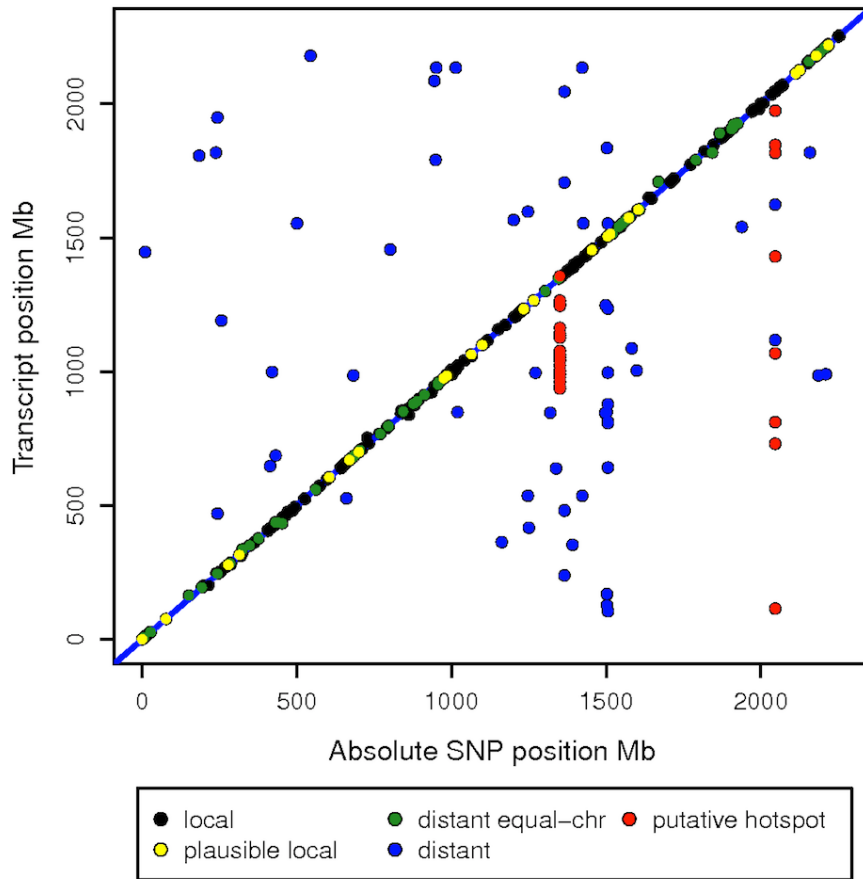




Expression QTL Analysis



mRNA eQTL Results



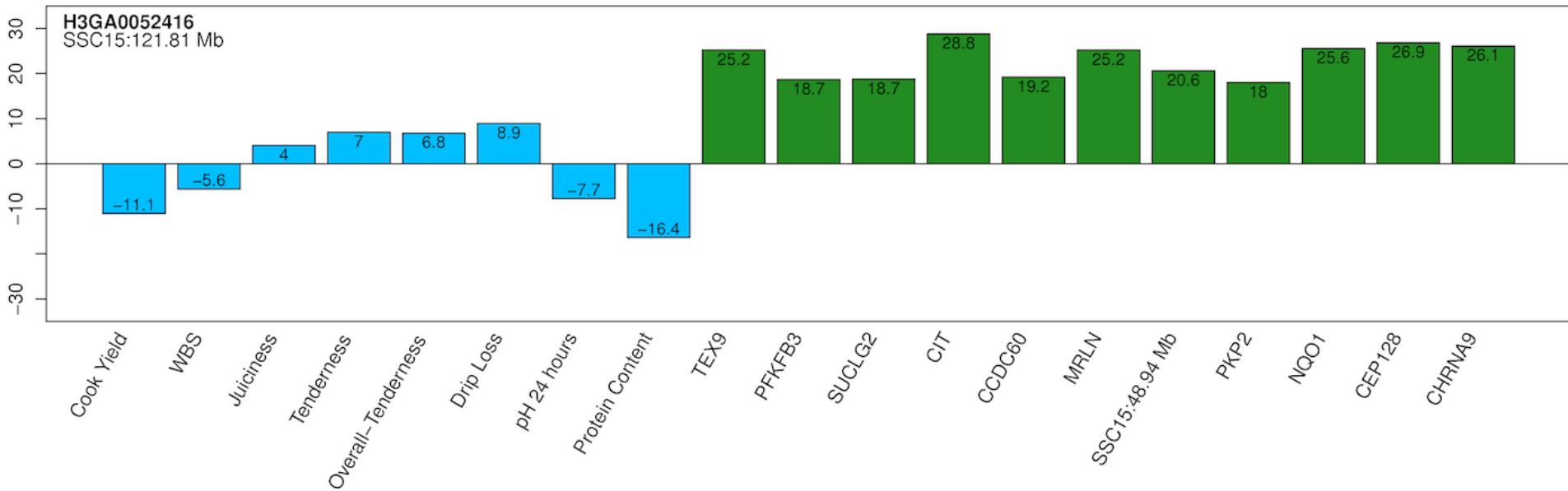
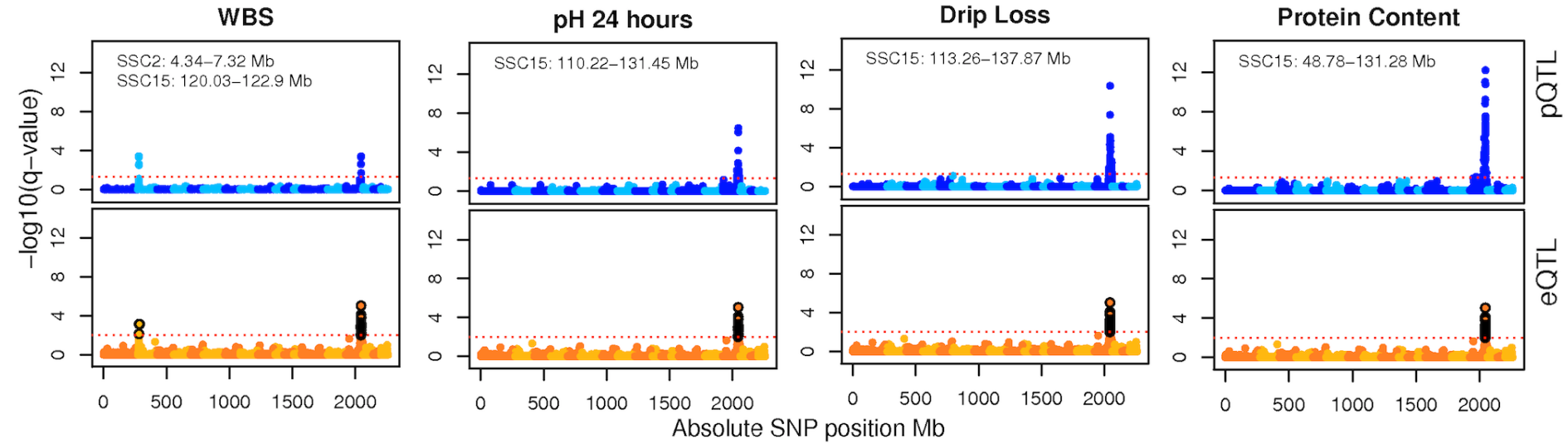
GBLUP-based GWA

eQTL Classification

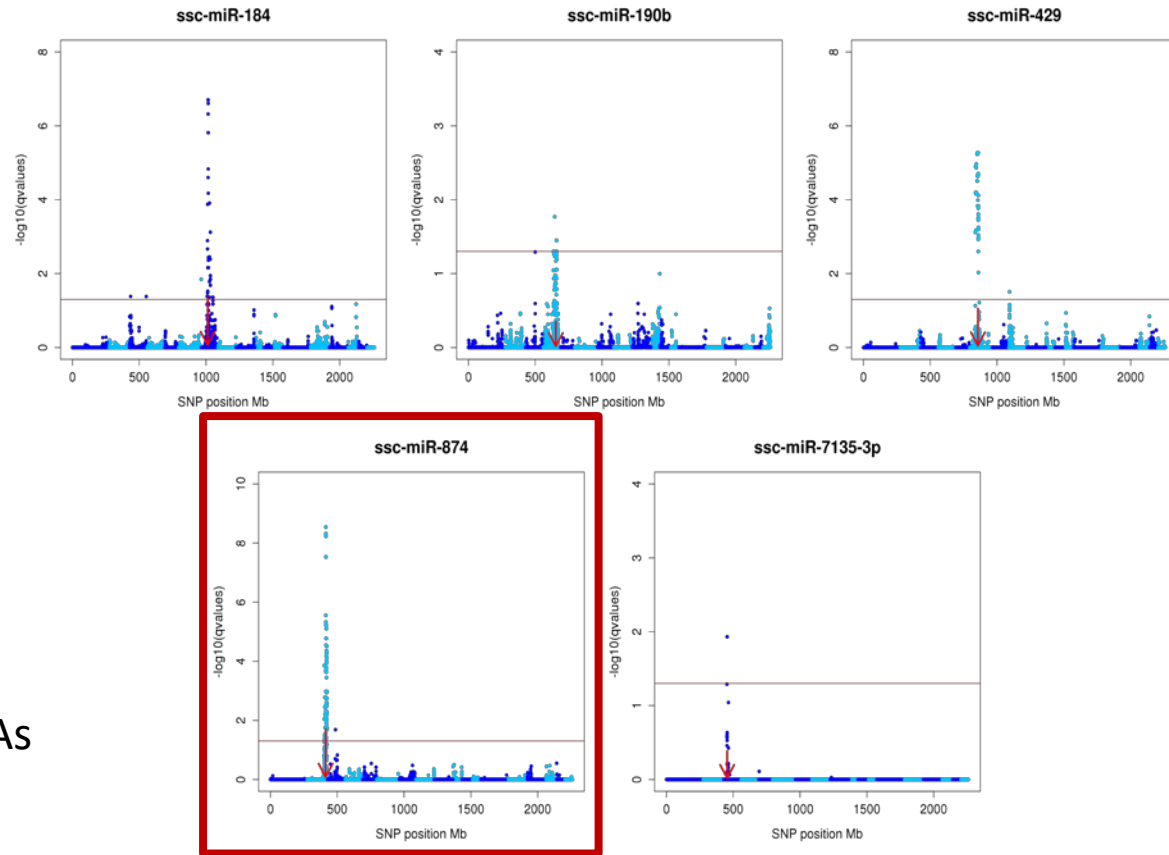
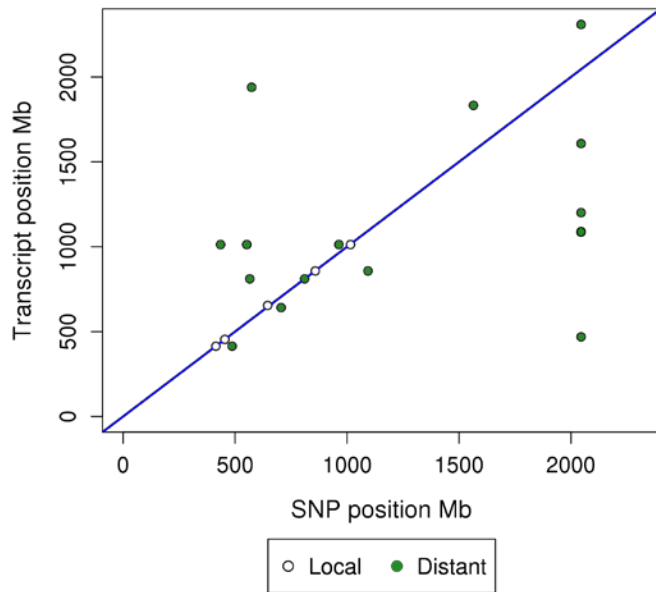
eQTL	Number ¹	%
Local	168	49
Plausible Local	23	7
Distant Same Chromosome	61	18
Distant	87	26
Total	339	-

¹FDR ≤ 0.01

Joint Association eQTL with pQTL



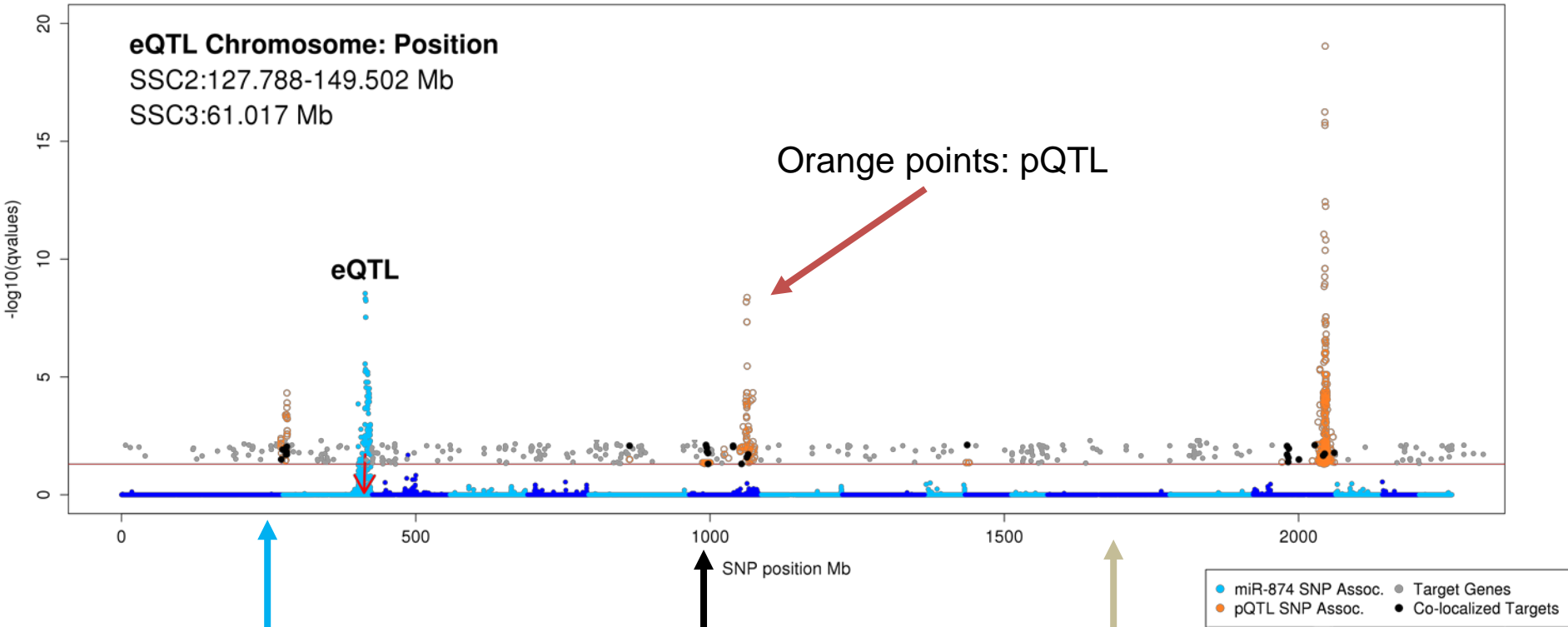
miRNA eQTL Results



- 23 miR-eQTL; 17 unique miRNAs
 - 5 local-acting
 - 16 distant-acting
 - 2 miRNAs on unplaced scaffolds

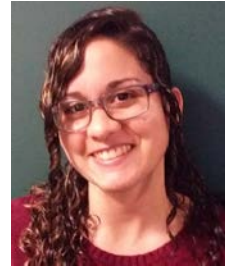
Genomic Co-Localization of miRNA eQTL with pQTL Results

ssc-miR-874

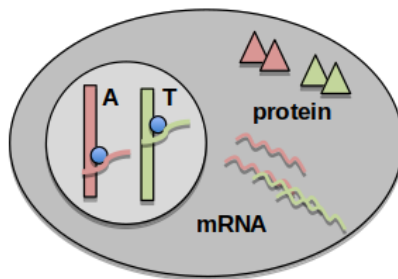


Blue points: miR-eQTL Black points: Co-localization (Target gene overlaps pQTL position) Gray points: Target Genes (Negative correlation)

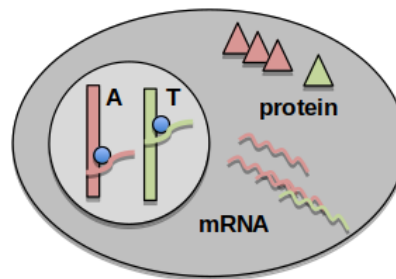




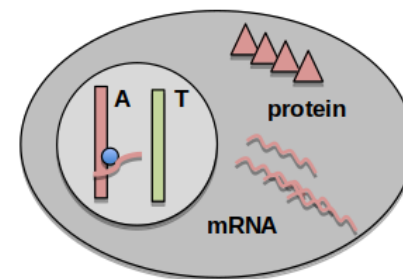
Allele Specific Expression Analysis



**Biallelic
Expression**



**Allele Specific
Expression**



**Monoallelic
Expression**



Test for Allele Specific Expression

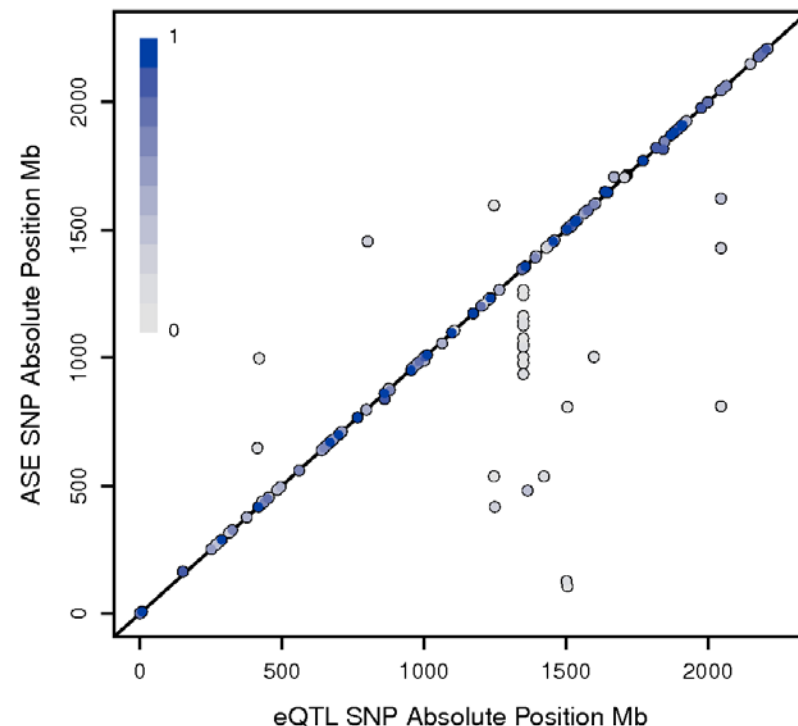
Quasi binomial model

cSNP	ASE	Genes
69,502	18,388	4,170

Expression QTL vs ASE

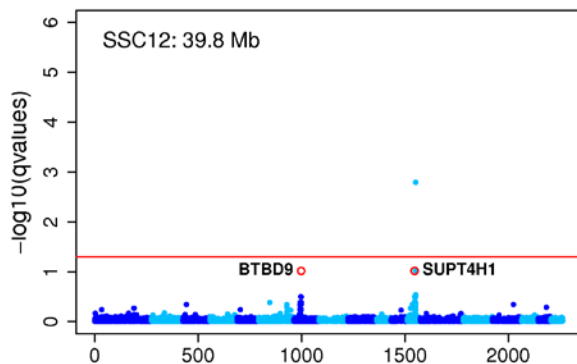
Analysis	Local	Distant Same Chr	Distant
eQTL	188	59	87
ASE	91	26	24
LD	70	20	-

Confirmed 91 local eQTL
78% in high LD with eQTL peak marker

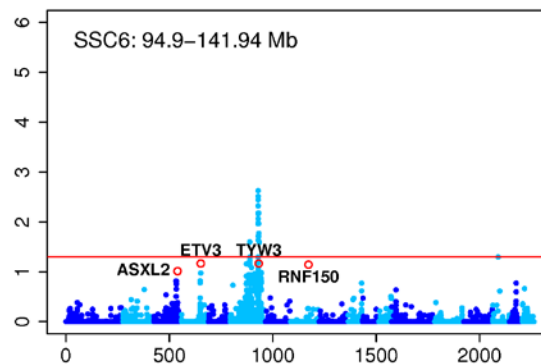


ASE Effects on Phenotype: cSNP pQTL

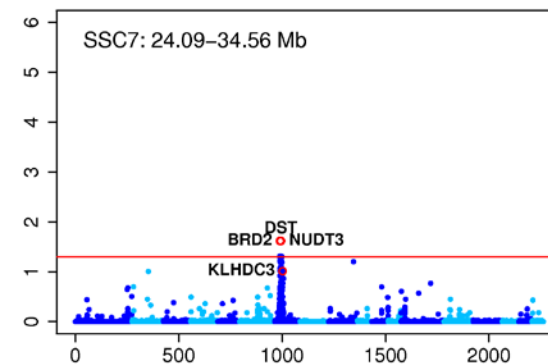
Last-Rib Backfat 22-weeks



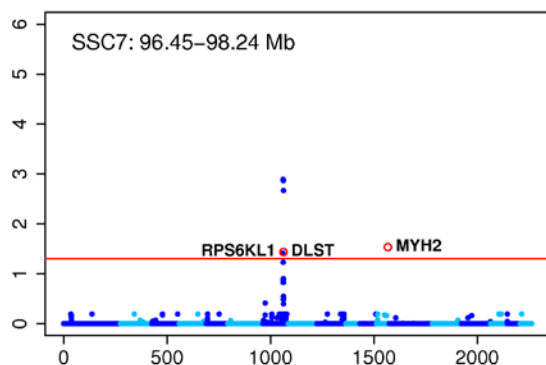
Carcass 10th-Rib Backfat



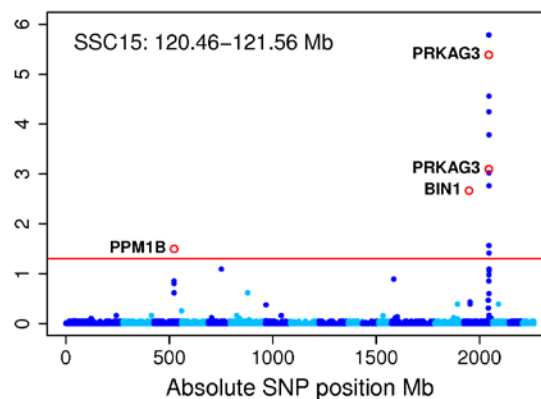
Carcass Length



Number of Ribs



Protein Percent

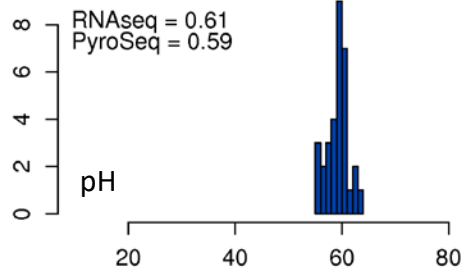


Animals: 168

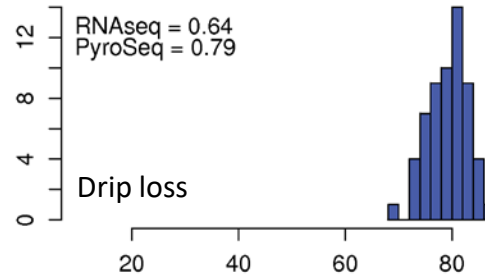
pQTL: 28,328 cSNP

ASE cSNP: 6,293

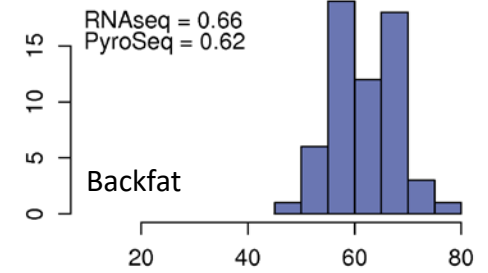
ZNF79



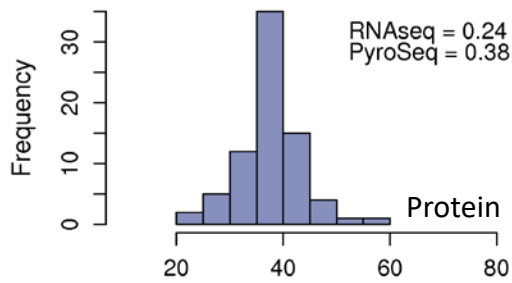
RNF141



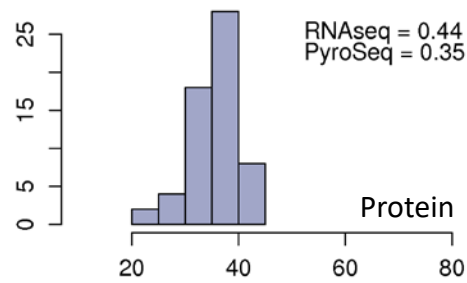
RNF150



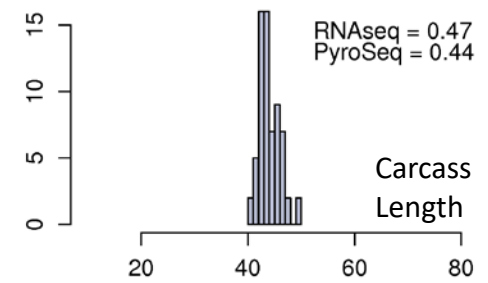
BIN1



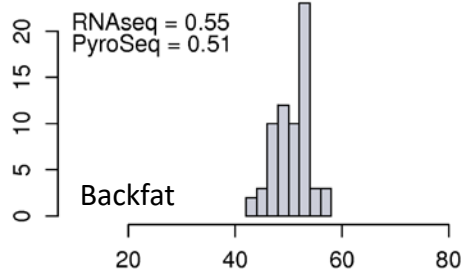
PRKAG3



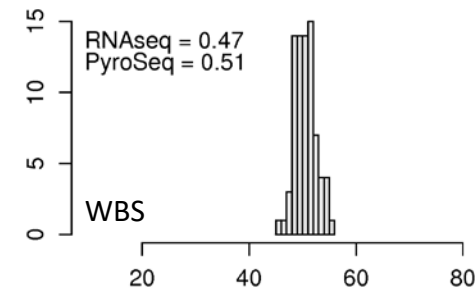
NUDT3



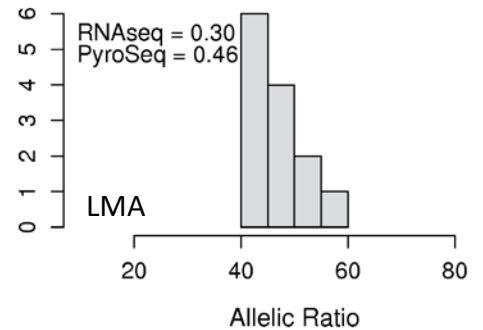
TYW3



NAMPT



PPARGC1B



Summary

- Joint analysis of pQTL with eQTL identified molecular markers associated with phenotypes and gene transcript abundance
 - mRNA eQTL
 - 334 mRNA eQTL
 - 16 mRNA eQTL associated with 21 pQTL
 - Redox homeostasis, energy metabolism, calcium signaling pathways
 - miRNA eQTL
 - 23 miRNA eQTL
 - 29 predicted miR-874 target genes co-localized with pQTL for 12 phenotypes
- ASE analysis identified 4,170 genes with cis-acting effects
 - Confirmed 91 eQTL genes with cis-acting effects
- Confirmed ASE for six genes with pyrosequencing
 - Zinc and ring finger genes show allelic imbalance associated with pH, drip loss and backfat phenotypes



Conclusion

- Integrated analysis combines multiple *-omics* datasets to provide a more comprehensive picture of genetic architecture of complex traits
 - Facilitate targeted research
 - Improve selection decisions
 - Increase production efficiency and pork quality

