

Expression quantitative trait loci (eQTL)

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DNA

ACTGACCTAGATCAGTGTAGCGATCGTATACGAGACCGATTTCATCGGCAT



transcription

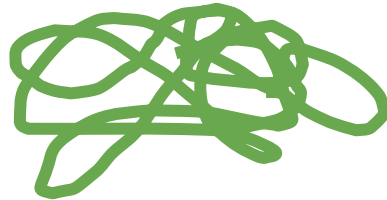
RNA

AUCAGUCGAUCACCGAU

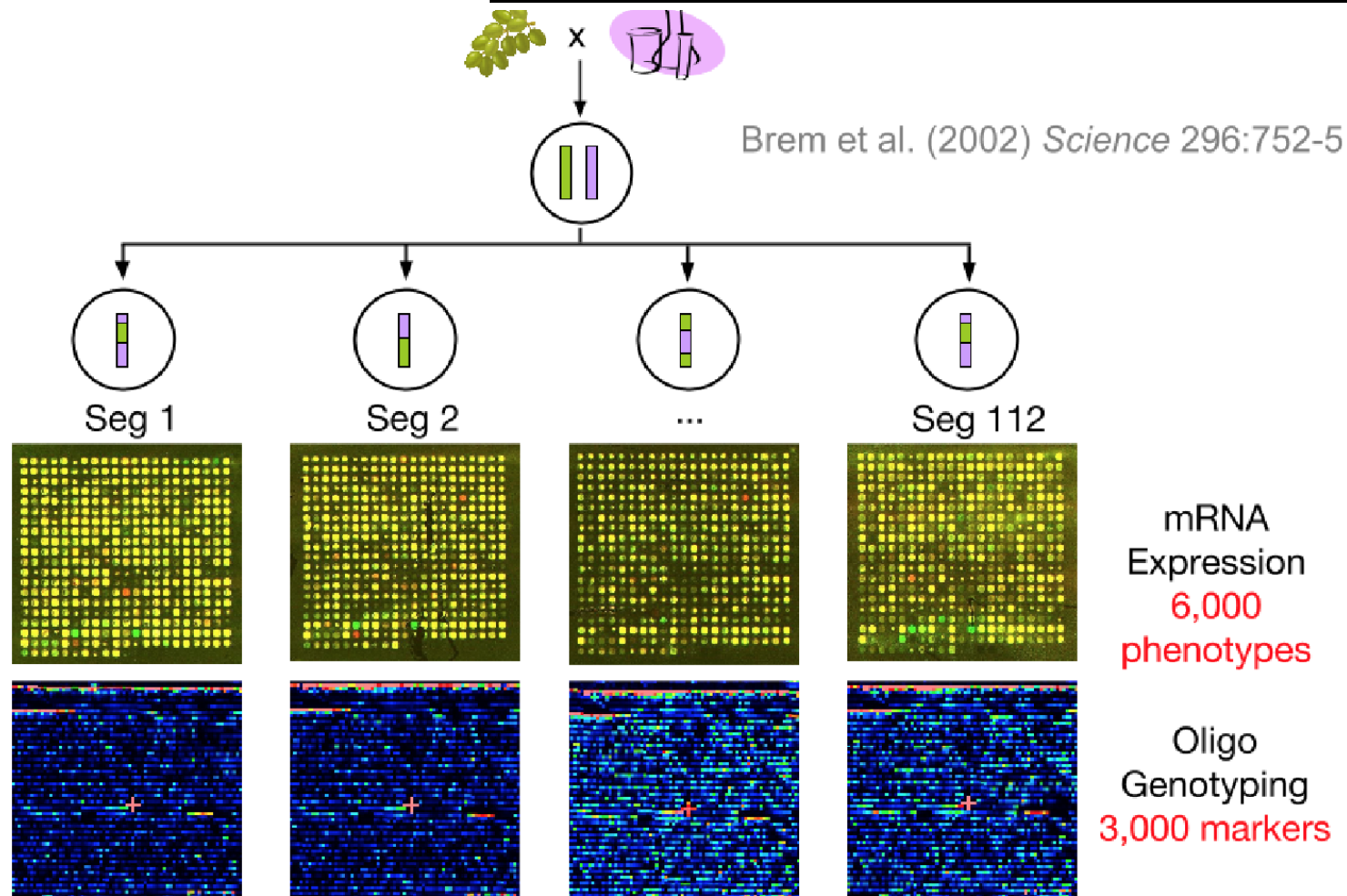


translation

protein



Study design



Chromosomes

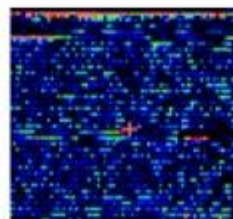
n=16 in yeast



...ACTACAGAC**CCA**ACCTGGGGATGCCGCGGAACAGGGAATAGCC...



Marker (SNP)
Position



Oligo
Genotyping
3,000 markers

Data

G_1, \dots, G_{1226}

$G_i = (g_{i1}, \dots, g_{i112})$

Chromosomes

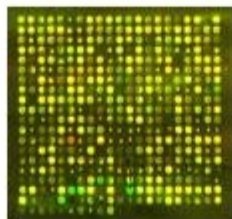
n=16 in yeast



...ACTACAGACC**CAACCTGGGG**ATGCCGCGGAACAGGGAATAGCC...



Trait (Gene)
Position

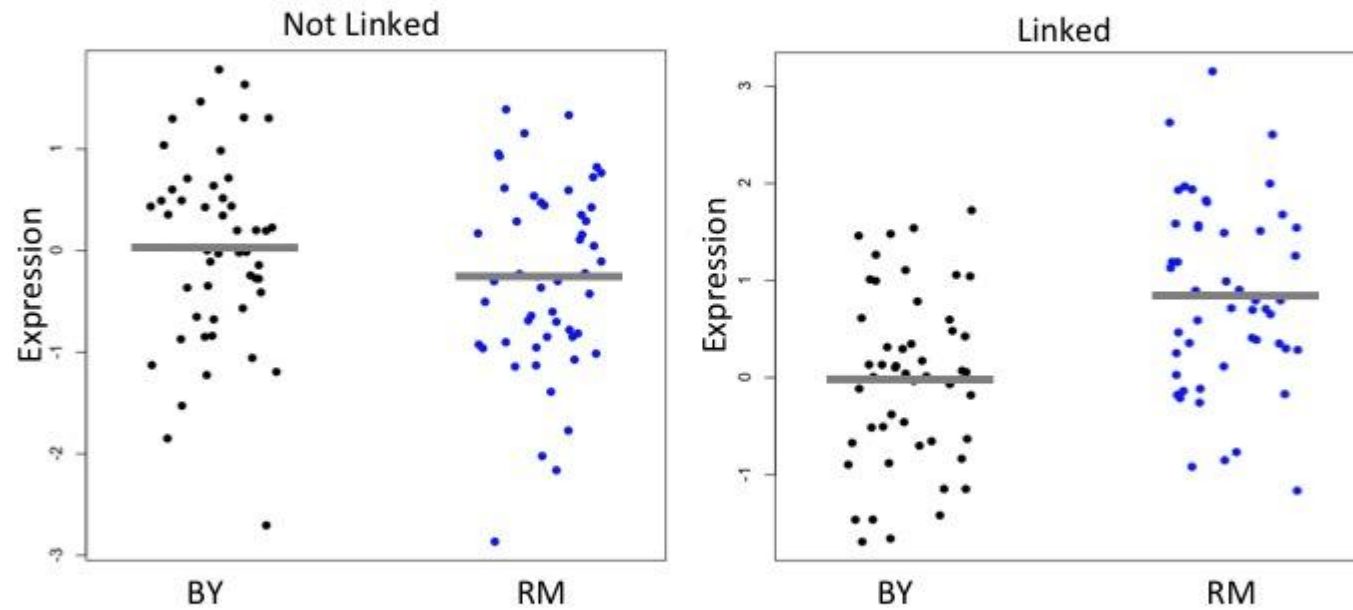


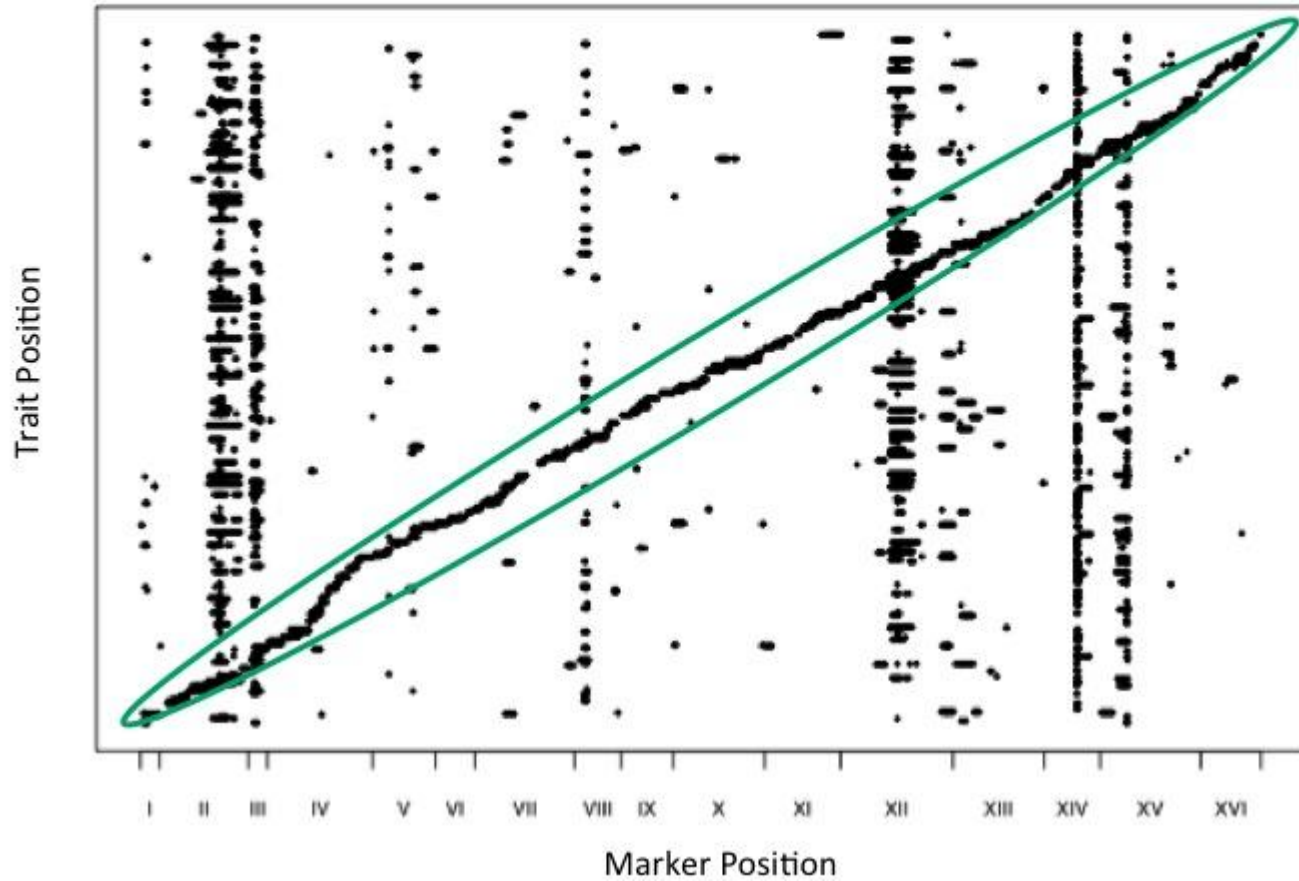
mRNA
Expression
6,000
phenotypes

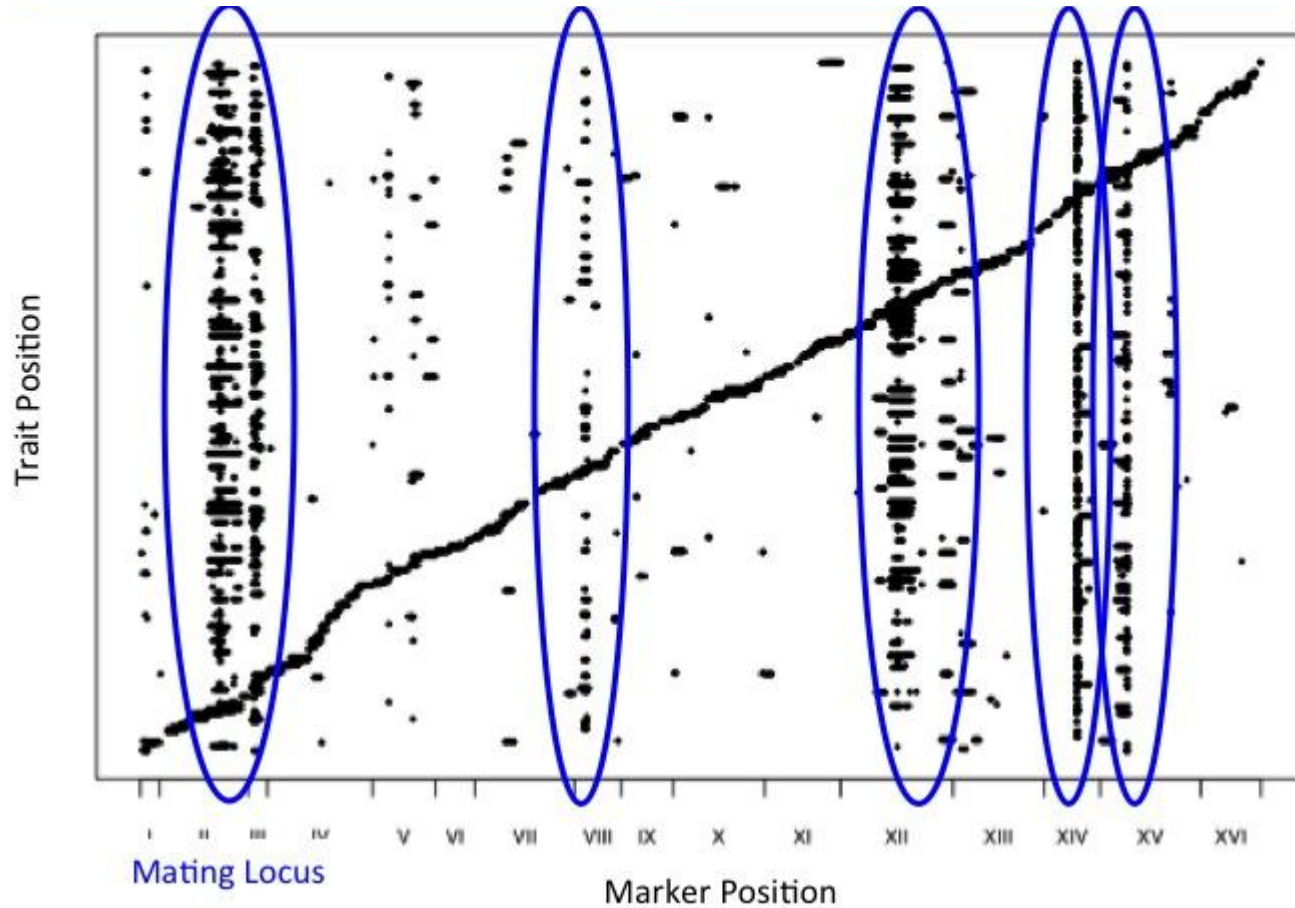
Data

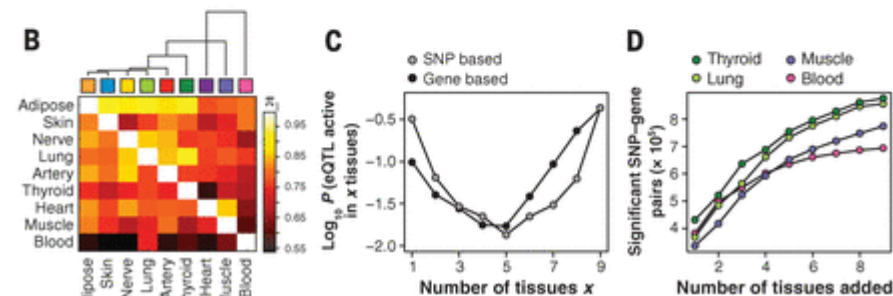
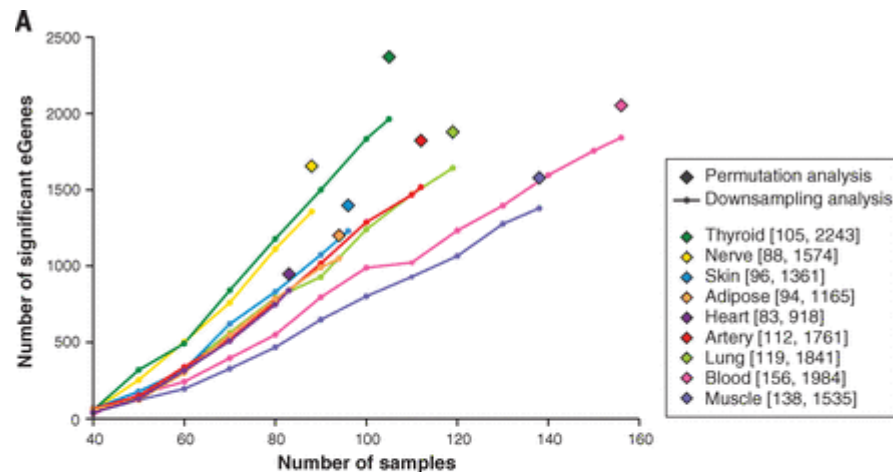
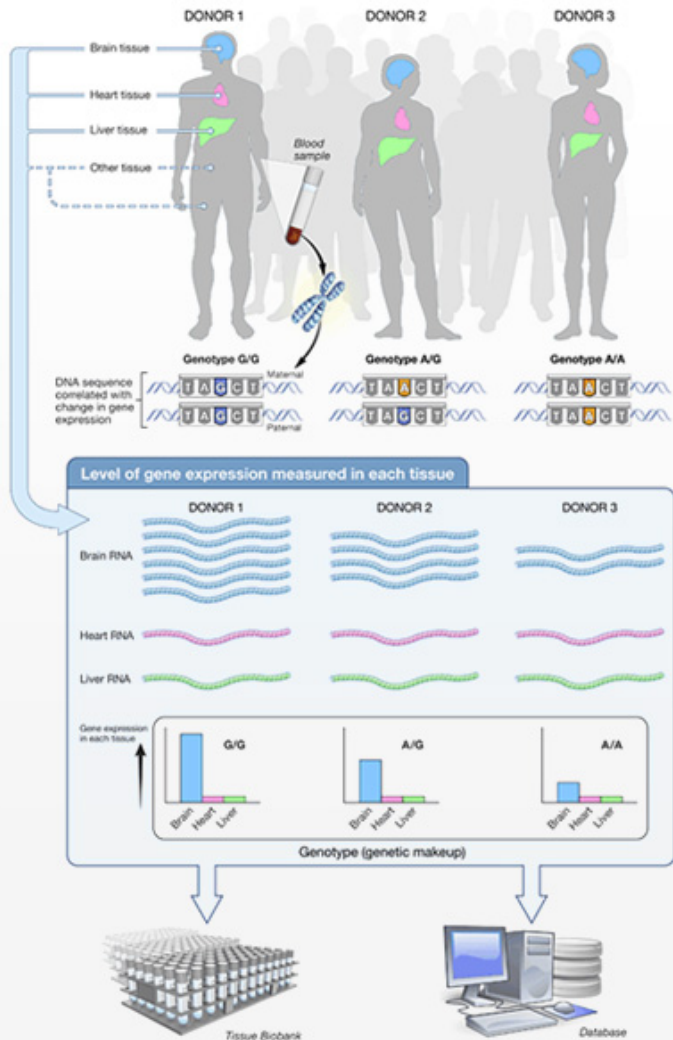
$$X_1, \dots, X_{6216}$$
$$X_i = (x_{i1}, \dots, x_{i6216})$$

Statistical model









Notes and further reading

- cis-eQTL are usually more believable than trans-eQTL
- There are many potential confounders
 - Population stratification, batch effects, sequence artifacts
- An excellent review
 - <http://www.nature.com/nrg/journal/v16/n4/abs/nrg3891.html>