

How genetic alterations propagate to organismal level traits?

Feb 2018

UNIL BSC course

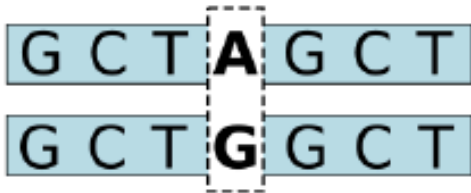
Supervisor: Sarvenaz Choobdar

Genetic variation



Genotype to Phenotype

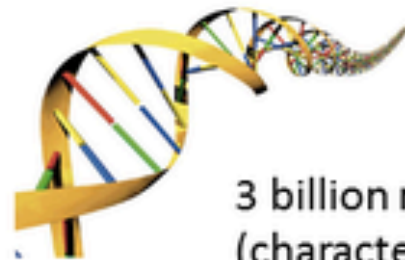
GWAS



Genome-wide Association Study (GWAS)



DNA Sequencing



3 billion nucleotides (characters)

Case (with disease)



DNA Sequencing



GWAS aims to find the associations between genetic variations and observable traits.

Control (without disease)



DNA Sequencing

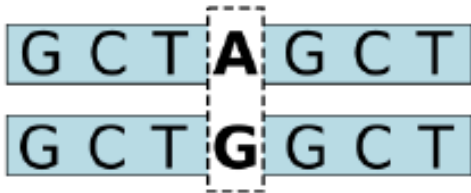


GWAS catalog



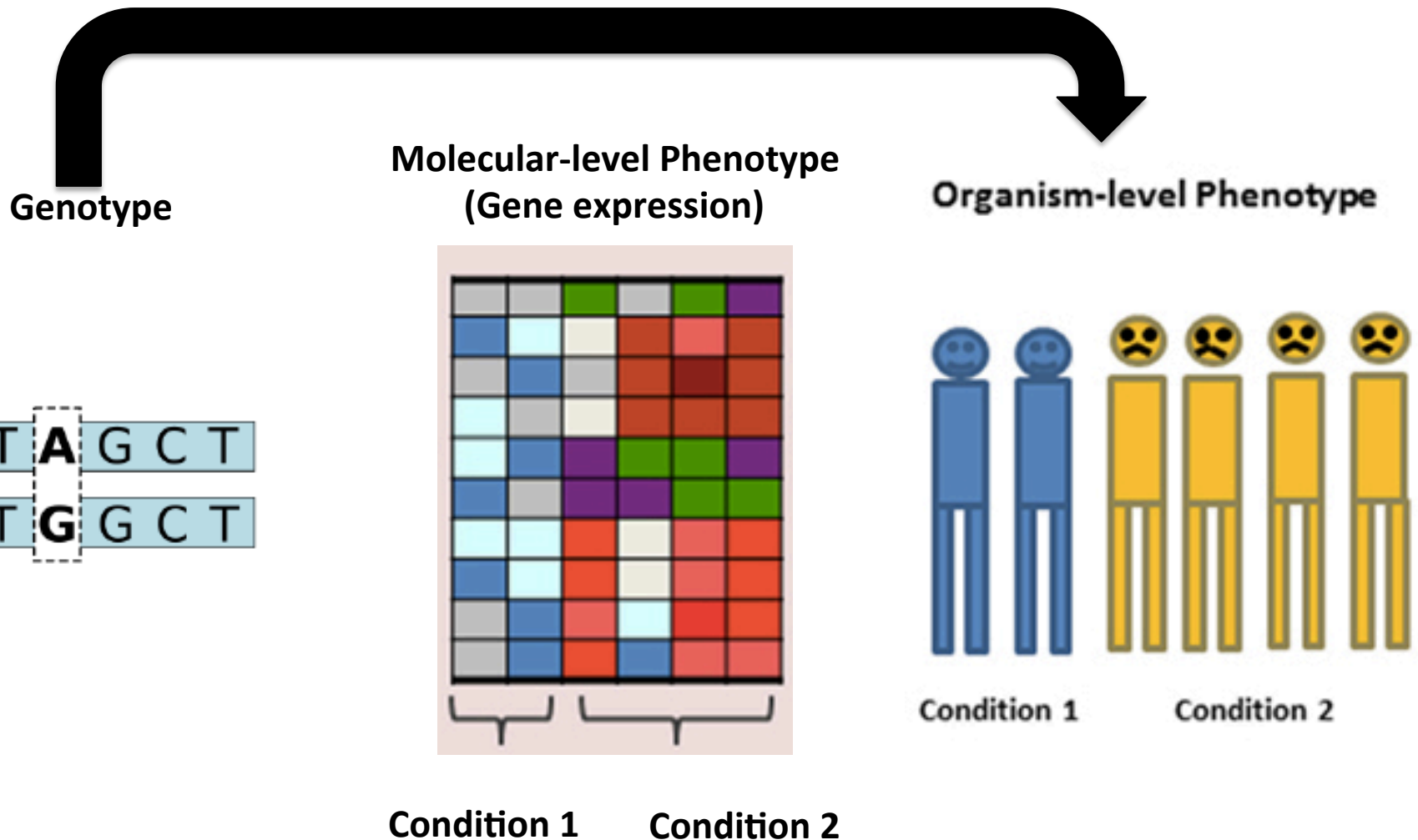
Genotype to Phenotype

GWAS



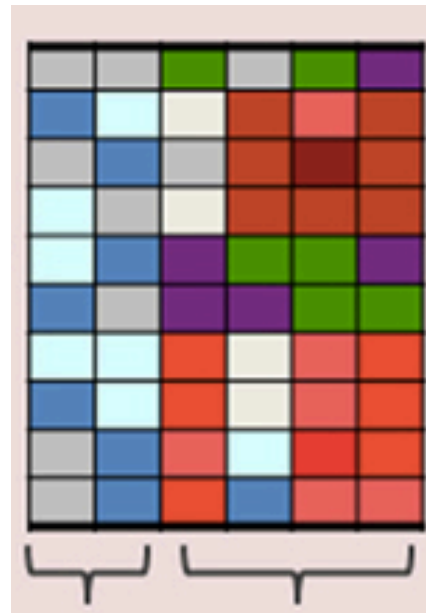
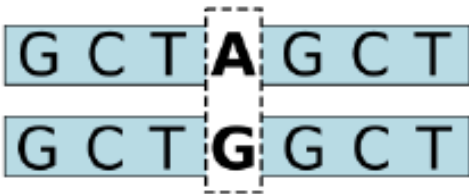
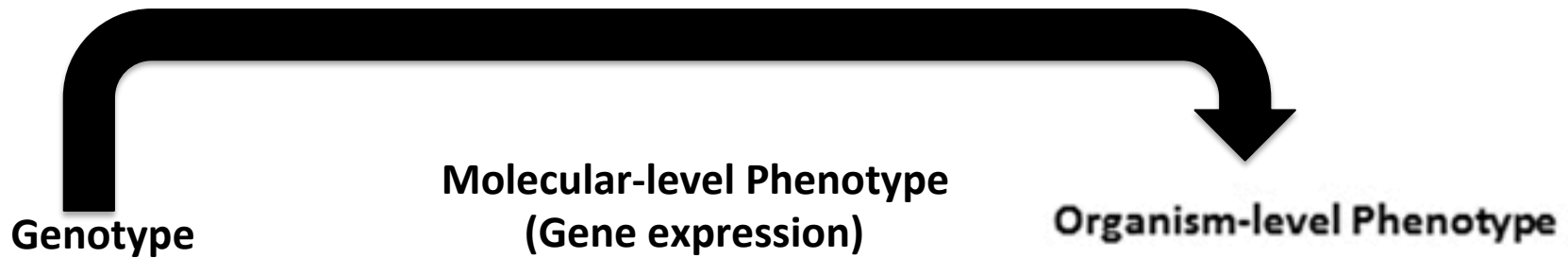
Genotype to Phenotype

GWAS



Genotype to Phenotype

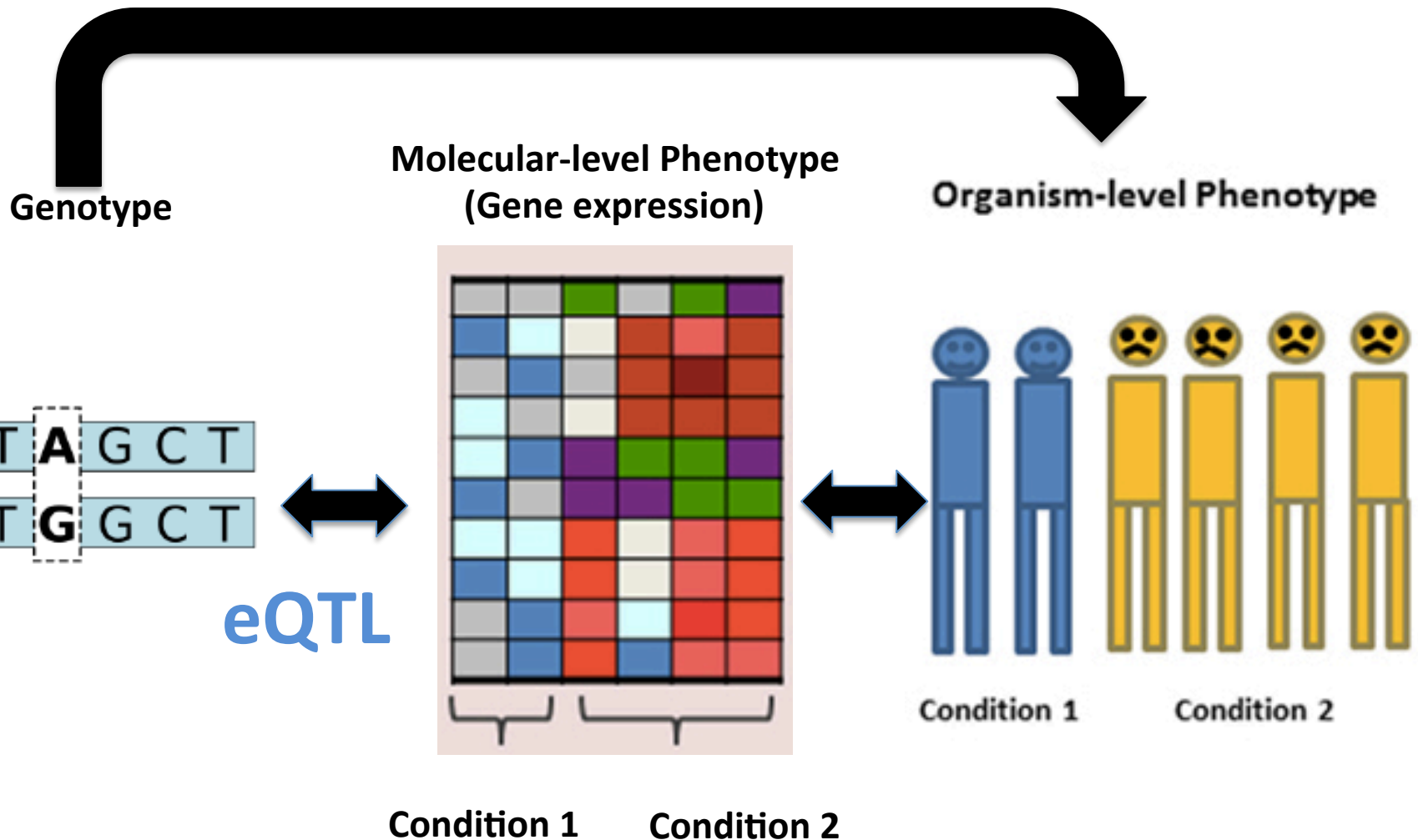
GWAS



Condition 1 Condition 2

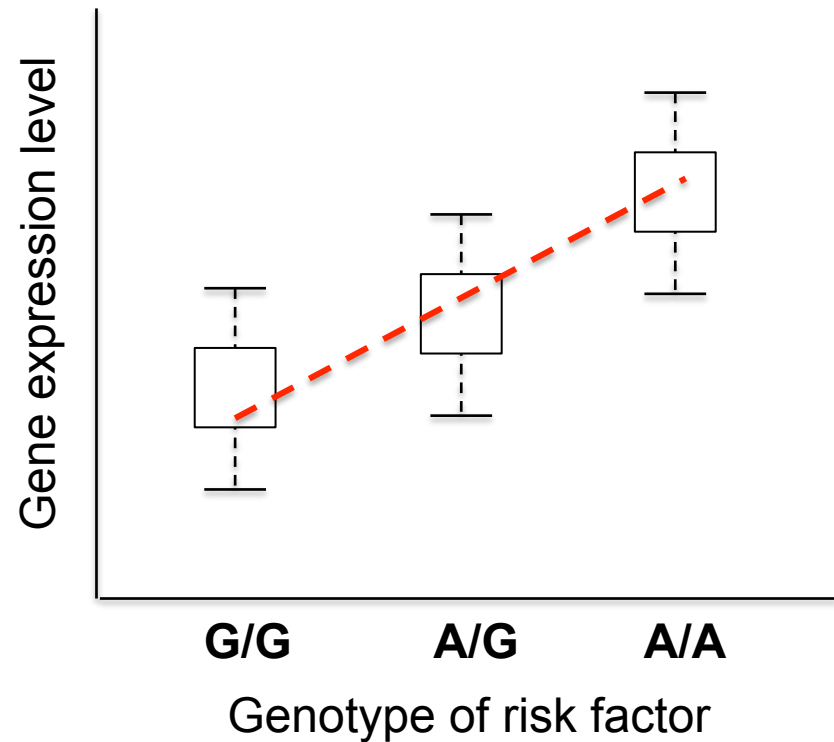
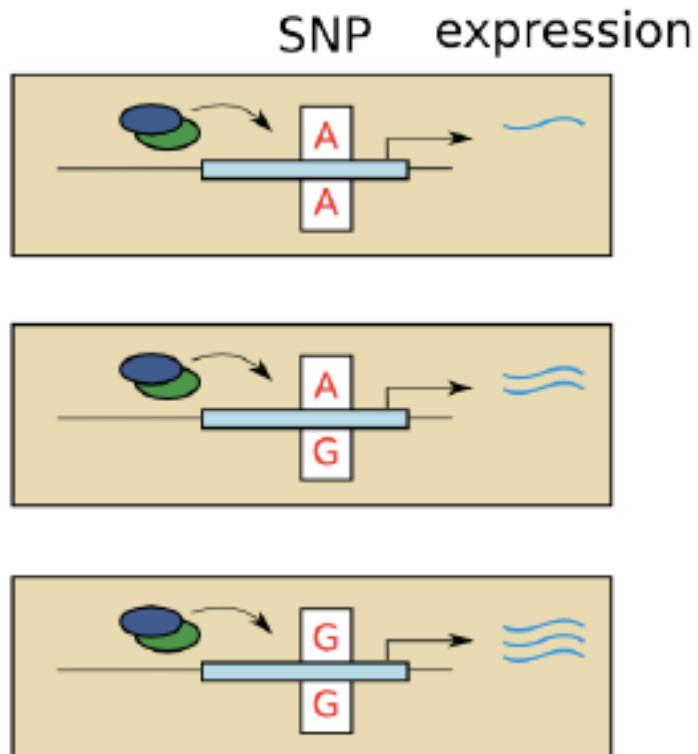
Genotype to Phenotype

GWAS



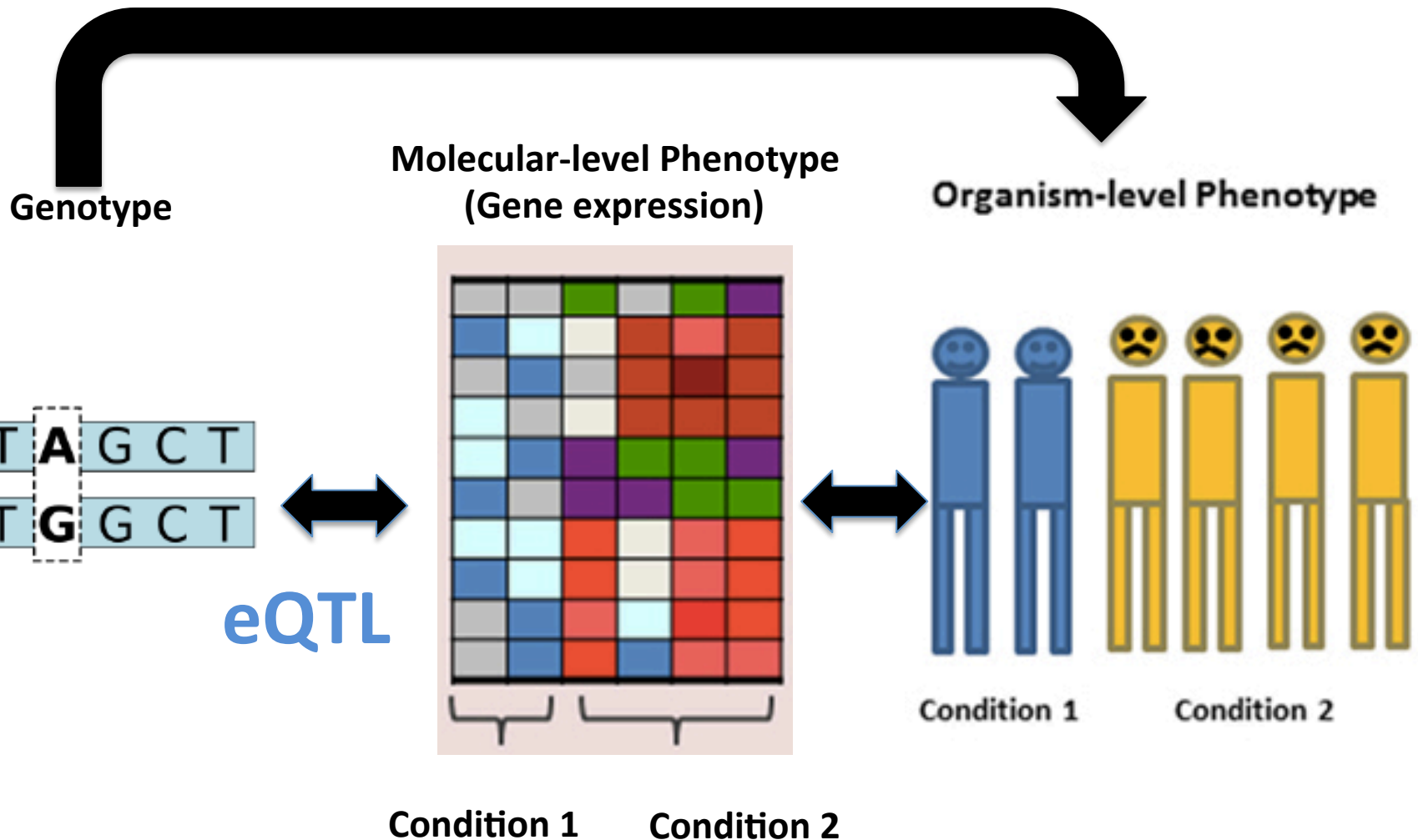
eQTL analysis

- **expression Quantitative Trait Loci (eQTL)**
- **Goal** : identify genomic locations where genotype significantly affects gene expression



Genotype to Phenotype

GWAS

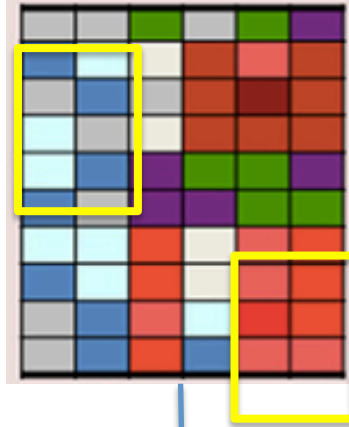


Project roadmap

Genotype

Phenotype

Molecular-level Phenotype
(Gene expression)



Modules

GWAS summary for
Cardio Vascular
Disease

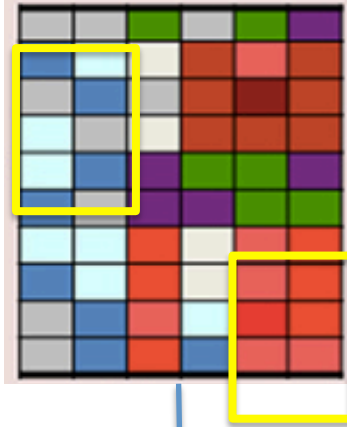


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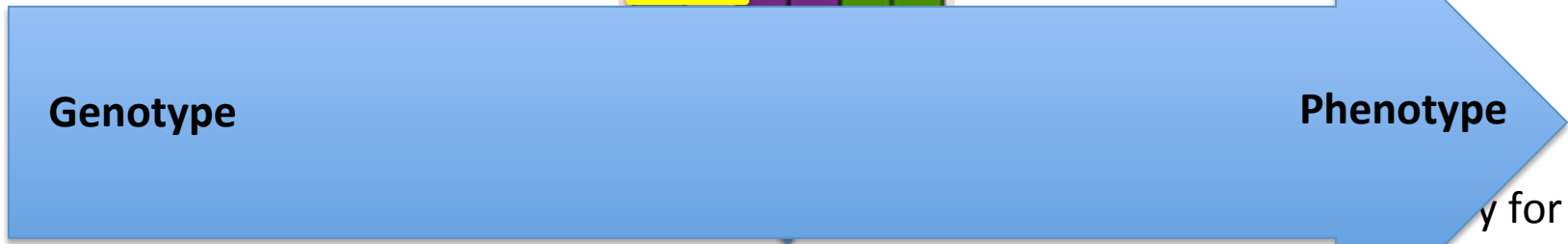
eQTL

Disease Modules



Project roadmap

Molecular-level Phenotype
(Gene expression)



Modules

Cardiovascular
Disease

eQTL

Disease Modules



Thank you

QUESTIONS?