

eQTL Analysis

Introduction of eQTL Analysis

The expression Quantitative Trait Loci (eQTL) refers to some regions on the chromosome that can specifically regulate the expression levels of mRNA and protein. The expression level of mRNA/protein is proportional to the quantitative traits. eQTLs can be divided into *cis*eQTLs and *trans*eQTLs. A *cis*eQTL is the eQTL of a gene that maps to the genomic region where the gene is located, indicating that the mRNA level may be changed by the difference of the gene itself. A *trans*eQTL means that the eQTL of a gene is mapped to other genomic regions, and differences in other genes lead to differences in the mRNA expression level of this gene.

In the process of genetic research, some disease-causing or susceptible mutations are often found, but how these mutations cause phenotypic changes is not intuitive. Therefore, the differential expression of a gene is used as a transition, from gene mutation to gene expression change, and finally to phenotype. The specific principle of eQTL analysis is to use gene expression as a quantitative trait to study the correlation between genetic mutation and gene expression, so as to study the correlation between genetic mutation and disease or phenotypic characteristics.

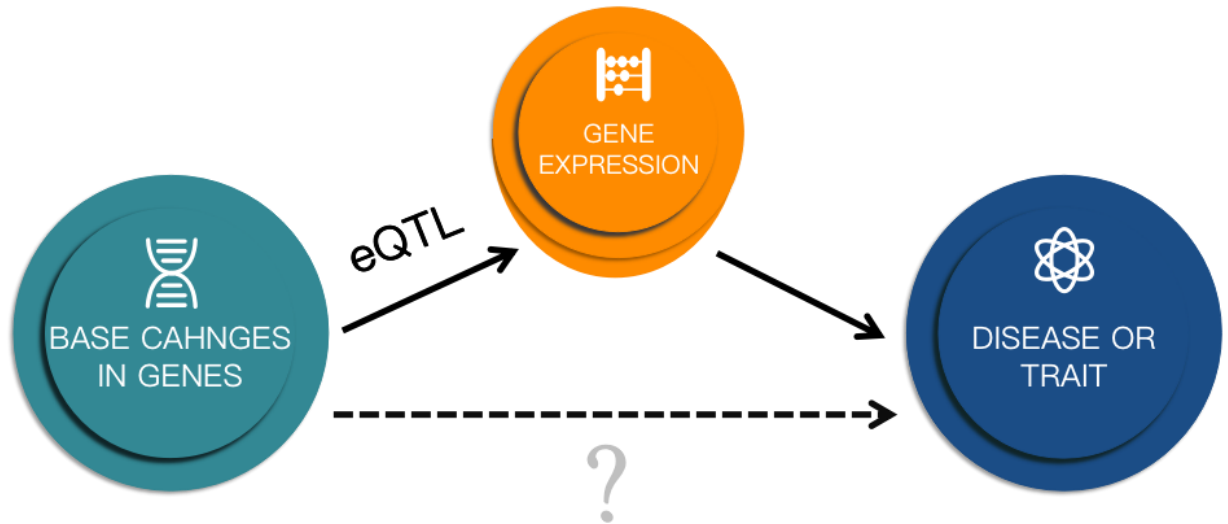


Fig 1. eQTL is a bridge between genetic changes and diseases.

eQTL Analysis Process

The eQTL analysis process can be divided into two three steps:

Firstly, perform whole genome-wide association analysis to detect various mutations in the coding region and non-coding region, and perform association analysis to screen disease-causing or susceptible sites.

Secondly, the mRNA sequencing data is analyzed, and the mRNA expression level is analyzed to obtain information such as differential genes, variable splicing, and new transcripts.

Finally, eQTL analysis is performed, and the candidate pathogenic sites or susceptible sites obtained from the whole genome-wide association analysis and the expression information obtained from mRNA data analysis are subjected to eQTL analysis to further narrow the candidate pathogenic or susceptible sites information, and then establish the network relationship between gene mutation and expression regulation through various models, and explore the molecular mechanisms related to the occurrence and development of diseases. The specific analysis content of eQTL is as follows:

- Cis eQTL analysis.
- Trans eQTL analysis.
- eQTL annotation, including ENCODE database comment and GETx database comment.
- eQTL combined with GWAS analysis.
- Differential cis eQTL analysis (two or more samples).
- Differential trans eQTL analysis (two or more samples).

Advantages of eQTL Analysis

- Using eQTL combined with GWAS analysis methods to find disease risk sites can be applied to complex disease research.
- The eQTL analysis can explain the regulatory relationship between gene mutations in non-coding regions and gene expression. And systematically understand the regulatory mechanism of gene transcription and construct a gene expression regulatory network.
- Performing eQTL difference analysis on the body or cells before and after drug treatment can well study the difference before and after drug treatment, and provide gene-level evidence for finding drug targets and drug action mechanisms.
- In population cohort studies, eQTL analysis can be used to compare eQTL differences in different populations, thereby enriching the results of population cohort studies, and finding the relationship between gene differences and gene expression regulation in different populations.

Application Filed

Using Bayesian sparse linear mixed model phenotype prediction model method can improve the prediction accuracy of genome-wide genetic locus information. The success of GWAS has greatly promoted the use of genetic information (except environmental and lifestyle information) to carry out complex disease risk prediction and assessment.

- Research on susceptibility sites of complex diseases.
- Analysis of drug targets.
- Population cohort study.

MedAI provides eQTL analysis based on PrediXcan to help you study the correlation between genetic mutation and disease or phenotypic characteristics. In addition, we also provide eQTL analysis with Matrix, FastMap, Merlin, Plink and cutting-edge analysis software. In the process of analysis, we will provide you with different software or different analysis models according to your needs. If you have any questions, please feel free to [contact us](#), we look forward to working with you.

References

1. Xu Z, Wu C, *et al.* A Powerful Framework for Integrating eQTL and GWAS Summary Data[J]. *Genetics*, 2017:*genetics.300270.2017*.