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Association Mapping - Disclosing Genetic Evolution (^{*}Anuka Yadav¹ and Aranav Yadav²) ¹National Dairy Research Institute, Karnal, Haryana, India ²Chandra Shekhar Azad University of Agriculture and Technology, Kanpur, U.P., India ^{*}Corresponding Author's email: <u>aranav.yadav@gmail.com</u>

Abstract

A technique for locating quantitative trait loci is association mapping, commonly referred to as "linkage disequilibrium mapping" (QTLs). Linkage disequilibrium is the term used to describe the non-random pairing of alleles at different loci in a population (LD). Linkage disequilibrium is a method for determining how closely connected two different loci's alleles are to one another. It is known as association mapping and is typically quicker and less expensive than traditional linkage mapping when looking for genotype-phenotype connections in unrelated individuals. Finding new genes or QTLs connected to crucial agronomic traits can be done by using association mapping. This approach is anticipated to be widely applied in agricultural plants in the long run as a result of the advancement of novel high-throughput genotyping and sequencing technologies. The enormous number of variations with tiny effects revealed by GWA research against the small number of genomic regions with substantial impacts discovered by linkage mapping hinders our current understanding of the genetic architecture of complex features. Population structure continues to be a substantial barrier to association research, calling for careful germplasm selection and the creation of novel statistical techniques. Association mapping significantly improves the capacity to address the specific limitations of each technique.

What is Association Mapping?

A technique for mapping quantitative trait loci (QTLs) known as association mapping, commonly referred to as "linkage disequilibrium mapping," links phenotypes—observable characteristics—and genotypes—an organism's genetic make-up—to show genetic relationships.



Linkage Disequilibrium (LD)

Linkage disequilibrium is the non-random association of alleles at different loci in a population (LD). Linkage disequilibrium measures the degree of relatedness between alleles at two distinct loci.

The population of alleles that share a certain chromosome is what the expression was first used to refer to. Even though LD is a phenomenon that occurs within populations, it is frequently seen that LD between alleles that are positioned closer together is greater. Linkage may thereby reduce the chance of an allele connecting randomly to another, leading to socalled disequilibrium. The organization of LD is influenced by a variety of genetic and nongenetic variables, including recombination, drift, selection, mating pattern, and mixing. For association mapping, the LD between functional loci and physically related markers is essential.

Linkage disequilibrium is an essential element in association mapping. The degree of LD can be estimated using a variety of statistical metrics, including r2, which assesses the correlation between the allelic states of two polymorphic loci. Linkage disequilibrium can be drastically overstated when sample sizes of less than 50 people are used.

Types of Association Mapping

• Genome wide association mapping

Main advantage: Allows untargeted fine mapping (blind approach), detection of common alleles.

Main disadvantage: Population structure's confounding impact will miss rare and minimally significant alleles.

General requirements: Large population size, a great number of genetic markers (fewer repeats are needed the larger the population), prior knowledge of LD, nucleotide polymorphism, breeding practices, and population structure.

Candidate gene association mapping

Main advantage: Allow fine mapping, relatively low costs.

Main disadvantage: No novel characteristic will be found; detailed functional understanding of the trait is required.

General requirements: Few genetic markers, big population size; fewer repetitions needed as population size increases. It is necessary to have prior understanding of the genetic and physiological underpinnings of a trait of interest, as well as knowledge of LD, nucleotide polymorphism, breeding systems, and population structure.

What is the goal of Association Mapping?

Association mapping is the process of looking for genotype-phenotype relationships in unrelated individuals, and it is typically quicker and less expensive than traditional linkage mapping.

Linkage and association mapping are complementary approaches and are more similar than is often assumed.

Advantage of Association Mapping

- Association mapping is a practical method for discovering new genes or QTLs linked to crucial agronomic features. This approach is anticipated to be widely applied in agricultural plants in the long run as a result of the development of novel high-throughput genotyping and sequencing technologies.
- Gene-based markers are more reliable than linked markers in predicting phenotype because the marker-trait relationship is maintained during segregation during breeding selection cycles.

- To determine the appropriate haplotype for the best expression of the target trait across one or more genes, association analysis data can be used.
- Genome-wide association studies are now being used by human geneticists to map disease genes.
- Genes that contribute to phenotypic variation in wild populations can be pinpointed with high resolution using association analyses based on correlations between alleles at different sites or LD.

Conclusion

There is a lot of promise for agricultural genetics improvement through association mapping. Due to the use of high throughput and reasonably priced next generation sequencing techniques, GWA research will become a popular and common methodology. However, association mapping still has value as a substitute for linkage mapping and other methods of gene identification and validation. The enormous number of variations with tiny effects revealed by GWA research against the small number of genomic regions with substantial impacts discovered by linkage mapping hinders our current understanding of the genetic architecture of complex features. For association studies, population structure remains a significant restriction that necessitates careful germplasm selection and the development of new statistical methods. The ability to resolve the individual restrictions of each technique is greatly enhanced by association mapping.

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