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Unlocking Genetic Mysteries: Genome Mapping in Polyploids

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Introduction

Polyploids are organisms with more than two sets of chromosomes. They are very important in agriculture and play a fundamental role in evolutionary processes, such as differentiation of species (Soltiset al., 2016). One of the most fundamental descriptions of any organism is its ploidy level and chromosome number. Plant scientists in particular will be familiar with this representation of the chromosomal constitution of the sporophyte generation (i.e. the adult plant). The second term in this seemingly simple equation describes the normal complement of chromosomal copies possessed by a member of that species, which is generally 2x ("two times") for diploids. Species where this number exceeds two are collectively referred to as polyploids. Not unexpectedly, each polyploid individual is the product of the fusion of gametes from two parents, just like their diploid counterparts. In other words, polyploids can also be defined as individuals derived from non-haploid gametes (in the case of triploids derived from diploid × tetraploid crosses, only one gamete satisfies this condition). The transmission of non-haploid gametes is one of the main "complexifying" features of polyploidy, leading to a whole range of implications for the genetic analysis of these "hopeful monsters" (Goldschmidt, 1933)

Gene mapping refers to the process of determining the location of genes on chromosomes. Today, the most efficient approach for gene mapping involves sequencing a genome and then using computer programs to analyze the sequence to identify the location of genes. Scientists are often

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most interested in the parts of a genome that directly encode proteins, in other words the protein-coding genes. So oftentimes, there is a priority to identify the location of all those genes in a genome. The process of determining the location of genes in a genome is called gene mapping. As with all forms of genome mapping, the old days involved approaches like sophisticated genetic studies or very tedious processes of cloning bits and pieces of a genome, studying them in the laboratory, and figuring out how things were organized relative to one another, including the relative locations of genes. But nowadays, such a mapping process usually involves sequencing a genome and analyzing the resulting sequence using computational tools that allow you to identify landmarks of interest, such as genes. So nowadays, most gene-mapping efforts first involved sequencing a genome.

How does Polyploidy affect genome mapping?

Polyploidy significantly affects the co-expression of duplicated genes, potentially eliminating, reducing, or increasing gene expression. These changes can occur with the onset of polyploidy or after several generations and can also be influenced by epigenetic factors. On the other hand, many changes in gene expression may occur only in a specific organ, so the relative expression of duplicated genes can vary in different parts of the plant.

In polyploids, the activity pattern of genes varies greatly. Studies on *Arabidopsis* plants have shown that some of the genes that were active in the diploid state were shut down in the autotetraploid state, but the cross between two tetraploid *Arabidopsis* species led to the production of allotetraploid plants in which these genes reactivated. In addition, a gene called rad54, which was not expressed in diploid *Arabidopsis* leaves, was activated in the leaves in the autotetraploid state. Recent experiments demonstrated that the various ploidy levels in a plant (autoploids or alloploids) and even the previous generations play a role in the level and stability of gene expression. There is considerable discussion about the factors that affect the activity of genes and their expression in polyploidy, including the activation of dormant transpositions in synthetic polyploids, which can cause gene extinction. Other factors that silence duplicated genes in polyploids include deacetylation, methylation, and changes in histones and chromatin structure.

Genome Mapping Techniques:

Recombination events are used in genetic mapping techniques to measure the distance between genetic markers.

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- Random Fragment Length Polymorphism, or RFLP, measures the differences in homologous DNA sequences to calculate the distance between two markers.
- Currently, gene mapping analyses targeting single gene disorders use short tandem repeat polymorphisms (STRP).
- SNP (Single Nucleotide Polymorphism) is used in genome-wide association and linkage analysis genetic research. Linkage analysis is studied using the inheritance of characteristic and genetic signatures like SNPs and microsatellites.
- Genome-Wide Association (GWA) studies the connections between traits and markers like SNPs and microsatellites by treating the population as a single family. The method is used to map the gene functions of common disorders
- Genetic mapping involves creating linkage maps and performing quantitative trait locus (QTL)
- Physical mapping enables the fingerprinting of large genome fragments and can be used to improve scaffolding.

Some molecular markers used for genetic mapping in polyploids

The molecular markers commonly used for genetic mapping in polyploids include:

- **Single Nucleotide Polymorphisms (SNPs):** SNPs are nucleotide positions that differ in the individuals being screened and are usually selected to be bi-allelic. They are important for genetic mapping in polyploids, and the methods developed for SNP markers are general to any bi-allelic marker system for which marker "dosage" counts can be accurately estimated
- **Genomic In Situ Hybridization (GISH):** This technique involves the use of total genomic DNA and is used for exploring polyploid genomes in plants. It can be valuable for genetic mapping in polyploids
- **Amplicon Sequencing and High-Resolution Melting (HRM):** HRM is a technique that can identify mismatches, even for single bases, in amplicons containing heteroduplex molecules, and is emerging as a powerful tool for polyploid genetics. It has been demonstrated to be a cost-effective approach for SNP discovery targeted to polyploids.

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- **Methylation-Sensitive Amplified Polymorphism (MSAP):** This method is based on the use of iso-schizomers affected by methylation and is used for methylation-sensitive molecular markers. It is valuable for studying DNA methylation in polyploids.

These molecular markers and techniques play a crucial role in genetic mapping and understanding the complex genomes of polyploid species.

Different between genetic mapping and physical mapping

	Physical Mapping	Genetic Mapping
Definition	It is a technique which shows the physical distance in a DNA sequence by working out the number of base pairs	It is a technique which shows how genetic information is shuffled in a chromosome.
Molecular Markers	It uses restriction enzymes to cut the specific sequence of DNA.	It uses genetic markers to map the distance between two genes.
Importance	It depicts the actual distance of base pairs along a stretch of DNA.	It depicts the region of polymorphisms (region where the DNA sequence differs) in different individuals.
Accuracy	It depicts a more accurate representation of the genome.	It gives mere insights for different regions of chromosomes.

Challenges in Genome mapping in Polyploids

Genome mapping in polyploids presents several challenges due to their complex genetic makeup, including multiple sets of chromosomes. One of the principal challenges faced by polyploid organisms is to evolve stable meiotic mechanisms to faithfully transmit genetic information to the next generation. The higher ploidy levels of polyploid plant genomes make the situation even more difficult, leading to highly fragmented genome assemblies, with disconnected contigs of repetitive sequences. Though reference genomes are now available for many crops, only diploid/haploid references are available for polyploidy crops like potato, coffee, strawberry and banana and most highly heterozygotic genomes have only one sequenced haplotype. Some of the elements contributing to agronomic traits like gene duplications, genome rearrangements and repeat integrations are cultivar specific, and cannot be found using resequencing strategies or reduced complexity references. High quality references of each sub

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genome (in polyploids) and each haplotype (in heterozygous crops) as well as multiple references per crop species are needed to survey true variation

Conclusion

In conclusion, genome mapping in polyploids presents unique challenges due to their complex genetic makeup, including multiple sets of chromosomes. However, significant progress has been made in this field through the development of various techniques and approaches. These include genetic and physical mapping, molecular marker-based genetic mapping, and the use of candidate gene approaches such as comparative genomics, transcriptomics, association mapping, and functional genomics. Despite the challenges, the ongoing development of tools and technologies offers the promise of understanding polyploid genomes at a deeper level. The study of meiosis in polyploid species has improved linkage estimation, allowing the construction of multi-locus genetic maps, and the recent inclusion of allelic dosage information has further advanced the field of genetic mapping in polyploids. Overall, the continued advancement of genomic tools and resources will undoubtedly enhance our understanding of polyploid genomes and their implications for agriculture, evolution, and biodiversity.

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