Poliploidija kao mehanizam specijacije u biljaka

Sesartić, Marko

UNIVERSITY OF ZAGREB
FACULTY OF SCIENCE
BIOLOGY DIVISION

SVEUČILIŠTE U ZAGREBU
PRIRODOSLOVNO-MATEMTIČKI FAKULTET
BIOLOŠKI ODSJEK

Polyploidy as mechanism of speciation in plants

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SEMINAR

SEMINARSKI RAD

Marko Sesartić
Undergraduate study of Molecular Biology
(Preddiplomski studij Molekularne biologije)
Mentor: Prof. dr. sc. Višnja Besendorfer

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1. Introduction

Polyploidization is an evolutionary process where two genomes are combined in one nucleus, it represents one of the most dramatic mutations know to occur. Nevertheless, polyploidy is well tolerated in many groups of eukaryotes. There are numerous examples in the plant kingdom, where chromosome fusions/fissions and rearrangements occurred following speciation [1]. Polyploidization is responsible for emergence of genotypic plasticity, providing polyploids with the ability to tolerate genomic variations better then their diploid ancestors. Most species of flowering plants have descended from ancestors who doubled their genomes, either trough autopolyploid or alloploidy [2]. Because ancient WGDs (Whole-genome duplications) in plants give rise to particularly species-rich groups, some have argued that polyploidy is not an evolutionary dead end but that it provides novel opportunities for evolutionary success.

To understand the origins of novel structure/function we need to answer two questions: what is the genetic source of the structure? And how the new structure became adapted to its function? Polyploidization is the increase in genome size caused by the inheritance of an additional set/sets of chromosomes. The duplicated set of chromosomes may originate from the same closely related species (autopolyploid) or from the hybridization of two different species (allopolyploid) [3].

1.1. Competitive advantage of polyploids

Genome duplication can reduce the risk of extinction through several means: by functional redundancy, mutational robustness, and increased rates of evolution and adaptation [3]. The most compelling evidence that genome duplication might help in avoiding extinction comes from flowering plants – legumes, cereals, Solanaceae – who independently underwent WGD ~60-70 mya. Alternatively, in a more neutral scenario, it could be assumed that environmental stress leads to increased incidence of polyploid formation: for instance, trough formation of unreduced, 2n gametes. In this case the cataclysmic events of the last mass extinction ~65 mya could have increased the established of polyploid lineages by chance. However, it is unclear whether such an increase alone could explain the extent to which polyploid plants replaced or overshadowed their diploid relatives [2]. In the adaptive scenario heterotic effects and

rapid genomic and epigenetic changes underlay the ability of polyploids to quickly adapt to more extreme environments. In allopolyploids and autopolyploids, increased heterozygosity can lead to increased variation in gene expression [4]. Transgressive segregation in polyploids might serve as pre-adaptation for survival in habitats not accessible to their diploid parent species [3].

In summery, increased phenotypic variability and heterotic effects have the potential to enable polyploids to survive environmental conditions that do not favour their diploid ancestors [5]. Polyploidy is also know to facilitate self fertilization and the formation of asexually reproducing species, which might be a selective advantage when sexual mates are scare.

1.2 Increased species diversity

Genome duplication is often followed by increased rates of evolution and directional selection on some genes [3]. Although a link between any specific genome duplication event and increased species diversity remains correlational rather than causal, several mechanisms might explain how gene duplication facilitates the formation of new species. Relatively quickly thereafter lineages diverge, which then continue to exhibit increased rates of evolution compared to nonduplicated lineages [3]. This corresponds to a postduplication “window of evolvability” due to relaxed constraint that has been previously postulated [3]. Finally, positive Darwinian selection on duplicate genes can be responsible for functional divergence and innovation. The evolutionary contribution of structural alterations to genome depends on their ability to persist. Although the probability of a gene duplication is low, the half-life of gene duplication is very long – over a million generations [6]. Lynch [7] proposed that the loss of different copies of duplicated genes in separated populations might genetically isolate those populations. Divergent evolution of thousands of genes and regulatory RNAs could therefor facilitate speciation. Scannell [8] showed that subsequent loss of duplicated genes after WGD, can lead species divergence. Different species have lost members of a duplicated gene pairs, so that 4-7% of single copy genes compered between any two species are not orthologous. This pattern of gene loss provides a strong evidence for speciation through a version of Bateson-Dobzhansky-Muller mechanism, in which the loss of alternative copies of duplicated genes leads to reproductive isolation.

The Bateson-Dobzhansky-Muller Model is a model of the evolution of genetic incompatibility. The model attempts to explain how incompatibilities between closely related species develop without either of them going through an adaptive valley. In its simplest form the model shows that changes in at least two loci are required to cause hybrid incompatibility, or at least a decrease in fitness between individuals from two ancestrally identical, but allopatric populations. This is based on the idea that a new allele which has arisen at one locus in one population should not cause decreased fitness when placed into the identical (except for one allele) genetic background of the second population. Therefore another allele at a second locus which is incompatible with the first must have arisen [9].

Other neutral scenarios might also promote speciation. One example would be a case in which both copies of a gene that has multiple functions – it's expressed in different stages of development – are retained in different populations after a duplication event. Should the two populations became geographically isolated, the two genes in each population could subfunctionalize and the orthologoues in different populations might evolve different functions. The partitioning of ancestral subfunctions between gene copies arising from this duplication could have contributed to the genetic isolation of populations, to lineage-specific diversification of development programs, and ultimately to phenotypic variation [10].

There seams to be a correlation between WGDs in plants and increased rates of speciation and divergence. Solitis [11] reported strong correlation between diversification rates and polyploidy following recent WGDs in many plant lineages. For instance, the WGD in the Poaceae lineage possibly coinciding with the origin and divergence of the core Poaceae – a large clade containing ~10,000 species. Whole-genome duplications have also been reported for Brassiceaceae (3,700 species), Asteraceae (23,000 species), Fabaceae (19,400 species) and Solanaceae (>3,400 species); also the rate of diversification in these families is high compered to other families in the same order [2].

1.3 Evolutionary innovations

In the longer run, polyploidy may pave the way for evolutionary innovations of elaborations of existing morphological structures radiation into fundamentally different regions of phenotypic space – a multi-dimensional continuum of all possible phenotypes. One of the prerequisites for developing more complex systems is an increase in number of gene regulators [12]. Intriguingly, genome duplications are the preferred way to accomplish such and increase. Genes retained after WGD event are not distributed equally among Gene Ontology, which indicates a non-random process of gene loss. Genes involved in signal transduction and transcription have been preferentially retained, and those involved in DNA repair have been preferentially lost [13]. Moreover, these regulatory gene classes cannot be easily expanded trough single-gene duplications, which highlights the importance of genome duplications in expanding the regulatory gene repertoire. Maere [14] developed an evolutionary model that simulates the duplication dynamics of genes, considering genome-wide duplication events and a continuous mode of gene duplication. Modelling the evolution of different functional categories of genes assesses the importance of duplication events for gene families involved in specific functions or processes. By applying the model to *Arabidopsis* genome, they report a striking difference of gene loss for large-scale and small-scale duplications; further more they also report highly biased gene loss and retention towards certain functional classes. Both under-retention of regulators after single genome duplication and their over-retention after genome duplications can be explained by dosage balance effects. Recent evidence suggests that a major contributor to this balance is the behaviour of molecular complexes that function in various regulatory processes affecting gene expression [15]. Study of individual gene families also points out the importance of genome duplication in expanding of regulatory repertoire of an organism. In plants, important development regulators, such as AUX/IAA family of auxin response regulators and certain MADS-box transcription factors subfamilies, seem to have expanded mainly trough genome duplication [16].

Gene duplication provides raw material for functional innovation. Although it's unclear whether or not polyploidy caused the evolution of the defining innovations in plants. Rather then facilitate the innovation from scratch, the power of

genome duplication may be their ability to perfect primitive versions or precursor of innovative features and fully exploit their potential [17]. It is plausible that an increase in regulatory gene complexity driven by WGD would tend to increase the potential of an organism to become more complex. But the fact that genome duplications provide the raw material for increased complexity dose not imply they always lead to such a result.

In this respect, the duplicated genome, although may not immediately useful, could be regarded as a genomic spandrel that occasionally might have been used for adaptive or complexity-increasing evolution [18]. An increased rate of speciation after polyploidy could have facilitated this process by providing a lineage with more opportunities to sample phenotype space. If enough species roam the fitness landscape, for some species further changes are likely to become adaptive or previous changes may be co-opted for a novel purpose. Donaghue [19] argued against a link between genome duplications and increased complexity based on the observation that when extinct lineages are taking into account, there are no bursts in morphological innovation or jumps in complexity post-WGD. However, morphological evolution after WGD dose not need to be saltational – sudden change from one generation to the next. Genome duplication merely enhances the diversification potential of a lineage; the diversification process is likely to take time and spawn intermediate forms that are likely to go extinct because they are later outcompeted by more derived relatives [20]. As with species, a better indicator of diversifying force of WGDs is obtained by comparing morphological innovations in WGD clades with their non-WGD sister clades. Phylogenetic placement of WGD events following the last mass extinction is to uncertain to allow accurate identification of WGD clades and non-WGD sister clades, therefore an assessment between correlation and causation between the occurrence of WDGs and morphological innovation is difficult.

2. Conclusion

Darwin famously wrote: “If it could be demonstrated that any complex organ existed, which could not be possibly formed have been formed by numerous, successive, slight modifications, my theory would absolutely brake down”. Darwin’s larger insight was that even “new” features created by evolution bear the marks of their ancestry, and the key role of co-option in creating new molecular functions exemplifies this principle [17]. In line with this hypothesis the gene duplication discussed here make up only a small piece of the story of an organisms adaptation to its environment. Achieving compressive answer to the two question from the beginning will require polling of insights form different fields.

Although the relative importance of the various processes preserving gene duplicates is under debate, everybody agrees that duplicated genes – whether produced by polyploidization or by single gene duplication – take a variety of new function over the long term. Is there then strong evidence that evolution has been facilitated by polyploidization? It is hard to point out a single set of duplicated genes, as proof that the same morphological evolution wouldn’t occur with out polyploidization. Facing the same selective pressure but without the polyploidization, it is likely that such hypothetical ancestors would undergo similar evolution via other means (evolution of regulatory elements, alternative splicing etc.) [6]. Arguably the greatest consequence of polyploidy is an increase in attainable phenotype space. But the potential phenotypic advantage provided by WGDs is less useful when there are no now niches in which newly developed phenotypes are advantageous. In stable ecosystems, newly formed polyploids are probably not able to compete with highly adapted occupants of existing niches, including their diploid ancestors [6]. Therefore, it’s conceivable that the availability of new ecological niches trough severely perturbed ecosystem – following mass extinctions – could be single most important determinant for the survival and long term evolutionary success of WGD. However, new niches may also be formed through biotic evolution. Arens [21] argues that angiosperms didn’t rise to ecological dominance by filing niches that became available after mass extinction, but rather by filling niches that were largely unpopulated because the necessary phenotypic characteristic had not yet been developed.

It may prove difficult to determinate heather polyploidy enabled organisms to survive extinctions or whether polyploidy facilitated evolutionary transitions and increased biological complexity. Further sequencing of more genomes and development of tool that are able to detect and correctly date ancient WGD may unveil the correlation between polyploidy and evolutionary changes that are currently unknown.

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4. Summary

Polyploidization is an evolutionary process where two genomes are combined in one nucleus, it represents one of the most dramatic mutations know to occur. Nevertheless, polyploidy is well tolerated in many groups of eukaryotes. Genome duplication can reduce the risk of extinction through several means: by functional redundancy, mutational robustness, and increased rates of evolution and adaptation. Genome duplication is often followed by increased rates of evolution and directional selection on some genes. Although a link between any specific genome duplication event and increased species diversity remains correlational rather than causal. It may prove difficult to determinate whether polyploidy enabled organisms to survive extinctions or whether polyploidy facilitated evolutionary transitions and increased biological complexity.

5. Sažetak

Poliploidizacija je evolucijski proces u kom se dva genom spajaju u jednu jezgru, ovo predstavlja jednu od najdramatičnijih mutacija. Bezobzira, poliploidija je dobro toleriranu u mnogim grupama eukariota. Duplikacije genoma mogu smanjiti rizik izumiranje kroz nekoliko mehanizama: funkcionalna redundancija, povećana otpornost na mutacije, i povećana stopa evolucije i adaptacije. Duplikacije genoma je često pračena povećanom stopom evolucije i selekcije određenih gena. Usporkos svemu veza između bilo koje duplikacije genoma i povećane raznolikosti vrsta ostaje korelacijska a ne uzročna. Vjerovatno će biti teško dokazati da li poliploidija omogućava organizmima preživljenje izumiranja ili poliploidija ubrzava evoluciju i povećava biološku kompleksnost.