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Rose's Thesis...

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Last edited: November 18 2019

4 Contents

5	1 Introduction	3
6	1.1 The Role of Genome-Doubling in Plant Diversification	4
7	1.2 Benefits	6
8	1.3 Costs	6
9	1.4 How do these mechanisms link?	7
10	1.5 Aims	7
11	References	8

Chapter 1

Introduction

Would some images of genome duplication be helpful?

Whole-genome duplication (polyploidisation) is an extreme mutation which occurs due to errors during cell division; such as non-disjunction where sister chromatids are not separated during anaphase. Failed disjunction leads to all the genetic material ending up in one of the two daughter cells; essentially doubling the genome within that cell. When this occurs during gametogenesis, it can lead to whole organisms with more than two complete sets of chromosomes (polyploids). According to ?, the estimated rate of polyploidisation is one order of magnitude higher than that of standard mutation (10^{-5}). In humans, it coincides with around 5% of miscarriages (Creasy et al. 1976). Moreover, while there have been some live birth cases of triploid human infants, they usually die within hours. In irregular cases, they survive for several months, but none have survived more than six years (Hashimoto et al. 2018).

26 Newer data for miscarriage rate?

27 No polyploid mammal species has ever been found (Svartman et al. 2005). The same
28 is also usually true for birds (?). However, while in these taxa, genome-doubling is not
29 well tolerated, for amphibians, fish, fungi, reptiles, and plants, the story is quite different.
30 Amongst these, polyploidy is exceptionally well tolerated in plants. Jiao et al. (2011) found
31 that genome-doubling is a ubiquitous feature in the lineages of almost all flowering plants
32 (angiosperms), and Wood et al. (2009) also found that it has occurred even more recently
33 (post genera formation) for 35% of all vascular plants. Plant surveys of natural populations
34 over the last century further support this, reporting mean polyploid frequencies of 25% (?).

35 **1.1 The Role of Genome-Doubling in Plant Diversifica-** 36 **tion**

37 Genome-doubling is thought to be fundamental in the diversification of plant species; hav-
38 ing been found to coincide with 15% of angiosperm speciation events, and 31% in ferns
39 (Wood et al. 2009). Furthermore, Jiao et al. (2011) showed that whole-genome duplication
40 could even be linked with major biological innovations that have lead to diversification of
41 the plant phyla. Their phylogenetic analysis brought to light two ancient groups of dupli-
42 cation events: the first grouping of duplications (around 319 millennia ago) influenced the
43 diversification of regulatory genes that were integral to seed development and the more
44 recent grouping (around 192 millennia ago) affected genes that enabled flower develop-
45 ment. The authors appropriately conclude that these events must have played a significant

46 role in the rise of seed plants, as well as in the later diversification of angiosperms. Seeds
47 and flowers were both crucial innovations that allowed plants to transition from an aquatic
48 life-cycle to a terrestrial one by removing their dependence on water for reproduction —
49 an event, broadly considered as one of the most significant in the history of our planet.

50 The full story, however, is not quite that clear-cut. According to Mayrose et al. (2011),
51 genome-doubling significantly increases speciation rates of diploids, but new polyploid
52 lines do not further speciate by that same mechanism. So, polyploid speciation rates are
53 far smaller in comparison. Furthermore, they found polyploid extinction rates to be far
54 higher than those of diploids because polyploids are clumped around the tips of phyloge-
55 nies. This Likelihood-based analysis of vascular plants provided clear, and quantitative,
56 support for the traditionally popular view that polyploidy most often leads to evolutionary
57 dead ends.

58 First proposed by? Stebbins 71?

59 So, what is it then that allows this extreme mutation to succeed and become fixated? What
60 scenarios and mechanisms combine to allow rare polyploid lines to succeed? Are the ma-
61 jority of the polyploids we see in natural populations doomed to extinction? Furthermore,
62 can we predict which conditions will lead to their survival?

63 Despite having been studied for over a century, the factors that drive the success of poly-
64 ploid establishment in the face of high extinction rates are still unclear. Arrigo and Barker
65 (2012) conclude that polyploids tend to become extinct at the establishment phase due to
66 reproductive disadvantages such as triploid sterility, or limited mate-choice; the latter oc-
67 ccurring via diploid pollen-swamping, or delayed flowering. On the other hand, polyploids

are frequently linked with distinct traits such as “gigas effects”, which describes their increased size and plant organs. Additionally, benefits of polyploidy can include enhanced capabilities for buffering of deleterious mutation (due to increased heterozygosity), hybrid vigour (heterosis), and — somewhat contentiously — a reversal of selfing inhibition (Woodhouse et al. 2009; Ramsey and Ramsey 2014). These traits are thought to overcome the reproductive disadvantages of polyploidy and, in those rare cases, make this mutation key to the invasive and adaptive potential of plants. Let us examine these mechanisms in more detail:

1.2 Benefits

Gigas-effects

Genetic buffering

Hybrid Vigour

1.3 Costs

Are they realistic? How do they work? Why/when are they important? Consider the conditions.

83 **Diploid Pollen-Swamping**

84 **Delayed Flowering**

85 **Triploid Sterility**

86 So what are the core mechanisms?

87 Or the most suspicious?

88 **1.4 How do these mechanisms link?**

89 Does limited mate-choice set the scene for the evolution of selfing vs out-
90 crossing?

91 Delayed flowering (cost) is associated with gigas-effects of increased size
92 (benefit). TRADE-OFF.

93 **1.5 Aims**

94 Based on all this pondering - what am I actually going to examine?

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