**Polyploidapalooza**

**Module 3: Polyploid Evolution**

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**Abstract**

Polyploidy plays an important role in the evolution of life but is often overlooked in undergraduate classrooms. In this series of modules, we explore how polyploid species are formed, how they evolve and adapt, and their economic and conservation importance. Module 3 explores how polyploid animals evolve, including what happens to duplicate genomes after polyploidization, the effects of polyploidization on rapid adaptation, and the role that whole-genome duplications have played in vertebrate evolution.

**Objectives**

* Examine the fates of sub-genomes after duplication including neofunctionalization, subfunctionalization, and pseudogenization
* Assess the process of rediploidization and the benefits of genome duplication followed by a return to a diploid system
* Explore a case study related to polyploidization, neofunctionalization, and rapid adaptation to new environments
* Explore a well-known example of genome duplication and rediploidization at the base of the vertebrate phylogeny

**Logistics**

Department: Biology

Level: Upper undergraduate

Prep time: 0 hours

Class time: 1-2 hours

**Pre-module preparation**

List of terms - come back to these terms if you need to while reading the text.

**Sub-genome**—one full genome that is part of a duplicated polyploid genome

**Neofunctionalization**—a process in which a gene acquires an entirely new function after a gene duplication event

**Subfunctionalization**—a process in which two duplicate genes each take on a subset of the functionality of the original gene

**Genetic silencing**—the regulation of a gene that prevents it from functioning

**Pseudogenization**—the accumulation of mutations in a gene that prevents it from functioning

**Rediploidization**—bivalent (diploid-like) pairing during meiosis within a polyploid species made possible by genetic divergence between sub-genomes

**Paralogues**—copies of the same gene that result from a duplication event. Compare t**o orthologs** — gene copies that are similar because of common ancestry

**Introduction**

Because of the meiotic challenges associated with whole-genome duplication (Module 2), the evolutionary fate of polyploids has been a topic of debate for many years, with many researchers believing that these species are evolutionary dead-ends. However, more recent studies suggest that major evolutionary lineages, including all flowering plants and all vertebrate animals, are descended from polyploid ancestors that later underwent **rediploidization** to form the genetic systems that we see today. Further research suggests that polyploidization may enable organisms to adapt more quickly to new environments because they have additional genetic material that can be repurposed for new functions (**neofunctionalization)**. In this module, we will assess some of the benefits and challenges of polyploid evolution and explore several case studies related to the role that polyploidy can play in both deep and recent evolutionary history. 

**Benefits and challenges of genome duplication**

Having an extra copy of the entire genome can present interesting challenges for a new polyploid species. These challenges include unstable, multivalent pairing during meiosis (covered in Module 2), as well as a huge increase in gene expression levels. Because of these challenges, polyploid species formation is often associated with many observable changes to the genome. These include rearrangements of the chromosomes, increased activity of transposable elements, and epigenetic changes to gene expression. If the species can make it through these initial steps towards genome stability, there are actually significant benefits to having a large amount of extra DNA. Polyploids often have high levels of heterozygosity compared to diploids. This may give them protection against dangerous mutations and also allow for more rapid responses to natural selection.

**The fates of the subgenomes**

Because polyploids have extra copies of their entire genome (each copy is called a **sub-genome**), they often have to develop mechanisms to regulate gene expression levels. Duplicate genes in these sub-genomes can either be turned off (**silenced**) to avoid creating too much gene product, adapted to have an entirely new function (**neofunctionalization**), or adapted to take over part of the function of the same gene in the other sub-genome (**sub-functionalization**). These changes in gene function may allow polyploids to adapt to different environments than those occupied by their parent species. Eventually, through evolutionary time, the sub-genomes diverge enough that they no longer form multivalent pairs during meiosis, and instead re-adopt the stable bivalent pairing normally seen in diploids, a process called rediploidization.

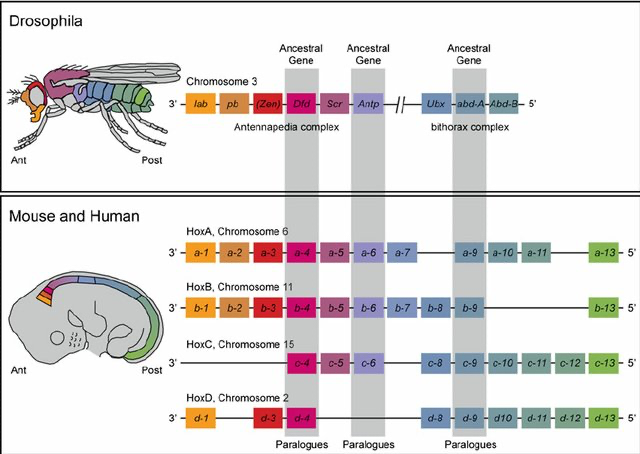
Please answer the following three (3) questions from the reading above:

1. What are the three most common fates of duplicated genes/sub-genomes?
2. What is the difference between neofunctionalization and subfunctionalization?
3. What is the benefit of rediploidization for a polyploid organism?

**Case study 1: Polyploidization in the history of vertebrates**

Scientists once hypothesized that whole-genome duplication was an evolutionary dead-end that was rare in the natural world. However, we now know that this process has played an important role in the evolution of life. For example, there is strong genetic evidence for two whole-genome duplication events followed by rediploidizations in the evolutionary history of vertebrates. These duplications were first discovered in the Hox genes. Hox genes specify the organization of an animal’s body. They specify where the head, trunk/body, and tail will develop as well as structures such as limbs, wings, and antennae. Interestingly, all animals—from worms to mammals—share a similar set of Hox genes but have them in different numbers. For example, fruit flies have one set of Hox genes, whereas humans, mice, lizards, and birds have four, and most fish have eight. The variation in the number of Hox gene copies across animals is due to whole-genome duplication events that occurred early in the evolutionary history of vertebrates. There were two duplications in the lineage leading to living vertebrates and a third duplication during the evolution of actinopterygian fish.

Below is a diagram from Pang & Thompson (2011) that compares the Hox genes of fruit flies to those of mammals. In this figure, the colors indicate corresponding genes as well as the part of the body where development is controlled by that gene. Duplicated Hox genes in the mouse and human are labeled HoxA through HoxD.

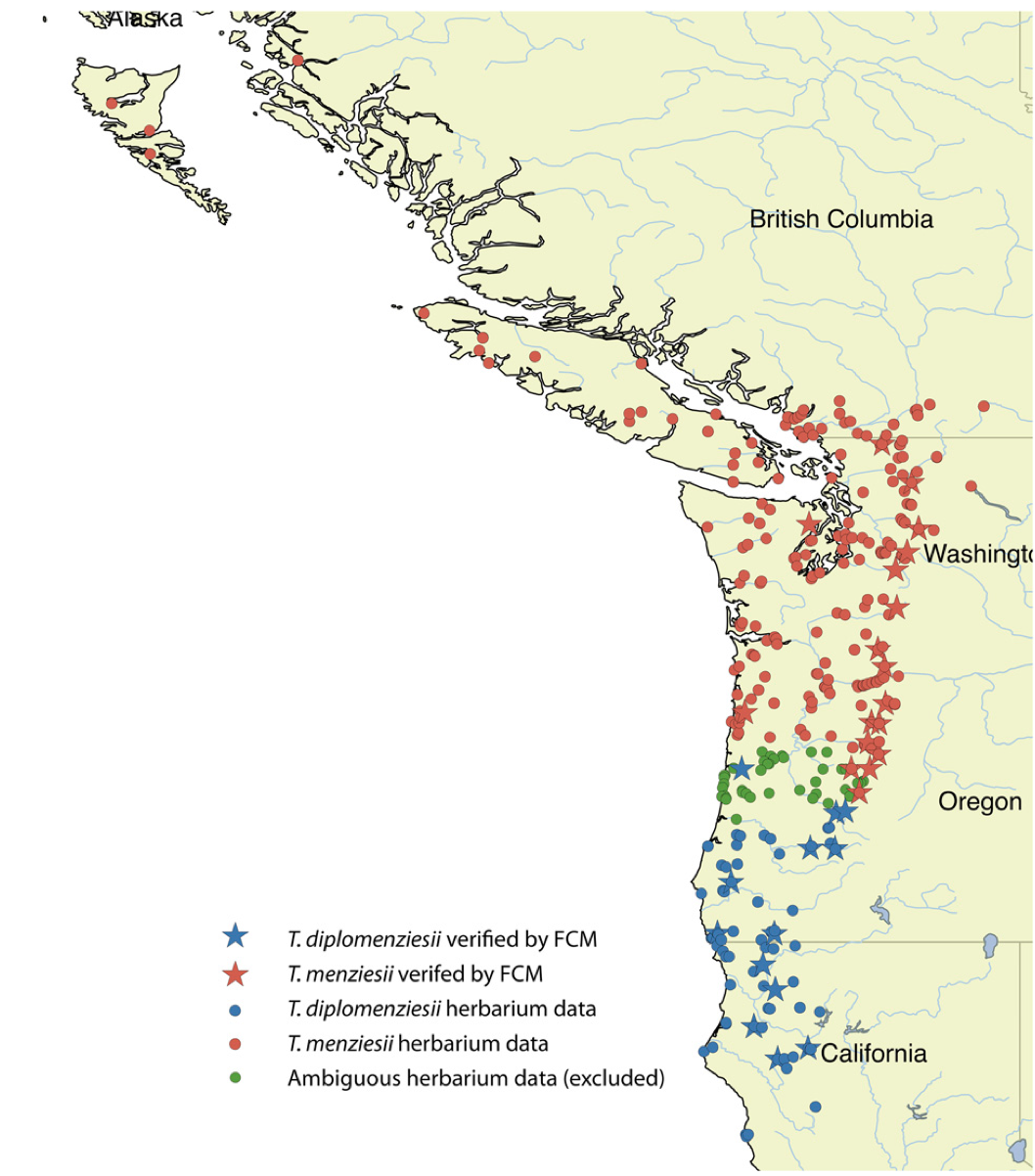


Using the diagram above, please answer the three (3) questions below:

1. What similarities do you notice between the regions on the body controlled by each Hox gene on the fly and on the mammal? Do you notice any differences? What does this say about the conservation of hox gene functions?
2. The diagram indicates an ancestral gene and “**paralogues**.” Paralogues are copies of the same gene that are a result of a duplication event. What is the maximum number of paralogues seen in the mammal genome? How many duplication events were needed to create this number of paralogues?
3. Even though there are four copies of the Hox genes in humans and mice, these species are still diploid. What is the name of the process that causes duplicated genomes to return to a diploid system? What are the benefits of diploidy?

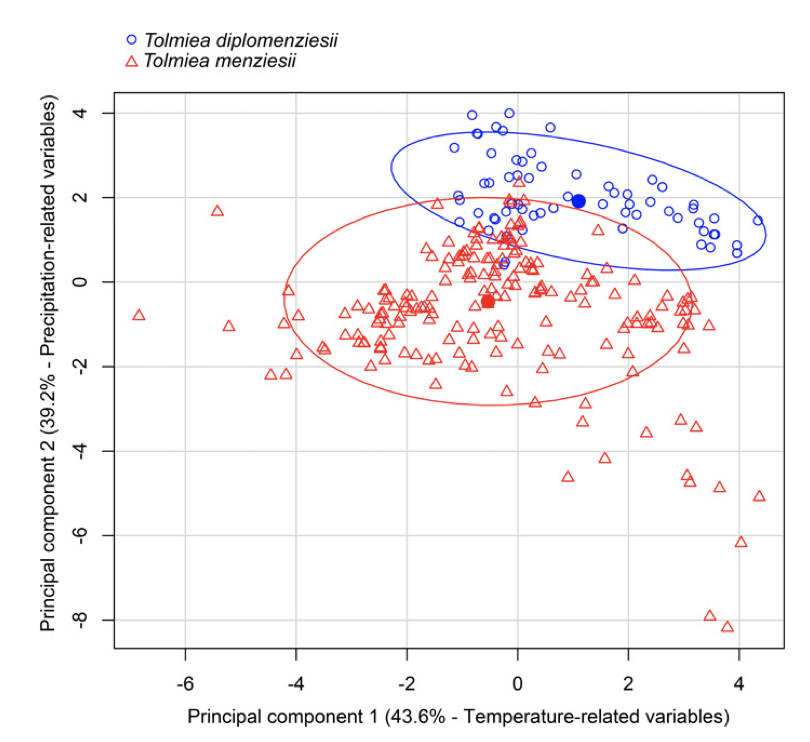
Interesting fact: You might notice that some Hox genes above have only two or three copies. This is because some copies of that gene have been lost to **pseudogenization** and no longer function.

**Case study 2: polyploidization and rapid adaptation**



While our first case study focused on the effects of ancient genome duplications, case study 2 will focus on the evolutionary advantages of recent polyploidization events. Here, we will look at the role that polyploidization plays in rapid adaptation to new environments. New polyploid species often overlap geographically with the parent species they derive from. This geographic proximity can lead to competition for resources and the need to adapt to new environments or niches. Because polyploids often have high levels of genetic variation, it is likely easier for them to adapt quickly to novel conditions. Additionally, the increased number of duplicated genes provides protection from dangerous recessive mutations due to the high levels of heterozygosity.

We will focus on the genus *Tolmeia*, a flowering plant species that is found in western North America (also discussed in Module 1). There are two species in *Tolmiea*: *T. diplomenziesii* (diploid)and *T. menziesii* (autotetraploid). These two species look identical but inhabit different habitats along the west coast, as you can see in the figure to the right from Visger et al. (2016). Geographically, we can see that the tetraploid *Tolmiea menziesii* (red dots and stars) inhabits regions far to the north of its parent species *Tolmiea diplomenziesii* (blue dots and stars)*.*

The difference in geography between the two species of *Tolmiea* is caused by a divergence in their niches (i.e., the conditions where they are able to survive and reproduce). As you can see from the below figure also from Visger et al. (2016), the locations where each species is found differ in temperature and precipitation (rainfall). There is a large difference in the amount of precipitation that each species can tolerate, with *T.*  *menziesii* being far more drought-resistant than its diploid parent species. This pattern is not uncommon between polyploids and their parental species. 

Please answer the following three (3) questions related to case study 2:

1. What challenges do newly formed polyploid species face that cause them to need to adapt to an environment where their parent species do not live?
2. Why is it believed that it may be easier for a polyploid species to adapt to a new environment than a non-polyploid species?
3. Choose one of the duplicate gene fates mentioned above: neofunctionalization, sub-functionalization, or pseudogenization. How do you think it can assist a polyploid species as it adapts to a new environment?

**Module summary question:**

At one time scientists believed that polyploidization was an evolutionary dead end. Based on your understanding of polyploid evolution in this module, what do you believe? What are some of the challenges and benefits of evolving as a polyploid?

**Acknowledgments**

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**References**

Pang, D., & Thompson, D. N. (2011). Embryology and bony malformations of the craniovertebral junction. *Child's Nervous System*, *27*(4), 523-564.

Visger, C. J., Germain‐Aubrey, C. C., Patel, M., Sessa, E. B., Soltis, P. S., & Soltis, D. E. (2016). Niche divergence between diploid and autotetraploid Tolmiea. *American Journal of Botany*, *103*(8), 1396-1406.

Sugar cane image

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