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Turning four into two: How duplicated genomes become diploid again

Genome duplication probably gave biodiversity a decisive evolutionary boost. A Chinese-German research team led by Axel Meyer from the University of Konstanz has now investigated the early phases of the process known as re-diploidization. The results show that the fusion of chromosome sets is asynchronous.

One chromosome from the mother, one chromosome from the father: For us humans and the vast majority of animal species, having a double set of chromosomes is the standard. Two complete sets of chromosomes in an organism’s cells is what we call "diploid". However, this is not always the case. It is rare, but possible for organisms to contain more than two sets of chromosomes in each cell’s nucleus, typically by doubling the complete genetic material (through autopolyploidization of hybridization within a species). Cases involving fourfold, sixfold or even twentyfold sets of chromosomes, such as those discovered and sequenced in the current study, are, however, exceedingly rare. These "polyploids" are quite common in plants but rarely occur in animals – usually only in certain groups of fish.

Over the last fifty years, scientists have increasingly recognized that the duplication of chromosome sets had played a major role in the early phases of evolution. They believe that this multiplication led to an explosive expansion of genetic variation and evolutionary innovation, giving newly emerged species a powerful boost toward new body plans and adaptations. However, such "polyploid" species usually do not retain the multiplied set of chromosomes indefinitely. Typically, polyploids gradually return to a more stable, diploid mode of inheritance ("re-diploidization"): over generations, they reduce the "surplus" chromosome sets to eventually arrive at a stable state with a "mere" double set of chromosomes once again. As the lab team around evolutionary biologist Axel Meyer from Konstanz had been able to show already 25 years ago, this process occurred in the common ancestor of all modern fishes (about 27,000 species) around 230 million years ago and is considered a key factor behind the evolutionary success of fish biodiversity

Exploring the early stages of evolution – with snow carp

It would be highly interesting for evolutionary research to understand these genetic processes of multiplication and subsequent re-diploidization in more detail as it would give us deeper insights into the early phases of the emergence of new species and their adaptations. However, because so much time has passed since the polyploidization of fishes, this can no longer be traced easily. Studying these processes directly is challenging, as recent polyploids are rare among living animals.

A Chinese-German research team led by Axel Meyer (University of Konstanz) has now been able to reconstruct the genetic processes of re-diploidization through extensive comparative genomic studies. Their findings were made with snow carps. From an evolutionary perspective, these fish are relatively young: they emerged only around 30 million years ago in the Himalayas – hence their name. They inhabit extreme altitudes of up to 5,000 metres – a world record – and underwent a duplication of their chromosome set comparatively recently in evolutionary terms. For Meyer and his team, they represented an ideal model for investigating the early phases of re-diploidization, as these fish are still in the midst of the process of re-reducing their chromosome sets and returning to a diploid state.

The first stage: the fusion of chromosomes

There have already been comparable studies on salmon and sturgeon, which also tolerate polyploid genomes. However, none of these studies were able to investigate the early phases of re-diploidization, because their polyploidization already happened a long time ago – and it is precisely these early phases that are highly interesting, but are no longer available for study in these ancient polyploids. "We can now prove that the first stage of re-diploidization is initiated by the fusion of chromosomes", explains Axel Meyer. Chromosomes are first united through fusion, and it is at these focal points in the genome that rediploidization begins, so that over generations fewer chromosome sets are ultimately found in each cell nucleus. This process does not proceed uniformly, but rather in a stepwise fashion, as Meyer and his team show. Initially, in some regions of the genome the chromosomes have already fused to form what are described as ohnolog pairs, while other regions still retain a fourfold set of chromosomes.

Chromosome unification thus begins at individual fusion sites within the genome and then spreads outward from there in stages toward the ends of the chromosomes. "The course of rediploidization thus depends strongly on proximity to these chromosomal fusion sites within the genome", Meyer says. "Identifying the initial regions of the genome in which rediploidization began at the earliest was key in order to uncover this genomic mechanism".

In snow carps, the fusion of chromosome sets therefore proceeds asynchronously: while some regions of the genome rapidly returned to a diploid inheritance, other regions retained a fourfold set of chromosomes for a prolonged period – over many millions of years. Beyond snow carps, it is also assumed that genome duplication and the subsequent reduction to two chromosome sets have profoundly shaped the evolution of vertebrates. Through these findings, we have now gained a deeper insight into the chromosomal mechanism underlying these macroevolutionary crucial events that preceded the major innovations in evolution.

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