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Did sex drive mammal evolution? How one species can become two

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There’s a difference in the sex chromosomes between various mammals, such as the platypus compared to humans. Credit: Flickr/Darren Puttock, CC BY-NC-ND

How new species are created is at the very core of the theory of evolution. The reigning theory is that physically separated populations of one species drift apart gradually.

But changes in chromosomes, particularly sex chromosomes, can interpose drastic barriers to reproduction. Mammals may be a good example. Comparisons of the sex chromosomes of the three major mammal groups show that there were two upheavals of sex chromosomes during mammal evolution.

The first corresponded to the divergence of monotreme mammals (platypus and echidna) from the rest, and the second to the divergence of marsupials from placental mammals (including humans).

In a paper published in *BioEssays*, I propose that drastic sex chromosome changes could have played a direct role in separating our lineage (placental mammals), first from the egg-laying monotremes, then from marsupials.

In humans and other placental mammals, such as mice, dogs and elephants, sex is determined by a pair of chromosomes. Females have two copies of the X while males have a single copy of the X and a small Y that contains the male-determining gene *SRY*.

Other vertebrate animals also have sex chromosomes, but they are different. Birds have an unrelated sex chromosome pair called ZW, and a different sex determining gene called *DMRT1*.

Snakes also have a ZW system, but again it is a different chromosome with different genes. Lizards and turtles, frogs and fish have all sorts of sex chromosomes that are different from the mammal system and from each other.

The rise and fall of sex chromosomes

Sex chromosomes are really weird because of the way they evolved. They start off as ordinary chromosomes, known as autosomes. A new sex gene arises on one member of the pair, defining either a male-determining Y as in humans or a female-determining W as in birds.

The acquisition of a sex factor on one member of the pair is the kiss of death for that chromosome, and it degrades quickly. This explains why only a few active genes remain on the human Y and the bird W.

When old sex chromosomes self-destruct, a new sex gene and sex chromosomes may take over. This is fraught with peril because the interaction of old and new systems of sex determination is likely to cause severe infertility in hybrids.

Rival sex genes may be at war with each other, causing intersexual development, or at least infertility. For instance, what will be the sex of a hybrid that has both a male-determining Y and a female-determining W?

Added to this are problems with gene dosage because the degenerate Y and the W have few genes. If an XY male mates with a ZW female, most of the progeny will be short of genes. There may also be problems with gene dosage because genes on the X and the Z are used to working harder to compensate for their single dosage.

Rearrangement of sex chromosomes with autosomes also causes severe infertility because half the reproductive cells of a hybrid will have too many, or too few, copies of the fused chromosome.

Such hybrid infertility poses a reproductive barrier between populations with the new and the old sex system. So could such barriers drive apart populations to form distinct species?

Reproductive barriers and new species

The idea that chromosome change could drive the formation of new species was popular 50 years ago.

But it was thoroughly dismissed by evolutionary geneticists in favour of the idea that speciation, the formation of new and distinct species, must occur in populations already separated by a physical barrier such as a river or mountains, or behaviour such as mating time, and occupied different environments.

Small mutations would accumulate slowly and the two populations would be selected for different traits. Eventually they would become so different that they could no longer mate with each other and would form two species. This allopatric speciation relied on external factors.

The alternate view, that sympatric speciation can happen within a population because of intrinsic genome changes, fell out of favour. Partly this was because it is hard to demonstrate speciation of populations sharing the same environment, the argument always being that the environment could be subtly different.

The other problem was imagining how a major chromosome change that occurred in one animal could spread to a whole population. Sex chromosome change is especially drastic because it directly affects reproduction. But our comparisons show that sex chromosomes have undergone dramatic changes throughout vertebrate evolution.

It is important to examine closely examples of evolutionary divergence that were accompanied by drastic sex chromosome change. Strangely, mammals may offer us a window into this evolutionary past. Their sex chromosomes are extremely stable, yet they have undergone rare dramatic changes, each of which lines up near when one lineage became two.

Sex chromosome change and mammal divergence

Placental mammals all share essentially the same XY. Marsupials, too, have XY chromosomes, but they are smaller; genes on the top bit of human X are on autosomes in marsupials.

Comparisons outside mammals shows that this bit was fused to ancient marsupial-like X and Y chromosomes before the different lines of placental mammals separated 105-million years ago.

Monotreme mammals (platypus and echidna) have bizarre multiple X and Y chromosomes. Surprisingly, comparing the genes they bear showed that they are completely unrelated to the XY of humans and marsupials. In fact, platypus sex chromosomes are related to bird sex chromosomes.

The human XY pair is represented by an ordinary chromosome in platypus. So our XY and *SRY* are quite young because they must have evolved after monotremes diverged from our lineage 190-million years ago.

Sex chromosome change has occurred very rarely in mammals, so it seems significant that each change corresponds to a major divergence. That's why I propose that sex chromosome turnover separated monotremes from the rest of the mammals, and sex chromosome fusion occurred later to separate our lineage from marsupials.

Strengthening the argument that sex chromosome turnover begets speciation is evidence of a new round of sex chromosome change and speciation.

In Japan and eastern Europe, species in two rodent lineages have completely eliminated the Y chromosome and replaced *SRY* with a different gene on a different chromosome. In each lineage the Y-less rodents have recently diverged into three species.

What does this mean for our own lineage? The primate Y seems to be more stable than the rodent Y. But if it continues to degrade at the same rate, it will disappear in about 4.6 million years.

Will it be replaced by some different gene and chromosome? And if so, will this unleash a new round of hominid speciation? We may have to wait another 4.6 million years to find out.

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