

Mathematical glimpse on the Y chromosome degeneration

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Abstract. The Y chromosomes are genetically degenerate and do not recombine with their matching partners X. Non-recombination of XY pairs has been pointed out as the key factor for the degeneration of the Y chromosome. The aim here is to show that there is a mathematical asymmetry in sex chromosomes which leads to the degeneration of Y chromosomes even in the absence of XX and XY recombination. A model for sex-chromosome evolution in a stationary regime is proposed. The consequences of their asymmetry are analyzed and lead us to a couple of conclusions. First, Y chromosome degeneration shows up $\sqrt{2}$ more often than X chromosome degeneration. Second, if nature prohibits female mortalities from being exactly 50%, then Y chromosome degeneration is inevitable.

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Human sex chromosomes share some intriguing features, including the genetic degeneration of Y chromosomes [1–6]. Suppression of XY recombination is known today to be the main factor responsible for the Y chromosome degeneration [7–9]. However, Lobo and Onody [10] have found that this lack of recombination is not the sole factor responsible for the Y degeneration; there is an additional mechanism of degeneration which considers the evolution of sex chromosomes with non-recombining XX and XY pairs. Sex chromosomes embody a very particular asymmetry: while Y chromosomes are forbidden to experience female bodies, the X chromosomes are free to move through both sexes. Here I present a simple mathematical model in which this asymmetry is explored. The impact of this asymmetry on Y chromosome evolution is very powerful [10–12].

Consider a population of sex-chromosomes organisms which has evolved over millions of years and reached a stationary regime where males consist of XY and females of XX pairs of chromosomes. Let us associate with the X and Y chromosomes, the final mortality probabilities x and y , respectively. This mortality probability is exclusively due to the genetic structure of the sex-chromosome organisms. It represents the overall effects of the total number of genes held in the sex chromosomes in the stationary regime. It is true that the survival probability of a biological organism is composed of a complex interrelationship between all sorts and varieties of genes. This simplification in the present model can be justified by the following. First, we are interested in studying the exclusive effects

of XY asymmetry on sex-chromosome evolution. Second, despite the complex interactions among genes in the sex chromosomes, one can assign a single variable (x or y) which represents the sum of the effects resulting from all genes within the chromosome.

Each diploid individual will carry a mortality rate due to their sex chromosomes. Suppose that, on average, non sex chromosomes (autosomes) have the same effect on male and female mortalities. The purpose here is to disregard those autosomes in order to study the exclusive effect of the XY asymmetry on the evolution of the sex chromosomes. Consider that both types of chromosomes — X and Y — will affect male and female organisms equally, that is to say, each chromosome contributes 50% to the individual's survival probability. Therefore, we have, for female and male individuals the following mortalities:

$$p_f = 0.5x + 0.5x = x, \quad p_m = 0.5x + 0.5y. \quad (1)$$

It is important to emphasize the meaning of the aforementioned probabilities. The values x and y represent the contribution from the X and Y chromosomes to the individual's mortality. The values p_f and p_m represent, respectively, the probability of a single female and male to die before sexual maturity has been reached. The idea of assigning the same contribution from X and Y chromosomes is to treat both chromosomes on an equal footing, i.e., not to privilege either type of chromosome. We are dealing with the stationary regime, which means that after millions of years of evolution, mutations and natural selection have led X and Y chromosomes to converge to their respective x and y mortalities.

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The population is divided into two age groups: adults and offspring. Adults are those individuals that have reached sexual maturity. The adults, however, are not necessarily semelparous, i.e., they can breed more than once. Let us consider adults as the sum of all qualified individuals for reproduction in the stationary regime. Note that there can be many ages that qualify for reproduction but we shall consider them pertaining to the same age group for the sake of simplicity. Let N_f and N_m stand for the number of adult females and males, respectively, and N represent the total number of babies. Considering there is $N/2$ offspring from each sex, the number of adult females and males, according to their mortalities p_f and p_m are, respectively:

$$N_f = \frac{N}{2} - \frac{N}{2}p_f = \frac{N}{2} - \frac{N}{2}x, \quad (2)$$

$$N_m = \frac{N}{2} - \frac{N}{2}p_m = \frac{N}{2} - \frac{N}{2} \left(\frac{x+y}{2} \right). \quad (3)$$

Suppose all adult individuals are sexually reproductive and monogamic. The number of fertile individuals, N_R , is therefore given by:

$$\begin{aligned} N_R &= N_m + N_f - N_{unmarried} \\ &= N_m + N_f - \sqrt{(N_m - N_f)^2}. \end{aligned} \quad (4)$$

Note that N_R stands for the total number of fertile individuals and not for the total number of couples. Since each individual consist of a pair of chromosomes and in order not to privilege either type of chromosome, let us allow the fertile individuals to produce two offspring each. It follows that the number of babies equals to:

$$N = 2N_R. \quad (5)$$

Equation (5) imposes the stationary condition, i.e., a stable population, independent of the number of babies, is achieved. Combining equations (2)–(5) we are led to the following:

$$3x + y + \sqrt{(x-y)^2} = 2. \quad (6)$$

Equation (6) provides a dependence between the mortality probabilities x and y due exclusively to the asymmetry present in sex chromosomes, namely, the fact that men possess XY and women XX pairs of chromosomes. This dependence is plotted in Figure 1a. Each and every set of points (x, y) can be interpreted as the converged mortalities of sex chromosomes from different initial configurations of evolution.

We can interpret equation (6) at two levels. First, let us define a measurable quantity that gives us how many times Y chromosome degeneration exceeds X chromosome degeneration by dividing the length of the $y > x$ curve by the length of the $x > y$ curve. By doing so, we can conclude that Y chromosome degeneration occurs $\sqrt{2}$ times more often than X chromosome degeneration, i.e., from all possible initial configurations of evolution Y chromosome degeneration would be predominant. In other words, if nature were to perform many experiments

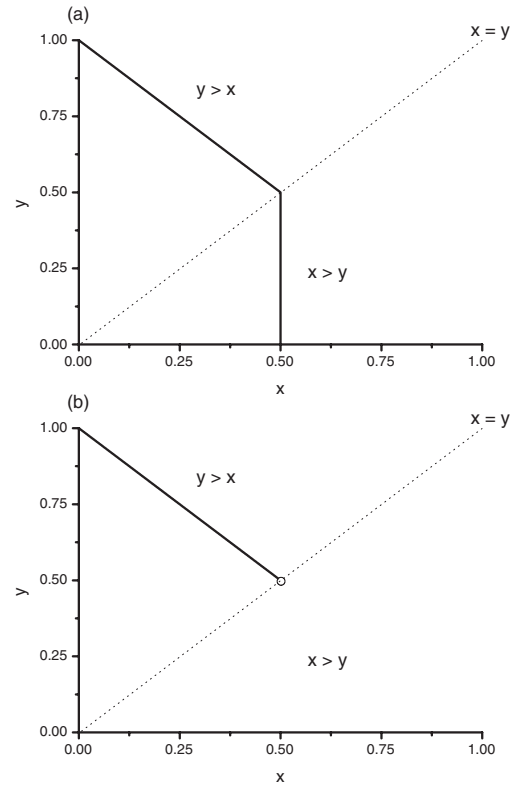


Fig. 1. Mortality probabilities for X and Y chromosomes. (a) The Y chromosome degeneration is predominant by a factor $\sqrt{2}$. (b) Degeneration of the Y chromosome is assured if $x \neq 0.5$.

of sex-chromosome evolution governed by equation (6) then Y chromosome degeneration would prevail most of the time.

The second interpretation considers an additional assumption. Suppose that, when the stationary regime has been reached, the randomness of mutations on the X chromosomes are such that it prohibits female mortalities from being exactly 50%. A careful reading of equation (6) for this special case, where $p_f = x \neq 0.5$, is illustrated in Figure 1b. Each and every set of points in Figure 1b lead us to the following conclusion: the mortality of Y chromosomes is always greater than that of X chromosomes. If nature prohibits females from having a mortality rate of exactly 50%, then Y chromosome degeneration is certain.

The mathematical asymmetry embodied by the sex chromosomes plays a central role for the Y chromosome degeneration. The purpose here is to call attention to this important asymmetry. Suppose, for instance, that at the beginning of sex-chromosome evolution both XX and XY pairs did not recombine. Even so, the mathematical asymmetry of the sex chromosomes would still lead to the degeneration of the Y chromosomes.

The power of this asymmetry is verified by a large amount of experimental evidence and therein lies its strength. In many groups of animals and plants, X and Y chromosomes have evolved genetically and morphologically in an independent fashion [1,13]. Curiously, there is an

almost complete deterioration of genes in the Y chromosome (or W chromosome, in groups with female heterogamety like birds) [9]. In other words, despite the diversity of lineages such as humans, birds, insects and plants, the member of the sex chromosome pair that is forbidden to exist in both sexes degenerates over time, thus losing the majority of its genes and so shrinks in size.

Although the simplicity of the approach used here is evident, it did not favor a priori either type of chromosome or either sex. Both X and Y chromosomes were treated on equal footing. The model studied here gives rise to two points of view: Y chromosome degeneration occurs more often than X chromosome degeneration with respect to different initial configurations of evolution; or Y chromosome degeneration is assured if nature avoids a single mortality rate for females, namely, 50%. The fulfillment of this condition leads Y chromosomes to decisive and spontaneous degeneration. I believe that this spontaneous degeneration mechanism was present in sex-chromosomes evolution since the very first Y chromosome appeared on Earth, depending on whether or not those primeval chromosomes recombined during the first moments of evolution.

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