# Y-chromosome evolution in humans

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The human Y-chromosome has increasingly reduced its size through its evolutionary history, currently being the shortest of the 23 human chromosomes. This raises questions as to whether it will continue to shorten, and if so, down to which size? Did it already stop losing genes, or will it eventually disappear completely?

## Objectives

Without neglecting that the X-chromosome is equally relevant, this study will focus on Y-chromosome.

- ❖ Which are the processes underlying Y-chromosome degeneration?
- ❖ Comparative study of a variety of diploid species

## Methodology

Study based on scientific literature research using *Pubmed* tool (NCBI) and *Google Scholar*.

**Keywords:** *sex-chromosomes, Y-chromosome, evolution* and *degeneration.*

Selection of principal interest papers through abstracts. Mind-blowing diversity. Exercise of synthesis and concretion.

**Sex chromosomes**
Chromosomes that determine the sex of an organism.

**Chromosome degeneration**
Process of gene loss.

**Heterochromatic**
Formed by heterochromatin, a tightly packed form of DNA, typically genetically inactive and containing repetitive sequences and few genes.

**Euchromatic**
Formed by euchromatin, a lightly packed form of DNA, typically rich in gene concentration and genetically active.

**Recombination**
Breaking and rejoining of DNA strands to form a new combination of genetic information.

**Gene conversion**
Non-reciprocal recombination.

**Chromosomal transposition**
Movement of DNA from one location to another.

**Chromosomal inversion**
Rearrangement in which a segment of a chromosome is reversed end to end.

**Proto-sex chromosomes**
New pair of chromosomes that recently acquired a sex-determining function but that otherwise contains identical genes.

**Beneficial mutation**
Mutation that increases the survivorship or fecundity (fitness) of its carrier.

**Deleterious mutation**
Mutation that decreases the survivorship or fecundity (fitness) of its carrier.

**Muller's ratchet**
Irreversible accumulation of deleterious mutations in a non-recombining population.

**Genetic hitchhiking**
Fixation of a deleterious mutation that is linked to a beneficial allele.

**Ruby in the rubbish model**
Elimination of a beneficial mutation that is linked to a deleterious allele.

## Results

### 1 Structural classification and functional aspects of Y-chromosomes

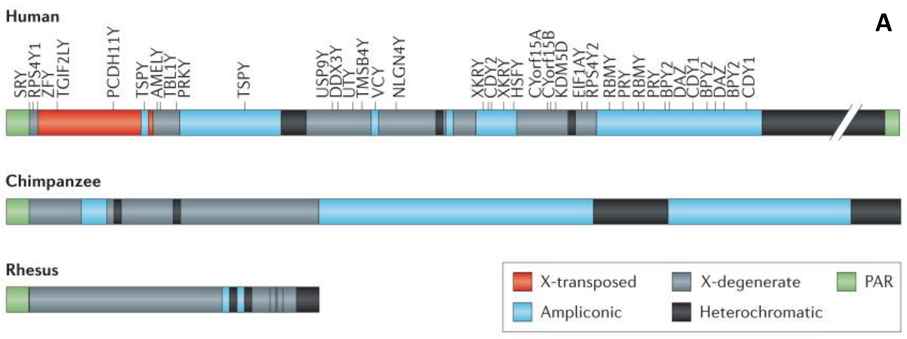


**Figure 1.A** Y-chromosomes in different species (taken from Ref. 1).

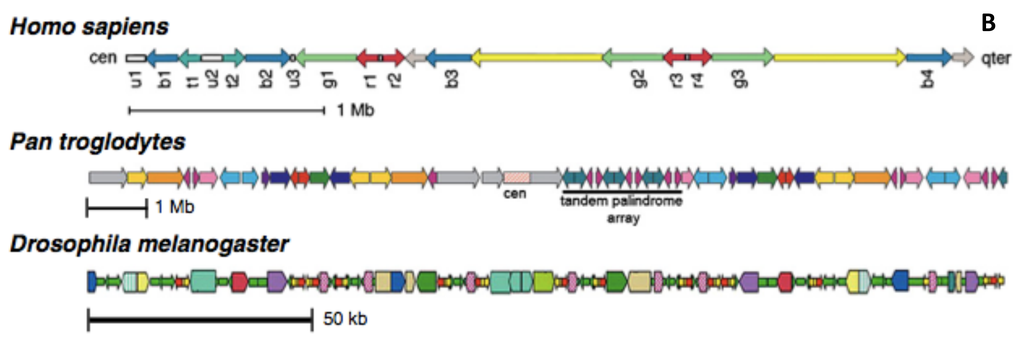


**Figure 1.B** DNA units sharing more than 99% sequence identity (amplicons) are represented by an identical colour code. Gray blocks denote single-copy genes (taken from Ref. 2).

### 2 Formation of human sex chromosomes

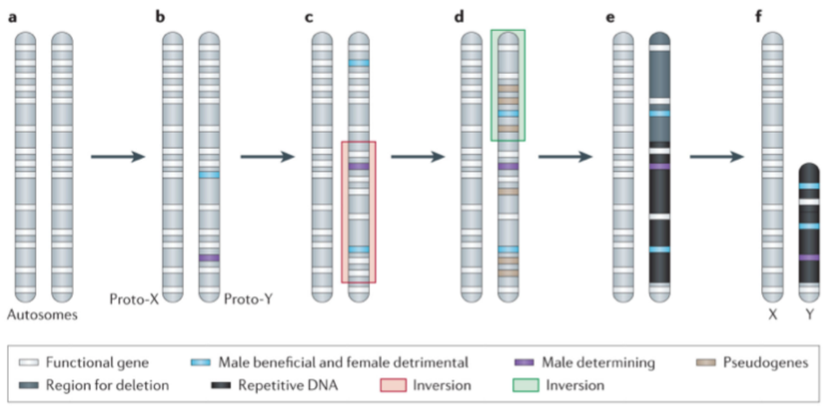


**Figure 2** Graphic representation of the potential path leading to heteromorphic sex chromosomes (taken from Ref. 1).

### 3 Population processes underlying Y-chromosome degeneration

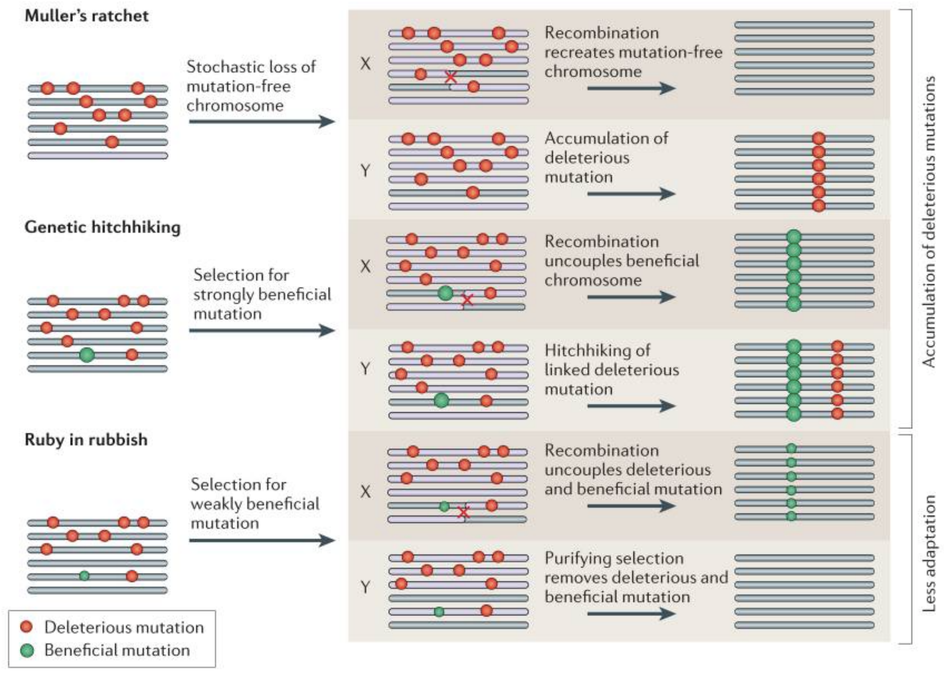


**Figure 3** Gene decay caused by a shutting down of X-Y crossing over can be consequence either of an accumulation of deleterious mutations (Muller's ratchet and genetic hitchhinking) or of a lower rate of adaptation (ruby in the rubbish) (taken from Ref. 1).

### 4 Formation of *Drosophila* neo-sex chromosomes

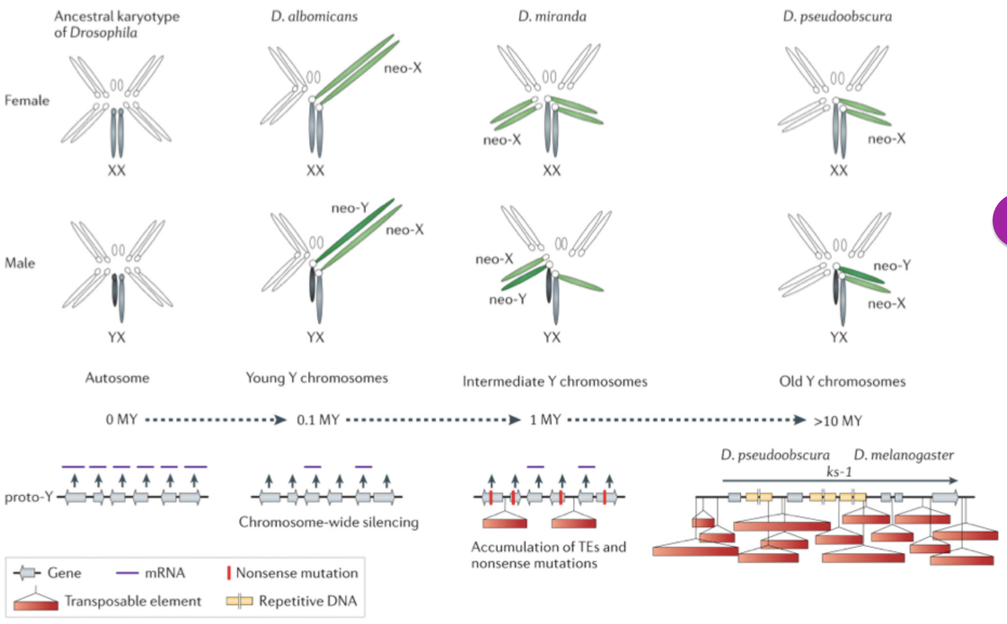


**Figure 4** Neo-sex chromosomes in *Drosophila species.* Neo-sex chromosomes (in green) and ancestral sex chromosomes (in grey). 0 million years is set as the time of their origin (taken from Ref.1).

### 5 Evolutionary dynamics of Y-chromosome degeneration

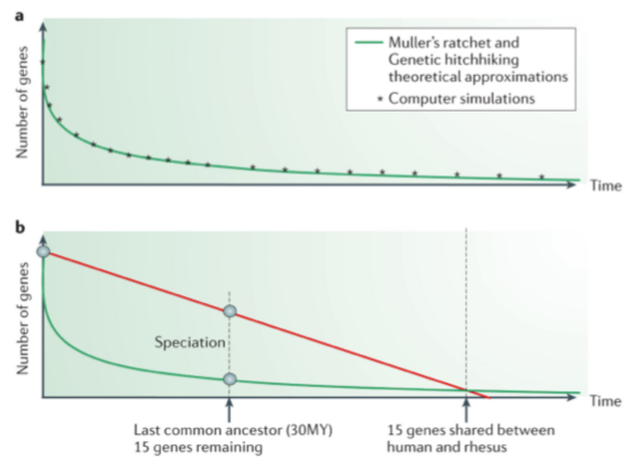


**Figure 5** Comparison between a simple model of constant gene decay and extinction of the Y-chromosome (red line), theoretical approximations (green line) and computer simulations (*) (taken from Ref. 1) .

## Conclusions

**OPEN QUESTIONS**

- ❖ Why not all homomorphic sex chromosomes stop recombining with each other and thus become heteromorphic over long evolutionary periods?
- ❖ If genetic diversity is one of the cornerstones of evolution, how can a chromosome largely devoid of recombination with a meiotic partner survive?
- ❖ Can any predictions be made about the evolutionary fate of the Y-chromosome?

What remains is a sex-determination system, regardless of how it is accomplished.

Further comparative analysis of Y-chromosomes at different ages of differentiation will give major insights into the characteristics and the evolutionary forces that act on sex chromosomes.

References

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[2] Navarro-Costa P. 2012. Sex, rebellion and decadence: The scandalous evolutionary history of the human Y-chromosome. *Biochim Biophys Acta*. **1822**: 1851-63.