The Evolution of Sex Chromosomes through the Baldwin Effect

Larry Bull

Computer Science Research Centre
Department of Computer Science & Creative Technologies
University of the West of England, Bristol UK
Larry.Bull@uwe.ac.uk

Abstract

It has recently been suggested that the fundamental haploid-diploid cycle of eukaryotic sex exploits a rudimentary form of the Baldwin effect. Thereafter the other associated phenomena can be explained as evolution tuning the amount and frequency of learning experienced by an organism. Using the well-known NK model of fitness landscapes it is here shown that the emergence of sex determination systems can also be explained under this view of eukaryotic evolution.

Keywords: Allosomes, Eukaryotes, Meiosis, NK model, Recombination
Introduction

The evolution and maintenance of sex is one of the defining characteristics of eukaryotes. Sex is here defined as successive rounds of syngamy and meiosis in a haploid-diploid lifecycle. It has recently been suggested that the emergence of a haploid-diploid cycle enabled the exploitation of a rudimentary form of the Baldwin effect [Baldwin, 1896] and that this provides an underpinning explanation for all the observed forms of sex [Bull, 2017]. The Baldwin effect is here defined as the existence of phenotypic plasticity that enables an organism to exhibit a significantly different (better) fitness than its genome directly represents. Over time, as evolution is guided towards such regions under selection, higher fitness alleles/genomes which rely less upon the phenotypic plasticity can be discovered and become assimilated into the population.

Whether the diploid is formed via endomitosis or syngamy, the fitness of the cell/organism is a combination of the fitness contributions of the composite haploid genomes. If the cell subsequently remains diploid and reproduces asexually, there is no scope for a rudimentary Baldwin effect. However, if there is a reversion to haploid cells under meiosis, there is potential for a mismatch between the utility of the haploids compared to that of the diploid; individual haploids do not contain all of the genetic material over which selection operated. That is, the effects of genome combination can be seen as a simple form of phenotypic plasticity for the individual haploid genomes before they revert to a solitary state and hence the Baldwin effect may occur. Processes such as recombination can be shown to vary the amount of learning occurring (after [Bull, 1999]).

In this short paper, following [Bull, 2017], the emergence of sex determination chromosomes is explored using versions of the well-known NK model [Kauffman & Levin, 1987] of fitness landscapes where size and ruggedness can be systematically altered. Results suggest that the X0 (Z0) and XY (ZW) systems can be shown to be beneficial under certain conditions as they also enable variation in the amount of learning occurring on rugged fitness landscapes.
Kauffman and Levin [1987] introduced the NK model to allow the systematic study of various aspects of fitness landscapes (see [Kauffman, 1993] for an overview). In the standard model, the features of the fitness landscapes are specified by two parameters: \( N \), the length of the genome; and \( K \), the number of genes that has an effect on the fitness contribution of each (binary) gene. Thus increasing \( K \) with respect to \( N \) increases the epistatic linkage, increasing the ruggedness of the fitness landscape. The increase in epistasis increases the number of optima, increases the steepness of their sides, and decreases their correlation. The model assumes all intragenome interactions are so complex that it is only appropriate to assign random values to their effects on fitness. Therefore for each of the possible \( K \) interactions a table of \( 2^{(K+1)} \) fitnesses is created for each gene with all entries in the range 0.0 to 1.0, such that there is one fitness for each combination of traits (Figure 1). The fitness contribution of each gene is found from its table. These fitnesses are then summed and normalized by \( N \) to give the selective fitness of the total genome.

![Figure 1: An example NK model (N=3, K=1) showing how the fitness contribution of each gene depends on K random genes (left). Therefore there are \( 2^{(K+1)} \) possible allele combinations per gene, each of which is assigned a random fitness. Each gene of the genome has such a table created for it (right, centre gene shown). Total fitness is the normalized sum of these values.](image-url)
Kauffman [1993] used a mutation-based hill-climbing algorithm, where the single point in the fitness space is said to represent a converged species, to examine the properties and evolutionary dynamics of the NK model. That is, the population is of size one and a species evolves by making a random change to one randomly chosen gene per generation. The “population” is said to move to the genetic configuration of the mutated individual if its fitness is greater than the fitness of the current individual; the rate of supply of mutants is seen as slow compared to the actions of selection. Ties are broken at random. Figure 2 shows example results. All results reported in this paper are the average of 10 runs (random start points) on each of 10 NK functions, ie, 100 runs, for 20,000 generations. Here $0 \leq K \leq 15$, for $N=20$ and $N=100$.

Figure 2: Showing typical behaviour and the fitness reached after 20,000 generations on landscapes of varying ruggedness ($K$) and length ($N$). Error bars show min and max values.
Figure 2 shows examples of the general properties of adaptation on such rugged fitness landscapes identified by Kauffman (eg, [1993]), including a “complexity catastrophe” as $K \to N$. When $K=0$ all genes make an independent contribution to the overall fitness and, since fitness values are drawn at random between 0.0 and 1.0, order statistics show the average value of the fit allele should be 0.66. Hence a single, global optimum exists in the landscape of fitness 0.66, regardless of the value of $N$.

At low levels of $K$ ($0<K<8$), the landscape buckles up and becomes more rugged, with an increasing number of peaks at higher fitness levels, regardless of $N$. Thereafter the increasing complexity of constraints between genes means the height of peaks typically found begin to fall as $K$ increases relative to $N$: for large $N$, the central limit theorem suggests reachable optima will have a mean fitness of 0.5 as $K \to N$. Figure 2 shows how the optima found when $K>6$ are significantly lower for $N=20$ compared to those for $N=100$ (T-test, $p<0.05$).

**Eukaryotes: Sexual Diploid Evolution (Typically)**

As noted above, bacteria and archaea can be described as “simpler” than eukaryotes for a variety of reasons including their typically containing a single genome. Hence, as in the NK model above, a given combination of gene values in their genome can be viewed as representing a single point in an $N$-dimensional fitness landscape. In contrast, eukaryotes can contain two or more genomes, typically two, and reproduce sexually via a haploid-diploid cycle with meiosis. Eukaryotes can therefore be viewed as a single point in a fitness landscape of all possible diploids. However, it has recently been suggested that diploid eukaryotes should be viewed as simultaneously representing two points in the fitness landscape of their constituent haploid genomes to explain why sex emerged [Bull, 2017].

Significantly, since the phenotype and hence fitness of a diploid individual is a composite of its two haploid genomes, evolution can be seen to be assigning a single fitness value to the region of the landscape between the two points represented by its constituent haploid genomes. That is, it is proposed that evolution is using a more sophisticated scheme by which to navigate the fitness landscapes of eukaryotes than for prokaryotes: an individual organism provides a generalization in the fitness landscape as opposed to information about a single point. This is seen as particularly useful as the complexity of the fitness landscape increases in both size and ruggedness.
Moreover, the haploid-diploid cycle can also be explained as creating a rudimentary form of the Baldwin effect [Baldwin, 1896], thereby enabling the beneficial smoothing of rugged fitness landscapes [Hinton & Nowlan, 1987]. Briefly, under the Baldwin effect, the existence of phenotypic plasticity potentially enables an organism to display a different (better) fitness than its genome directly represents. Typically, such plasticity is assumed to come from the organism itself, eg, through the modification of neural connectivity. However, a genetically defined phenotype can also be altered by the exploitation of something in the environment, eg, a tool. It is suggested that haploids forming pairs to become diploid is akin to the latter case of learning/plasticity with the partner’s genome providing the variation. Being diploid can potentially alter gene expression in comparison to being haploid and hence affect the expected phenotype of each constituent haploid alone since both genomes are active in the cell - through changes in gene product concentrations, partial or co-dominance, etc. If the cell/organism subsequently remains diploid and reproduces asexually, there is no scope for a rudimentary Baldwin effect. However, if there is a reversion to a haploid state for reproduction under a haploid-diploid cycle, there is the potential for a significant mismatch between the utility of the haploids passed on compared to that of the diploid selected; individual haploid gametes do not contain all of the genetic material through which their fitness was determined.

The variation processes can then be seen to change the bounds for sampling combined genomes within the diploid by altering the distance between the two end points in the underlying haploid fitness landscape. That is, the degree of possible change in the distance between the two haploid genomes controls the amount of learning possible per cycle. Numerous explanations exist for the benefits of recombination (eg, [Bernstein and Bernstein, 2010]) but the role becomes clear under the new view: recombination moves the current end points in the underlying haploid fitness space which define the fitness level generalization either closer together or further apart. That is, recombination adjusts the size of an area assigned a single fitness value, potentially enabling higher fitness regions to be more accurately identified over time. Moreover, recombination can also be seen to facilitate genetic assimilation within the simple form of the Baldwin effect: if the haploid pairing is beneficial and the diploid cell/organism is chosen under selection to reproduce, the recombination process can bring an assortment of those partnered genes together into new haploid genomes. In this way the fitter allele values from the pair of partnered haploids may come to exist within individual haploids more quickly.
than the under mutation alone (see [Bull, 2017] for full details). Mutation can also be seen to be adjusting the distance between the two genomes at a generally reduced rate per generation. It has previously been shown how both the amount of learning per step and the rate at which it occurs can affect utility, with more learning typically proving increasingly beneficial with increasing $K$ [Bull, 1999].

![Diagram](image)

Figure 3: Showing the haploid-diploid cycle with meiosis as implemented with a converged population here.

**Two-step Meiosis with Recombination in the NK Model**

As discussed in [Maynard-Smith & Szathmary, 1995, p150] the first step in the evolution of eukaryotic sex was the emergence of a haploid-diploid cycle, probably via endomitosis, then simple syngamy or one-step meiosis, before two-meiosis with recombination. Following [Bull, 2017], the NK model can be extended to consider aspects of the evolution of sexual diploids. Firstly, each individual contains two haploid genomes of the form described above for the standard model. The fitness of an individual is here simply assigned as the average of the fitness of each of its constituent haploids. These are initially created at random, as before. Two-step meiosis with recombination is here implemented as follows (Figure 3): on each generation the diploid individual representing the converged population is copied twice to create two offspring. In each offspring, each haploid genome is copied once, a single
recombination point is chosen at random, and non-sister haploids are recombined. One of the four resulting haploids in each offspring individual is chosen at random. Finally, a random gene in each chosen haploid is mutated. The resulting pair of haploids forms the new diploid offspring to be evaluated.

![Figure 4](image.png)

Figure 4: Showing typical behaviour and fitness reached after 20,000 generations on landscapes of varying ruggedness ($K$) and size ($N$) for diploids undergoing two-step meiosis with recombination.

Figure 4 shows examples of the general properties of adaptation in the NK model of rugged fitness landscapes for diploid organisms evolving via two-step meiosis with recombination. When $N=20$ the fitness level reached is significantly lower than for $N=100$ for $K>4$ (T-test, $p<0.05$), as was seen in the traditional haploid case due to the effects of the increased landscape complexity. Following [Bull, 2017], it can be seen that fitness levels are always higher than the equivalent haploid case (Figure 2) when $K>0$ due to the Baldwin effect as discussed (T-test, $p<0.05$).

**Sex Chromosomes**

The emergence of isogamy, ie, mating types, was not considered in the explanation for the evolution of two-step meiosis with recombination above (as in [Bull, 2017]). However, the presence of allosomes - XY in animals and ZW in birds, some fish, reptiles, insects, etc. – can also be explained as a mechanism by which a haploid genome may vary the amount of learning it experiences when...
paired with another to form a diploid organism. This is because, as noted above, the most beneficial amount of learning can vary with landscape ruggedness [Bull, 1999]. For convenience, XY sex determination will be used in the discussions here but the argument applies equally well to the ZW system. The simpler X0 system, as found in most arachnids, for example, is used as a starting point.

In the X0 system, females have two X chromosomes and are generally hermaphrodite, whereas the typically rarer males have one X chromosome. Following the approach in [Bull, 2017] of considering the haploid genome fitness landscape of primary importance, as opposed to that of the diploid, the males in the X0 system can be viewed as not using the rudimentary learning process for the genes contained in the X chromosome. That is, the females contain two X chromosomes and the fitness contribution of the genes therein will be some combination of the two, eg, their average was used above. Note that when X-inactivation occurs the choice is random (in placental cells) and hence the organism as a whole remains a composite of the two chromosomes. In contrast, in the male the fitness contribution of the genes in the X chromosome is simply the same as in the haploid case, ie, without the potential for any learning since they are not paired with another set of corresponding genes. Thus a male can be seen as a mechanism through which the learning experienced in the region of the haploid genome fitness landscape covered by the genes contained in the X chromosome is removed; a male provides additional "raw" haploid fitness contribution information.

Figure 5 (left) shows examples of the basic two-step meiosis with recombination model from above extended to contain two mating types as in the X0 system. For simplicity, the (converged population of) females here consist of two X chromosomes only and the (converged population of) males of one X chromosome only, ie, males are strictly haploid. After the first haploid genome has been generated from the female under meiosis, a mutated copy of the male genome is then generated and accepted probabilistically half of the time. When accepted, a new (diploid) female is said to have been created and is evaluated as above, replacing the original female if fitter, and vice versa to create a new male offspring. Males and females are seeded with the same genomes. Considering the fitness of the females, no benefit over the hermaphrodites above (Figure 4) is found with $N=20$ for any $K$ but with $N=100$, a significant increase in fitness is seen when $K=10$ ($T$-test, $p<0.05$). Note this remains the case if the average fitness of the male and female is used for the comparison (not shown).
Figure 5 (right) shows examples from extending the model further to the XY system. Here a male consists of an X and a Y chromosome/genome and since, as in the XY system only a small - if any - section of X and Y chromosomes typically recombine, a basic syngamy process is used for males, ie, one-step meiosis without recombination. Hence if a Y chromosome is chosen from the pair generated by the male, a male offspring is created and vice versa. Male fitness is the average of its two constituent haploid genomes. Considering the fitness of the females again, no benefit over the hermaphrodites is found with \( N=20 \) for any \( K \) but with \( N=100 \), a significant increase in fitness is seen when \( 6 \leq K \leq 10 \) (T-test, \( p<0.05 \)). Again, this remains the case if the average fitness of the male and female is used for the comparison, and this is also the case regardless of whether the Y chromosome is evaluated on the same haploid fitness landscape as the X or another created at random (not shown).

![Graphs showing fitness versus K for N=100 in X0 and XY systems](image)

Figure 5: Showing the typical fitness reached after 40,000 generations by females under the X0 system on landscapes of varying ruggedness (left) and under the XY system (right).

It can be noted that there is no significant difference in the fitness levels reached for any \( N \) or \( K \) combination between the X0 and XY systems as described (T-test, \( p \geq 0.05 \)). Clearly, in the XY system the male can no longer be seen to provide a haploid-like evaluation of the X chromosome. It therefore seems that it is the extra (approximate) fitness value information that proves beneficial to the learning/generalisation process described above. Note this is not the same as organisms becoming triploid since the X chromosome is evaluated in a different context in a male: two fitness values for the X chromosome will always exist with two mating types. A potential benefit of the XY system over X0
exists when the correlation between the diploid fitness landscape and underlying haploid fitness landscape is considered (after [Mayley, 1996]), as will be discussed.

Conclusion

The evolution of allosomes was explored and a beneficial effect from the extra (approximate) fitness value information the X0 (Z0) and XY (ZW) systems enable was found. The diploid hermaphrodites exploiting two-step meiosis with recombination can be expected to eventually converge upon organisms carrying two copies of the same (or very nearly) haploid genome. This means that over evolutionary time they become increasingly restricted to the region of symmetry within the diploid fitness landscape, ie, the region where constituent haploid genome A is genetically identical to haploid genome B. However, the highest levels of fitness may exist in other areas of the diploid landscape. One way to avoid a near complete set of homozygotes is to maintain a region(s) in the haploid genome where recombination does not occur, thereby inhibiting the assimilation process. It is here suggested this is what the XY (ZW) system enables over the X0 (Z0) system. Asexual reproduction and one-step meiosis, ie, a haploid-diploid cycle without recombination, can also hinder convergence. Whilst the results in [Bull, 2017] suggest two-step meiosis with recombination is generally beneficial over asexuality and one-step meiosis due to the increased amount of learning, there is some overlap at low levels of landscape ruggedness (eg, \(K=2\) [Bull, 2017]). Parabasalid are known to exploit simple syngamy and since they have lost their mitochondria may be seen to exist on less rugged landscapes (after [Bull & Fogarty, 1996]). It can be speculated that they may therefore be exploiting the potential to move away from the region of symmetry within their diploid fitness landscapes. Having adopted the generally more beneficial two-step meiosis with recombination, the XY (ZW) system enables eukaryotes to maintain heterozygotes for some regions of their overall genome space in a relatively controlled manner. This explains such things as why recombination does not occur between X and Y chromosomes, why the situation is reversed in the ZW system since which mating type maintains the single copy is not important, why the two sex chromosomes can be of different sizes which is potentially related to the degree of heterogeneity required between the two haploids to reach the higher fitness areas, etc.
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