

SEX AND EVOLUTION IN EUKARYOTES

C. William Birky, Jr.

Department of Ecology and Evolutionary Biology, The University of Arizona, Tucson, Arizona, USA

Keywords: amphimixis, apomixis, asexual, automixis, clone, extinction, inbreeding, meiosis, mitosis, outcrossing, parasexual, parthenogenesis, recombination, selection, selfing, sexual, speciation, syngamy

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Summary

Sex, in the sense of the combination of genes from two different individuals, occurs in prokaryotes as well as eukaryotes. However, sex with meiosis and syngamy (amphimixis) is limited to eukaryotes, which are the focus of this chapter. Eukaryotic life cycles include strictly asexual reproduction (apomixis) involving only mitotic cell divisions and as well as amphimixis which is sex in the most narrow sense. The most common and probably ancestral life cycle includes both, but some organisms are strictly

asexual while reproduction in a few groups always involves amphimixis. Besides apomixis, asexual reproduction can also occur by automictic parthenogenesis which involves meiosis but not syngamy. The origin of amphimixis is so conjectural that it is not covered here. The loss of sex via mutation, polyploidization, or hybridization is common and largely irreversible. Moreover, it is often selectively advantageous at the individual level so that asexual genomes have an excellent chance of replacing their sexual conspecifics, turning a sexual species into an asexual species. These processes would, if unchecked, result in the replacement of most or all sexual species by asexuals. Because many or most eukaryotes still reproduce sexually, there must be selection in the opposite direction that favors sex. This selection probably occurs mainly at the level of species, such that asexual species have a higher rate of extinction and a lower rate of speciation than sexual species. Numerous theoretical models have been proposed that give a selective advantage to sex, although some give sex a disadvantage. The most generally applicable of these models are stochastic, invoking random genetic drift that makes natural selection less effective with asexual than with sexual reproduction. Consequently, compared to sexual species, asexual species tend to accumulate more deleterious mutations, which increases the rate of extinction. Conversely asexual species are more prone to lose advantageous mutations by chance, which retards adaptation to new environments. This further increases the probability of extinction when environments change; it also decreases the rate of speciation via disruptive selection in asexuals compared to sexuals.

Although the theory described above is very general, the advantage of sex depends on many details such as the amount of inbreeding. Also that advantage can be overcome by various kinds of adaptations. Consequently the magnitude of the effects of the loss of sex varies greatly among different groups of eukaryotes, and this is reflected in variation in the proportion of asexual and sexual species.

1. Introduction and Overview

As geneticists and evolutionary biologists see it, sex involves the exchange of genes between individuals and the recombination of alleles of different genes to produce new genotypes. It has apparently evolved at least four different times, three in prokaryotes where there are three different vehicles for gene exchange and once in the ancestors of eukaryotes when meiosis evolved. These were incredibly important evolutionary advances and understanding them is one of the most important problems in biology. However, it is also an extremely hard problem. No present-day taxa show obvious intermediate stages in the evolution of sex. Unraveling these stages will probably require resolving the deep branches of the phylogenetic tree of life, the use of complete genomes to trace the origins of genes involved in sex, and detailed studies of the genetics of groups where the mechanisms of mitosis, meiosis, and syngamy are still vague. These are problems that will occupy future biologists for a number of generations. At the present time, theories about the origins of sexual processes are very speculative and will not be covered in this article.

Our focus will be on the maintenance of meiotic sex in eukaryotes. Sex in eukaryotes occurs exclusively between members of a species and usually involves syngamy (fusion of haploid cells) followed eventually by meiosis with recombination and cell division.

(Parasexual cycles which do not involve meiosis but can have similar genetic consequences will be dealt with separately in Section 4.) In contrast, sex in Bacteria and Archaea involves the transfer of genes from one cell to another by means of transformation, transduction, or conjugation. Prokaryotes also differ in that some forms of gene exchange may cross species boundaries. Consequently much of theory and conclusions about the genetic consequences of sex that were developed for eukaryotes and will be discussed here may not apply to prokaryotes, or may apply but only with modification. But it is also possible that sex in prokaryotes arose and is maintained by very different mechanisms. It has been argued that transformation is an adaptation that allows starving cells to obtain food (DNA) from dead cells. Conjugation is mediated by selfish genetic elements, the F factors, and may be maintained by those elements because it facilitates their spread. Transduction is mediated by bacteriophage and may be an accidental byproduct of their replication.

We will see that both asexual reproduction involving mitosis and sexual reproduction involving syngamy and meiosis probably arose in the ancestor of all living eukaryotes. The ancestral eukaryote probably reproduced by mitosis most of the time but with occasional periods of sexual reproduction, as do most protists and many invertebrates, plants, and fungi today. The sexual phase was subsequently lost in a number of eukaryote species, and in fact is still being lost today. Life cycles with exclusively sexual reproduction probably evolved later, independently in multicellular animals and plants, but in some species it was replaced by parthenogenesis. Asexual individuals are easily produced by mutation or hybridization and the process is almost always irreversible. Moreover, asexual individuals usually have a selective advantage over sexual members of the same species and often replace them, transforming a sexual species into an asexual species; also asexual individuals tend to be good at colonizing new habitats in which they form new asexual species. If these processes continued unchecked, there would be few or no sexual eukaryotes left. Since this has not happened, we can assume that most asexual mutants and species must have some compensating selective disadvantage.

Consequently, in order to understand the phylogenetic distribution of asexual and sexual forms, we must understand the evolutionary consequences of *losing* sex in eukaryotes. We will see that many of the best evolutionary biologists and geneticists have proposed a variety of competing theories about the evolutionary advantages of sex. Happily, in the past few decades some unifying principles have finally emerged; in particular, the most general statement one can make about the evolutionary difference between asexual and sexual reproduction is that natural selection is more effective with sex. This means that asexual species are more prone to accumulate detrimental mutations and less able to retain and fix advantageous mutations that would allow them to adapt to changing conditions or colonize new habitats. The net result is a higher rate of extinction and lower rate of speciation. The formal theories behind this conclusion have given us the tools to begin to understand why sex is so common in eukaryotes, and also why specific groups have managed without it.

2. Species in Sexual and Asexual Organisms

The presence or absence of sexual reproduction has consequences for both individuals

and species. For example, the rate of speciation is likely to be lower in asexuals than in sexuals. Consequently, we cannot develop and test theories about the evolutionary consequences of sex unless we have a species concept (a theoretical model of species and speciation) that identifies comparable units in sexual and asexual organisms. The evolutionary species concept defines species as inclusive populations of individuals that are independent arenas for mutation, selection, and random drift. Thus it is applicable to both asexuals and sexuals and is especially suitable for addressing evolutionary and population genetic aspects of sex. This species concept will be used in theoretical discussions throughout the article; however, experimental studies of speciation and extinction have involved species defined in a variety of ways, often simply as clusters of phenotypically similar individuals. In sexual organisms species are sometimes defined on the basis of interbreeding as in Mayr's "biological" species; these will generally be evolutionary species as well.

3. Eukaryotic Life Cycles

As humans we are most familiar with a life cycle in which a diploid multicellular organism includes a germ line of cells that undergo meiosis and gametogenesis to produce haploid gametes. Gametes from two individuals, an egg from a female and sperm from a male, undergo syngamy (fuse) to produce a diploid zygote; the zygote undergoes mitosis to produce a multicellular individual of the next generation. But of course this life cycle is not the only one seen in nature, and in fact is probably not even the most common one. To really understand the evolution of sex, it is necessary to take a much broader view of life cycles in eukaryotes, including the following variations:

1. Meiotic sexual reproduction in animals and plants involves differentiated female and male individuals or hermaphrodites with both male and female reproductive structures, but in fungi and protists sex involves the fusion of individuals of different mating types that are often phenotypically similar or identical.
2. In plants the haploid products of meiosis can undergo mitotic divisions to produce several cells or nuclei, one or more of which become gametes.
3. Some animals and plants undergo fission or budding to produce offspring; this asexual or vegetative reproduction involves only mitosis in diploid, or occasionally haploid, cells. When there are differentiated sexes, both males and females can reproduce vegetatively.
4. Some animal and plant species consist entirely of parthenogenetic female individuals in which gametogenesis produces a diploid egg by one of several methods described below.
5. In single-celled plants (algae), the majority of the life cycle may be spent as haploid cells; in some algae such as *Chlamydomonas* these haploid cells fuse and the diploid products undergo meiosis without any intervening mitotic divisions.
6. Many other organisms, mostly single-celled, exist only as haploid cells reproducing asexually by mitosis; an example is the green alga *Chlorella* and the euglenoid algae.

7. Both haploid and diploid cells can reproduce asexually by mitotic divisions in the yeast *Saccharomyces cerevisiae*. When starved for nitrogen, diploid cells undergo meiosis to produce haploids.

A generalized eukaryotic life cycle is shown in Figure 1. Some organisms such as yeast have precisely this generalized life cycle; others (like us) lack the mitotic divisions in the haploid or diploid stage; still others lack the sexual stage and are stuck in the haploid or diploid asexual life cycle.

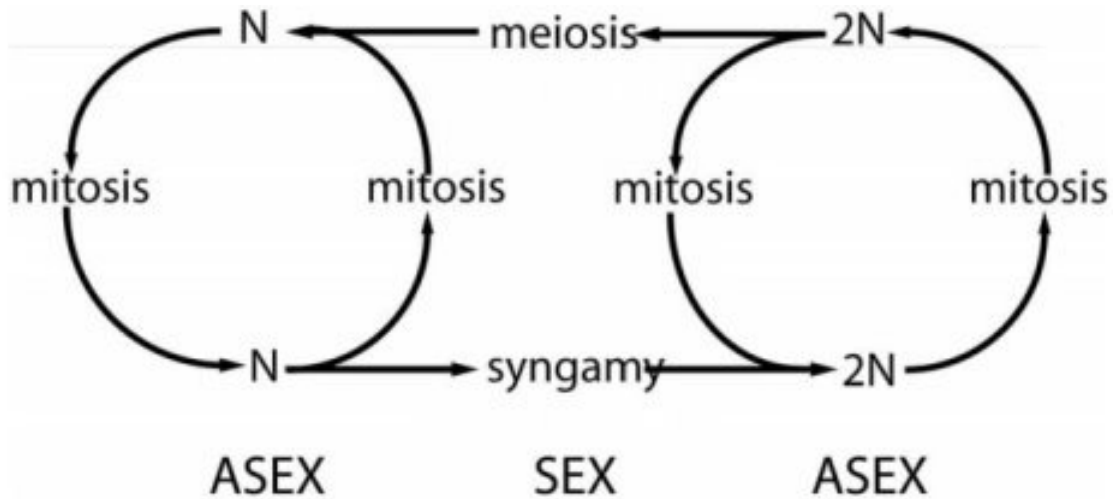


Figure 1. Generalized eukaryotic life cycle. Asexual reproduction occurs via mitosis in haploids, diploids, or both. Transition from diploidy to haploidy occurs via meiotic cell divisions, while the diploid phase is restored by syngamy.

It should be noted that sex and reproduction are not necessarily coupled. As described above, organisms can reproduce without sex via mitotic divisions alone. The sexual part of the eukaryotic life cycle can also occur in the absence of reproduction. For example, during conjugation in ciliated protozoa, two cells join and their diploid nuclei undergo meiosis, then the haploid products are exchanged via cytoplasmic bridges between the two cells and fuse to produce diploid nuclei. The cells then separate, after which they may undergo mitosis and fission.

4. Definitions and Varieties of Sexual and Asexual Reproduction

The variety of mechanisms and genetic consequences of sexual and asexual reproduction make them difficult to classify and define. Moreover, the genetic consequences of some forms of sex are very similar to those of some forms of asexual reproduction. Some important distinctions are:

1. Sexual reproduction can unite haploid genomes of the same individual or clone, in which case it is called selfing, or it can unite genomes from different individuals or clones, called outcrossing. These have very different genetic consequences. Between pure selfing and pure outcrossing there can be any proportion of selfing or other forms of inbreeding and outcrossing. A common model, familiar because it is used

in the Hardy-Weinberg law, is panmixis or random mating in which individuals are chosen at random from the population without regard to their genotype or relatedness. Random mating models may include or exclude the possibility of selfing.

2. Sexual reproduction can involve two individuals, in which case it is said to be biparental, or one individual, called uniparental or clonal reproduction. Asexual reproduction is always uniparental and always produces clones, which are the offspring of single individuals by asexual reproduction only.

A convenient terminology was developed early in the history of genetics to distinguish these phenomena:

1. Amphimixis = meiosis and syngamy with at least some outcrossing. This is sex in the strictest sense.
2. Selfing = meiosis and syngamy involving only one individual. This is also usually called sex, but when it is obligatory (no outcrossing at all, as in some hermaphrodites), it is sometimes called clonal reproduction.
3. Automixis = meiosis with diploidy restored by fusion of two haploid products of the same division or by duplication of one haploid genome. This is generally considered to be asexual or clonal.
4. Apomixis = mitosis only. It is also clonal.

There is an interesting variation on parthenogenesis called gynogenesis in animals or pseudogamy in plants. Here, eggs will not mature unless they are fertilized by males of the same species or a closely related species. The male and female nuclei do not normally fuse so that the male parent does not contribute genes to the offspring. However, in some cases there is a significant rate of “leakage” of paternal genes, so this kind of parthenogenesis might be most accurately described as infrequent and inefficient sexual reproduction interspersed with asexual reproduction.

In this article we will focus mainly on the two extremes of sexual and asexual reproduction, amphimixis and apomixis.

5. Parasexuality

Parasexual cycles have been described in some fungi and trypanosomes, but may occur in other organisms as well, especially in protists. Parasexuality is usually seen in organisms with a predominant haplophase. A parasexual cycle is initiated when two genetically-different cells or hyphae fuse and exchange nuclei. The result is that one or both cells are heterokaryons, having two genetically distinct nuclei. Thus the fusion of cells of genotypes Ab and aB produce a heterokaryon with nuclei of both genotypes. Fusion of the haploid nuclei produces diploid nuclei that are heterozygous with chromosomes Ab and aB . As the cells or hyphae grow, the nuclei undergo mitosis. Mitotic recombination occurs and produces nuclei with recombinant chromosomes, e.g. AB and ab . However, the diploid nuclei are unstable and frequently lose chromosomes,

probably by mitotic nondisjunction. Eventually, repeated loss of chromosomes restores haploidy, but some of the resulting haploid nuclei can have recombinant genotypes. Sorting out of nuclei of different genotypes can then produce recombinant homokaryotic cells.

In the pathogenic yeast *Candida albicans*, parasexual recombination requires the gene *Spol1* that appears to be specific for meiotic recombination in sexual fungi and animals. Moreover, mating in *Candida* involves cells of different mating types, which suggests that *Candida* arose from ancestors that underwent meiosis, and perhaps parasexual cycles as well. Both meiotic sex and a parasexual cycle has been reported in the cellular slime mold *Dictyostelium*. Among the hemoflagellates, *Trypanosoma brucei*, the causative agent of sleeping sickness, is diploid and undergoes meiotic sex in its intermediate host the Tsetse fly, as well as reproducing asexually by mitosis. In contrast, its congener *T. cruzi*, which causes Chagas disease, appears not to have meiosis but produces recombinants via a parasexual cycle. *Giardia lamblia* has recently been shown to engage in outcrossing with recombination and to exchange genes between nuclei in the encysted state; the results of crosses implicate meiotic segregation, although meiosis has not been observed cytologically.

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Bibliography

- Agrawal A.F. (2006) Evolution of sex: Why do organisms shuffle their genotypes? *Current Biology* 16:R696-R704. [Good general review of the subject.]
- Avise, J.C. (2008) *Clonality*. New York, Oxford University Press USA. [Good introduction to the varieties of asexual reproduction in vertebrate animals.]
- Barton N. H. and Charlesworth B. (1998). Why sex and recombination? *Science* 281: 1986-1990. [An early review of the modern population genetic theory approaches to the evolution of sex.]
- Birky C. W. Jr. and Barraclough T. G. (2009) Asexual speciation, in *Lost Sex* (Van Dijk, P., Martens K., Schon, I. eds), in press, [Discusses speciation in asexual organisms and describes two methods of defining evolutionary species in asexuals.]
- Bell G. (1982). *The Masterpiece of Nature*. London, Croom Helm. [Classic and magisterial book on the evolution of amphimixis and parthenogenesis in animals and plants; theoretical treatment outdated.]
- Gladyshev E. and Meselson M. (2008). Extreme resistance of bdelloid rotifers to ionizing radiation. *Proceedings of the National Academy of Sciences USA* 105: 5139-5144. [Recent paper on the ability of the bdelloid rotifer genome to repair DNA damage and its evolutionary consequences. This and the Mark Welch 2008 reference provide an entre into the recent literature on bdelloids.]
- Judson O. P. and Normark B. B. (1996). Ancient asexual scandals. *Trends in Ecology and Evolution* 11: 41-46. [Summarized and popularized the influential idea that very old asexual groups are unexpected and difficult to explain; see Neiman et al. below for evidence to the contrary.]

Kouyos R.D., Silander O.K. and Bonhoeffer S. (2007) Epistasis between deleterious mutations and the evolution of recombination. *Trends in Ecology and Evolution* 22:308-315. [Critical review of epistasis and the mutational deterministic hypothesis but also a lucid overview of other models of the evolution of sex.]

Mandegar M. A. and Otto S. P. (2007). Mitotic recombination counteracts the benefits of genetic segregation. *Proceedings of the Royal Society B*. 274: 1301-1307. [Mitotic recombination can compensate for the lack of meiotic segregation in apomictic species.]

Mark Welch D. B., Mark Welch J. B., and Meselson M. (2008). Evident for degenerate tetraploidy in bdelloid rotifers. *Proceedings of the National Academy of Sciences USA* 105: 5145-5149. [Recent paper on the structure of the bdelloid rotifer genome and its evolutionary consequences. This and the Gladyshev 2008 reference provide an entre into the recent literature on bdelloids.]

Maynard Smith J. (1978) *The Evolution of Sex*. Cambridge, Cambridge University Press. [Classic and influential book on the theory of the evolutionary advantages and disadvantages of the evolution of amphimixis and parthenogenesis, mainly in animals and plants; introduced the two-fold cost of sex; somewhat dated.]

Michod R. E. and Levin B. R. Eds. (1988). *The Evolution of Sex*. Sunderland, Mass., Sinauer Associates, Inc. [A collection of reviews of most of the major theoretical approaches to the evolution of sex by various authors.]

Neiman M., Meirmans S., and Meirmans P. G. (2009) What can asexual lineage age tell us about the maintenance of sex? *Annals of the New York Academy of Sciences*. 1168:185-200. [Review of data on the ages of asexual lineages and the implications of those data for theories of the evolutionary advantages of sex; shows there is no justification for calling ancient asexuals "scandalous".]

Normark B. B., Judson O. P., and Moran N. P. (2003). Genomic signatures of ancient asexual lineages. *Biological Journal of the Linnean Society* 79: 69-84. [Review of genetic and molecular methods for distinguishing asexual from sexual species.]

Otto S. P. and Gerstein A. C. (2006). Why have sex? The population genetics of sex and recombination. *Biochemical Society Transactions* 34: 519-522. Excellent non-mathematic review of the theory of the evolutionary advantage of sex; concise and readable.]

Otto S. P. and Lenormand T. (2002). Resolving the paradox of sex and recombination. *Nature Reviews Genetics* 3: 252-261. [Excellent review of the theory of the evolutionary advantage of sex; modest amount of math.]

Rice W. R. (2002). Experimental tests of the adaptive significance of sexual recombination. *Nature Reviews Genetics* 3: 241-251. [Reviews theory as well as experimental tests.]

Schwander T. and Crespi B. J. (2009) Twigs on the tree of life? Neutral and selective models for integrating macroevolutionary patterns with microevolutionary processes of asexuality. *Molecular Ecology* 18:28-42. [The first rigorous test of the idea that asexual species are short-lived compared to sexual species.]

Wilkins A. S. and Holliday R. (2009) The evolution of meiosis from mitosis. *Genetics* 181:3-12. [A recent, and novel, example of hypothetical models of the origin of meiosis.]

Biographical Sketch

C. William Birky, Jr. is professor emeritus of Ecology and Evolutionary Biology at the University of Arizona. His research is focused on two fundamental biological problems: the evolutionary consequences of the loss of sex, and the nature of species. In the laboratory he and his students study the parthenogenetic bdelloid rotifers and their relatives.