

# Genetic Drift in Human Populations

Advanced article

Online posting date: 30<sup>th</sup> April 2008

Andrew J Bohonak, *San Diego State University, San Diego, California, USA*

Genetic drift consists of changes in allele frequencies due to sampling error. Even if all individuals in a population have the same opportunities to mate, their reproductive contributions to the next generation will vary due to random chance alone. In any population of finite size, this sampling error will cause gene frequencies to fluctuate from generation to generation. Genetic changes due to drift are neither directional nor predictable in any deterministic way. Nonetheless, genetic drift leads to evolutionary change even in the absence of mutation, natural selection or gene flow.

Studies of evolution have focused strongly on natural selection since the time of Darwin (1859) and Fisher (1930). However, even Darwin (1859) acknowledged that heritable polymorphisms which do not affect survival or reproduction 'would be left a fluctuating element'. During the modern synthesis, Wright (1931, 1932) developed a holistic theory of evolution in natural populations that involved complex interactions among alleles, inbreeding, gene flow and the random effects of *genetic drift*. Wright's influences on the fields of population genetics and molecular evolution include the dominant mathematical framework for genetic drift that is still in use today.

Genetic drift consists of changes in allele frequencies due to sampling error. Even if all individuals in a population have the same opportunities to mate, their reproductive contributions to the next generation will vary due to random chance alone. In any population of finite size, this sampling error will cause gene frequencies to fluctuate from generation to generation. Genetic changes due to drift are neither directional nor predictable in any deterministic way. All populations of finite size experience genetic drift to some extent, with effects that are inversely proportional to the *effective population size*,  $N_e$ . Effective population size is a mathematical abstraction used to correct for factors such as unequal sex ratios, variance among individuals in reproductive output and temporal fluctuations in population size (Ewens, 1982). More precisely, the effective population size is the number of individuals that would be found in an ideal population that undergoes the same amount of drift as the real population. **See also:** [Effective Population Size](#)

A simple mathematical model of drift can be constructed by assuming that individuals are diploid (i.e. the gene pool has a size of  $2N_e$ ), and that each gene copy produces an

infinite number of gametes. The homozygosity of the population, denoted as  $F$ , is the probability that two gametes chosen randomly are identical by descent, because they trace back to the same gene copy in a previous generation. By definition,  $F$  ranges from 0 to 1. Because the probability of two gametes being identical by descent is  $1/(2N_e)$ ,  $F$  increases over time as

$$F_{t+1} = \frac{1}{2N_e} + \left(1 - \frac{1}{2N_e}\right)F_t$$

*Heterozygosity*, the probability that two gametes are not identical by descent, is denoted  $H$  and calculated as  $H = 1 - F$ . From this, it follows that  $H$  will decrease over time:

$$H_{t+1} = \left(1 - \frac{1}{2N_e}\right)H_t$$

and

$$H_t = \left(1 - \frac{1}{2N_e}\right)^t H_0$$

The major implication is that all populations of finite size will eventually drift to 'fixation' where  $H=0$  and only a single allele remains, unless diversity is maintained by mutation, gene flow or natural selection. Because genetic drift is a random process, different populations may reach fixation for different alleles. In the absence of other factors, the probability that any particular allele will eventually reach fixation is its current frequency.

Genetic drift provides a theoretical framework to evaluate the effects of habitat fragmentation, captive breeding programs, population *bottlenecks* (severe reductions in population size that accelerate drift) and founder effects (bottlenecks associated with the founding of a new population). Drift and mutation are implicit in the most common null models for molecular genetics, relegating natural selection to an alternative that is invoked when the null is rejected. For example, the *neutral theory of evolution* (Kimura, 1968) posits that observed polymorphisms in natural populations are due to a balance between genetic drift and mutation, rather than natural selection. Analyses of empirical genetic data typically rely on equilibrium

**ELS subject area:** Evolution and Diversity of Life

**How to cite:**

Bohonak, Andrew J (April 2008) Genetic Drift in Human Populations. In: Encyclopedia of Life Sciences (ELS). John Wiley & Sons, Ltd: Chichester. DOI: 10.1002/9780470015902.a0005440.pub2

expectations for the neutral theory (in its pure or a modified form) (e.g. Tajima, 1989). However, for evolution at the phenotypic level, the utility of drift versus natural selection as a null model has long been the subject of periodic debates (reviewed by Provine, 1986; Beatty, 1992).

Empirically, random genetic drift has been studied in humans, as well as natural and experimental populations of innumerable other organisms. For example, Helgason *et al.* (2003) showed through a variety of analyses that patterns of genetic variation in Icelanders (such as low genetic diversity) are consistent with pronounced levels of past genetic drift. Expectations that the importance of genetic drift will exceed that of natural selection in small populations have been repeatedly validated in studies of human molecular evolution. For example, high frequencies for particular diseases in relatively closed ethnic groups are generally attributable to high rates of genetic drift, either during or after the founding of these groups (e.g. Ostrer, 2001). The high frequency of achromatopsia on Pingelap Atoll in Micronesia provides an often-cited example of how extreme bottlenecks in population size can affect the frequency of deleterious alleles (Hussels and Morton, 1972). Neutral models of genetic drift and mutation have been extended to quantitative traits as well (e.g. Lande, 1976; Orr, 1998). Using these analyses, several studies have suggested that many aspects of evolution in early *Homo* facial morphology may be more consistent with genetic drift than natural selection (e.g. Ackermann and Cheverud, 2004; Weaver *et al.*, 2007). Often, the effective size of one or more populations is inferred from their genetic composition, based on theoretical models that include drift (e.g. Hey, 2005). Although methods for estimating changes in effective population size across the entire depth of a gene genealogy are available, they have been applied primarily to pathogens rather than human populations (e.g. Shackelton *et al.*, 2006). **See also:** Darwin and the Idea of Natural Selection; Mutational Change in Evolution

Molecular inferences concerning the history of human populations are an area of active research, providing data that cannot easily be obtained from fossils, linguistics or other sources. However, because drift and mutation are stochastic processes, historical inferences are only robust when multiple genes are analysed in a framework that accounts for this stochasticity. Based on the analyses of 25 gene regions, Templeton (2005) has posited numerous range expansions over the past 2 million years of human evolution. Similarly, Gherman *et al.* (2007) suggested that an unusually high burst of large-scale changes in the human genome occurred *c.* 54 million years ago, attributable to high rates of drift in the common ancestor of New World monkeys and the clade that includes New World monkeys and Hominoidea. **See also:** Fixation Probabilities and Times; Genetic Variation: Polymorphisms and Mutations; Heterozygosity; Polymorphisms: Origins and Maintenance; Population Genetics: Historical Aspects; Population Genetics: Overview

## References

- Ackermann RR and Cheverud JM (2004) Detecting genetic drift versus selection in human evolution. *Proceedings of the National Academy of Sciences of the USA* **101**: 17946–17951.
- Beatty J (1992) Random drift. In: Keller EF and Lloyd EA (eds) *Keywords in Evolutionary Biology*. pp. 273–281. Cambridge, MA: Harvard University Press.
- Darwin C (1859) *On the Origin of Species by Means of Natural Selection, or the Preservation of Favoured Races in the Struggle for Life*. 6th edn. London: John Murray.
- Ewens WJ (1982) On the concept of effective population size. *Theoretical Population Biology* **21**: 373–378.
- Fisher RA (1930) *The Genetical Theory of Natural Selection*. New York: Dover.
- Gherman A, Chen PE, Teslovich TM *et al.* (2007) Population bottlenecks as a potential major shaping force of human genome architecture. *PLoS Genetics* **3**: e119.
- Helgason A, Nicholson G, Stefánsson K and Donnelly P (2003) A reassessment of genetic diversity in Icelanders: strong evidence from multiple loci for relative homogeneity caused by genetic drift. *Annals of Human Genetics* **67**: 281–297.
- Hey J (2005) On the number of New World founders: a population genetic portrait of the peopling of the Americas. *PLoS Biology* **3**: e193.
- Hussels IE and Morton NE (1972) Pingelap and Mokil Atolls: achromatopsia. *American Journal of Human Genetics* **24**: 304–309.
- Kimura M (1968) Evolutionary rate at the molecular level. *Nature* **217**: 624–626.
- Lande R (1976) Natural selection and random genetic drift in phenotypic evolution. *Evolution* **30**: 314–334.
- Orr HA (1998) Testing natural selection vs. genetic drift in phenotypic evolution using quantitative trait locus data. *Genetics* **196**: 2099–2104.
- Ostrer H (2001) A genetic profile of contemporary Jewish populations. *Nature Reviews. Genetics* **2**: 891–898.
- Provine WB (1986) *Sewall Wright and Evolutionary Biology. Science and Its Conceptual Foundations*. Chicago: University of Chicago Press.
- Shackelton LA, Rambaut A, Pybus OG and Holmes EC (2006) JC virus evolution and its association with human populations. *Journal of Virology* **80**: 9928–9933.
- Tajima F (1989) The effect of change in population size on DNA polymorphism. *Genetics* **123**: 597–601.
- Templeton AR (2005) Haplotype trees and modern human origins. *Yearbook of Physical Anthropology* **48**: 33–59.
- Weaver TD, Roseman CC and Stringer CB (2007) Were Neandertal and modern human cranial differences produced by natural selection or genetic drift? *Journal of Human Evolution* **53**: 135–145.
- Wright S (1931) Evolution in Mendelian populations. *Genetics* **16**: 97–159.
- Wright S (1932) The roles of mutation, inbreeding, crossbreeding and selection in evolution. *Proceedings of the Sixth International Congress of Genetics* **1**: 356–366.

## Further Reading

- Tishkoff SA and Verrelli BC (2003) Patterns of human genetic diversity: implications for human evolutionary history and disease. *Annual Review of Genomics and Human Genetics* **4**: 293–340.