Pop Gen/Human Origins

* The first hominin group to migrate out of Africa was Homo erectus sensu lato (Homo georgicus) about 1.9 million years ago. (Gabunia et al. 2000)
  + The ancestors of Neanderthals, Denisovans, and modern humans—presumably Homo heidelbergensis or Homo rhodesiensis (included in Homo erectus sensu lato)—left Africa between 600 and 300 kya. (Hublin 2009; Stringer 2002)
* The first foray of Homo sapiens out of Africa to the Levant occurred about 125 kya, but this is generally viewed as a failed dispersal as fossil evidence of Homo sapiens vanishes after about 92 kya. (Oppenheimer 2012)
  + Molecular analyses of modern human genomes suggest that a successful expansion of Homo sapiens occurred around 60-50 kya. (Petraglia et al. 2010; Stringer 2011)
  + In one of the most influential genetic studies of human origins, Cann et al (1987) examined the mitochondrial DNA (mtDNA) of 147 contemporary individuals from 5 geographic populations around the world. Their findings traced the last common ancestor of maternally inherited mtDNA in modern human populations to an African origin about 200,000 years old. Non-African lineages were inferred to be about 50,000 years old. This study was heavily criticized, but it laid the groundwork for the debate over human population history.

The debate over the origins of modern Homo sapiens can largely be summarized by two major competing theories and the spectrum of permutations that exist in between them.

1. The multiregional (MR) model of human evolution, which is also known as the regional continuity model, posits that modern humans evolved by anagenesis following the expansion of Homo erectus sensu lato (Weidenreich 1943; Wolpoff 1994). Presumably, Homo erectus gave rise to Neanderthals and then modern humans in Europe, Homo erectus in Asia gave rise to modern Asians, and archaic hominins in Africa gave rise to modern Africans. Continuity of the modern human species was necessarily maintained by gene flow between the different regions.
2. In contrast to the MR hypothesis, the recent African origin (RAO) model of human evolution proposes a single outward migration of anatomically modern Homo sapiens from Africa who replaced all other archaic hominins in other regions with little to no admixture (Stringer and Andrews 1988; Lahr 1994).

These models predict that different patterns of morphological and genetic variation should be found in different regions of the world. The RAO model predicts that the very earliest anatomically modern fossils would first appear in Africa and archaic populations would be found throughout Eurasia, descendants of H. erectus or later archaics. Outside of Africa, there should be few alleles older than the origins of anatomically modern humans found in Eurasia, and a clinal decrease in genetic diversity should emanate from Africa. The MR and other models that posit regional continuity would predict various levels of ancient alleles found in different regions along with both transitional fossil forms and levels of morphological continuity in the major areas of the Old World. (Disotell 2012)

Until recently, genetic data favored the RAO model of complete replacement over the MR model. (Klein 2008)

* As of the publication of Disotell’s review (2012), more than 18,000 whole human mtDNA genomes have been published, and they all coalesce within the last 200,000 years with non-African lineages coalescing around 50-60 kya or more recently (e.g. Ingman et al. 2000; Kivisild et al 1999).
* Studies of modern paternally inherited Y chromosomes found a world-wide coalescene of the 11ancestor of all Y chromosomes in Africa about 150 kya (Hammer et al. 1998) and infer an expansion out of Africa round 44 kya (Underhill et al. 2000)
* Initial comparisons of Neanderthal and modern human mtDNA found no evidence for a Neanderthal contribution to the modern human genome (Krings et al. 1997; Serre et al. 2004).
* The lack of ancient mtDNA in modern humans today does not necessarily mean that Homo sapiens never interbred with Neanderthals.
  + It is possible for archaic mtDNA lineages to be lost to genetic drift or replaced by more recent lineages (Relethford 2001)
  + Lack of archaic mtDNA in the contemporary human population is consistent with at most 120 hybridization events over a 12,000-year period of potential contact between Neanderthals and Homo sapiens. (Currat and Excoffier 2004)

Since the sequencing of Neanderthal (Green et al. 2010) and Denisovan (Meyer et al. 2012) genomes and analyses of nuclear DNA, the strict RAO model with no introgression has largely fallen out of favor.

* 1-4% of modern non-African genomes are derived from admixture with Neanderthals. (Green et al. 2010)
  + Neanderthals are as closely related to a Chinese and Papuan individual as to a French individual, even though morphologically recognizable Neanderthals exist only in Europe and western Asia. Lack of a clinal decrease in relatedness to Neanderthals with distance from Europe suggests that the gene flow between Neanderthals and modern humans occurred before the divergence of European, East Asian, and Papuan populations. (Green et al. 2010)
* Melanesians were found to have up to 4.8% Denisovan ancestry with an additional component of Neanderthal ancestry, that is, up to 7.4% of their ancestry is derived from archaic hominins. (Reich et al. 2010)
  + Denisovan alleles are also found in aboriginal Australians, near Oceanic, Polynesian, Fijian, and east Indonesian populations but not in South Asia or East Asia. Denisovans contribute to about 3.5% of these modern human genomes. (Reich et al. 2011)
* Indirect studies of patterns of linkage disequilibrium in modern Africans found support for some introgression from archaic African lineages. (e.g. Plagnol and Wall 2006; Wall et al. 2009; Hammer et al. 2011; Lachance et al. 2012)
  + Up to 2% of modern African polymorphisms can be traced to an archaic African population that introgressed around 35,000 years ago. (Hammer et al. 2011)
  + Evidence of archaic introgression into modern hunter gatherers 20-80 kya was found in Pigmy, Hadza, and Sandawe populations in Africa (Lachance et al. 2012). The ancient alleles are inferred to have split from the modern lineage about 1.2 million years ago.
* The finding of almost equal levels of Neanderthal introgression in all Eurasians has been interpreted as evidence for a unique pulse of admixture in the Middle East between Neanderthals and the ancestors of Eurasians (Green et al. 2010; Sankararaman et al. 2012). The fact that Denisovan admixture had been first evidenced in Papua New Guineans suggested that admixture had occurred as a single pulse in Southeast Asia, after the separation of the ancestors of Oceanians and other Asians (Reich et al. 2010, 2011).
  + See Alves et al (2012) for alternative hypotheses of human dispersal and admixture with archaic populations.

Most recent findings:

* East Asians are more similar to the published Neanderthal genome sequence than are Europeans. (Meyer et al. 2012; Wall et al. 2013)
  + This implies there were at least 2 separate episodes of admixture between Neanderthals and modern humans
  + At least one episode of admixture must have occurred after the separation of the ancestors of modern Europeans and East Asians.
  + Rather than 2 distinct episodes of admixture, Wall et al. (2013) suggest that admixture took place over a protracted period 50-80 kya. The ancestors of Europeans experienced less admixture after the divergence from the ancestors of East Asians.
* Wall et al. (2013) also found significant Neanderthal admixture into the Maasai population of East Africa, probably because of secondary contact with a non-African Homo sapiens population, rather than admixture directly from Neanderthals.
  + This supports the Currat and Excoffier (2011) model that suggest low levels of introgression in Europe and Asia from distinct admixture events occurring beyond the Middle East, after the split of modern Europeans and Asians.