On the Origins of Man: Understanding the Last Two Million Years

Introduction:

In the past two decades, our understanding of the human genome has increased at remarkable rate. This expansion of our genetic knowledge promised to resolve longstanding questions regarding the origins and evolution of mankind (Eller, 2002). However, after nearly fifteen years of research, our understanding remains limited; genes have raised far more questions then they have answered. Two models, proposed nearly thirty years ago, still hold weight in the anthropological community: the Multiregional and the Replacement model of the evolution of mankind (Eller, 2002). A critical review of various studies concerning the two models and, specifically, of recent genetic evidence concerning the interaction of *Homo sapiens* and *Homo neanderthalensis* (Caramelli et al., 2003; Gibbons, 2001; Kahn & Gibbons, 1997; Krings et al., 2000; Krings et al., 1997; Ovchinnikov et al., 2000), will reveal that – while both models are far too simplistic – a pattern closer to Replacement is more likely.

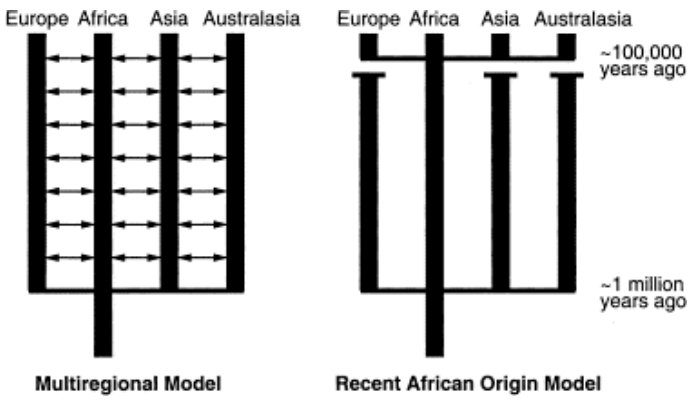
Defining the Two Models:

The Multiregional model of evolution, in its most basic from, argues that the since the original spread of *H. erectus* out of Africa, approximately 1.7-mya (million years ago), the 'human' population has been one large interbreeding mass (Eller, 2002). This population slowly evolved into what we consider modern humans with no side branching or distinct speciation events occurring in Eurasia, Australasia, or Africa (Eller, 2002; Templeton, 2002). Naturally, the model dictates that the morphological differences and geographical distances between all populations of humans were not enough to genetically isolate them and, thus, the modern human form evolved simultaneously across the entire old-world (Templeton, 2002). Milford Wolpoff, of the Univer-

sity of Michigan, originally proposed this model based solely upon archeological and morphological evidence (Lahr, 1994). Since the models incarnation, however, researchers have forwarded limited genetic evidence and some new morphological evidence to support the model (Hawks & Wolpoff, 2001; Templeton, 2002). Furthermore, research, done by Vinayak Eswaran, into population dynamics may also support a multiregional-like model – though Eswaran's model is more complex then Wolpoff's (Eswaran, 2002).

Contrary to Multiregionalism is the Replacement model. This model dictates that after *H. erectus'* initial expansion into the new world, a new form of hominids evolved in Africa, *H. sapiens* (sometimes written: *H. sapiens sapiens*) (Treisman, 1995). This new species – after migrating into Eurasia and Australasia approximately 100-kya (thousand years ago) – replaced *H. erectus* and *H. neanderthalensis* without inbreeding or hybridization (Eller, 2002). In this model, *H. neanderthalensis* evolved outside of Africa. The evidence from the Replacement model comes mostly from genetic analysis of modern human lineages and limited fossil evidence (Collard & Franchino, 2002; Jorde et al., 2000). Recently, genetic evidence from *H. neanderthalensis* and Cro-Magnon man (ancient presumed *H. sapiens* from around 25-kya) further supports the Replacement model (Caramelli et al., 2003; Krings et al., 2000; Krings et al., 1997; Ovchinnikov et al., 2000).

Fig 1: Multiregional vs. Replacement Model (Eller, 2002)



These two models are graphically represented in Figure 1, where the single line from Africa in the rightmost image represents *H. Sapiens*.

ARCHEOLOGICAL MULTIREGIONALISM:

The thrust of the evidence for the Multiregional model comes from comparisons of morphological differences between all known fossils of the last 2 million years and separate comparisons of their archeologically represented cultural traits – that is tool use and burial behaviors (Hawks & Wolpoff, 2001). John Hawks and Wolpoff believe that the morphological differences between the presumed "species" are insignificant variations – perhaps due to local climatic adaptation – and have not isolated the groups genetically (Hawks & Wolpoff, 2001). In fact, they content that these morphological differences are not large enough to advance a species difference between any hominid fossil since the emergence *H. erectus* (Hawks & Wolpoff, 2001). A review of one recent research endeavor concerning *H. neanderthalensis* and *H. sapiens* will show that the conclusions about morphological differences are not well founded, thereby, discrediting some claims held by the multiregionalists (Collard & Franchino, 2002; Kramer, Crummett, & Wolpoff, 2001).

A recent study, done by Andrew Kramer, claims to support Wolpoff's contention that *H. neanderthalensis* does not form a separate species. The study analyzes differences in cranial features between presumed *H. neanderthalensis* and anatomically modern humans (*H. Sapiens*), both found in the Levant region (Kramer et al., 2001). This region is thought to be the central hub of migrations between Eurasia and Africa (Walter et al., 2000). Using pairwise analysis differences of six fossils presumed to be anatomically modern humans (Skhul IV, V, & IX and Qafzeh III, VI, IX) and two fossils presumed to be Neandertals (Tabun and Amud) – all from Levant – Kramer concludes that morphological differences are not great enough to make the Neandertals a separate species in this region. In addition, they include thirteen other skulls from around the same time, but from different locations throughout Eurasia. These skulls are composed of six presumed Neandertals, six presumed modern humans, and one non-human primate

(KNM-ER 3733) – as an out-group – and are employed to test their methodology. Finally, they analyze twelve character traits using various subsets of these seventeen skulls with the program PAUP* 4.0 and create various phylogenies. (Kramer et al., 2001)

From their analysis of just the Levant fossils and the out-group fossil, they do not find a separate clade for the Neandertals and other Hominids of Levant (Kramer et al., 2001). However, an analysis of just the non-Levant male Neandertals, the non-Levant male modern humans, and the out-group reveals a *separate* clade for the Neandertals; this abnormality is brushed off as outside the aim of the research and, thus, insignificant. Furthermore, and more curious, the researchers never present or discus a PAUP* 4.0 analysis of *all* seventeen fossils, again their reasoning being that they are only concerned with the Levantine relationships. They then conclude that inbreeding was present at Levant and, therefore, possible throughout the old-world, thereby, supporting a multiregional inbreeding model. This conclusion is not easily reconciled with the data, which suggests that, even if only in Europe, two separate clades of Neandertals and modern humans existed (Kramer et al., 2001). Therefore, their research scarcely supports the multiregional hypothesis and may even support Replacement.

Furthermore, Mark Collard's research, in the use of pairwise difference analysis for morphological cranial features, suggests that the theoretical assumptions made by Kramer in the aforementioned study may be suspect (Collard & Franchino, 2002). Specifically, Collard proposes that the pairwise analysis – popular for DNA research – is not reliable for cranial morphology analysis. Specifically, Collard attempts to reconstruct the phylogenies of various extant primates groups (*Homo*, *Pan*, *Gorilla*, *Pongo*, and *Hylobates*) using craniodental and soft tissue features, many similar to the features used by Kramer (Collard & Franchino, 2002; Kramer et al., 2001). These groups have known phylogenies based on molecular data and other non-facial morpho-

logical differences (Boyd & Silk, 2003; Collard & Franchino, 2002). Collard attempts to make phylogenies based on: [1] 96 qualitative craniodental features, [2] 171 soft tissue characters (qualitative), and [3] 129 quantitative craniodental characters, all from two sets of 182 and 100 individuals (combination of all species) with at least 20 individuals from each species in the sets. This is a considerably larger data set then the one used by Kramer (Collard & Franchino, 2002; Kramer et al., 2001). *None* of the generated phylogenies agree with the known phylogenies of the extant primates (Collard & Franchino, 2002). However, Collard does not use PAUP* 4.0 for is analysis but a different program to create his trees. Thus, Collard's research may not completely invalidate the use of facial features, but it certainly cast doubt on the practical use of them.

Further evidence also casts doubt on the archeological basis for Multiregionalism (Lahr, 1994; Walter et al., 2000); however, there is still much debate on the conclusions presented above (Hawks & Wolpoff, 2001). In the next section, the genetic evidence presented will generally support these conclusions, which invalidate traditional Multiregionalism. However, the evidence will also suggest that a complete replacement is unlikely. Instead, a complex system with more aspects of Replacement, but some aspects Multiregionalism, is necessary.

GENETIC REPLACEMENT:

In contrast to Multiregionalism, the Replacement model relies heavily upon genetic evidence, owing its existence to a flawed mtDNA (mitochondrial DNA) analysis in 1987 (Templeton, 2002). Despite the models beginnings, much more evidence has arisen to support the theory (Jorde et al., 2000; Kimmel, 1999; Quintana-Murci et al., 1999; Templeton, 2002; Thomson, Pritchard, Shen, Oefner, & Feldman, 2000; Underhill et al., 2000). The DNA evidence falls fundamentally in two categories: [1] comparisons of modern human diversity and [2] comparisons

of ancient DNA Neandertals and Cro-Magnon man to modern humans (Jorde et al., 2000; Krings et al., 1997). For the time being, we will only concern ourselves with the modern human diversity argument, leaving the Neandertal evidence for later discussion.

The modern human diversity argument proposes that given the genetic diversity of modern man, a large bottleneck likely occurred sometime in the last 100,000 years (Eller, 2002). This bottleneck presumably represents a small population of *H. sapiens*, who evolved in Africa, replacing all other living hominid species (Eller, 2002; Kimmel, 1999). In many ways, the date of this bottleneck epitomizes the arguments between the two models: both agree that man arose in Africa, but disagree on the date of his diaspora and the effects of his diaspora on population dynamics (Wall, 2000). The theoretical reasoning behind the detection of the bottleneck lies in a mathematical function known as a Laplace transformation (Kimmel, 1999). While the actual workings of this function are unimportant, Marek Kimmel has observed that – despite the instability inherent in the equation – simulation studies have proven that it can effectively predict population size changes and, thus, be applied to detect signs of a population bottleneck.

Researchers have used several classes of DNA evidence (Y-Chromosome DNA, mtDNA and Autosomal DNA) to investigate the possibility of a bottleneck and, subsequently, have attempted to date any observed bottleneck event(s) (Jorde et al., 2000). All attempts at dating, however, have utilized the same theoretical assumption: They have all relied upon a stable mutation rate of their respective DNA, assuming "selective neutrality" of their DNA regions (Thomson et al., 2000). Thus, the researchers are able to predict a date of divergence by effectively counting the number of mutations in the present population and comparing them to the molecular clock of mutation rates. To set their clocks, the researchers must use DNA from the last known ancestor of the human, the chimpanzee, which diverged from modern man approximately 5 million years

ago (Thomson et al., 2000). Again, by comparing the overall differences between chimpanzee and human DNA they are able to establish a rate of mutation for whatever region they are studying; of course, this inherently assumes that the divergence date from *Pan troglodytes* is correct, this date being based on fossil evidence (Boyd & Silk, 2003).

Two recent studies using these methods have employed Y-chromosome DNA, and offer nearly identical dates if divergence (Thomson et al., 2000; Underhill et al., 2000), however they do not prove the Replacement model since so much theory is still in question (Hammer et al., 1998; Hawks & Wolpoff, 2001; Templeton, 2002; Thomson et al., 2000; Underhill et al., 2000; Wall, 2000).

The first researcher, Peter Underhill, analyzes 167 polymorphisms on a portion of NRY DNA (non-recombinant y-chromosome DNA) from 1062 individuals (Underhill et al., 2000). Underhill attempts to accurately represents the genetic diversity of the human population by choosing an ethnically diverse group – a rather common practice in studies of this nature (Jorde et al., 2000; Templeton, 2002; Thomson et al., 2000; Underhill et al., 2000). Underhill found that the largest amount of genetic diversity occurs in Africa and detected a bottleneck event approximately 59-kya. This estimate assumes 6,900 years between new mutations, with an average of 8.6 mutations in any population. Underhill accounts for the many sources of error with an upper and lower limit of 35-kya and 89-kya. This estimate is much more recent than the original 100-kya (Eller, 2002; Underhill et al., 2000).

The second researcher, Russell Thomson, analyzes three genes found on the Y-chromosome (SMCY, DBY & DFFRY) (Thomson et al., 2000). Combined the genes represent 64,120bp, show 56 unique (non-repeating) polymorphisms, and have an average calculated mutation rate of 1.24e-9bp per year. Extrapolating a phylogeny from this data set using GENETREE, Thomson

found a bottleneck event approximately 50-kya, with the most diverse population being African. This all suggests a modern African origin, with an expansion around 50-kya.

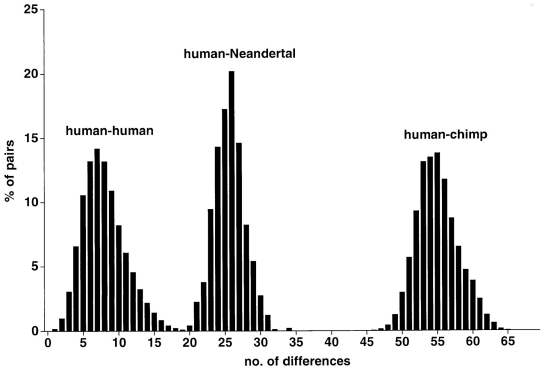
However, while both of these studies seem to support the Replacement model, there is still a considerable amount of further research needed. Both Underhill and Thomson have stated that the lack of genetic diversity on the Y-chromosome may be a result of selection pressure (Thomson et al., 2000; Underhill et al., 2000) and, therefore, invalidating their mutation rate estimates. Furthermore, Thomson recognizes that these date-estimates do not agree with other genetic evidence, such as the β–globin loci, which place the bottleneck at a much earlier date more in agreement with the Multiregional model (Templeton, 2002; Thomson et al., 2000). Conversely, Thomson and Underhill's estimates agree with other genetic evidence such as mtDNA and Autosomal DNA (Jorde et al., 2000). All this conflicting evidence remains to be thoroughly reconciled. However, the large body of genetic evidence seems to support to a pattern akin to the Replacement model.

NEW NEANDERTAL DNA :

Some of the problems associated with the genetic examination of human history lie in its inability to examine DNA from the extinct hominids (Hawks & Wolpoff, 2001). However, with new techniques, pioneered by Svante Paabo, this type of analysis has become possible (Krings et al., 1997). In the last six years, scientists have been able to reconstruct two partial genomes of *H. neanderthalensis* (Krings et al., 1997; Ovchinnikov et al., 2000). Analysis of these genomes has suggested to some that the Neandertal did not contribute to the modern day gene pool (Caramelli et al., 2003; Gibbons, 2001; Kahn & Gibbons, 1997; Krings et al., 2000; Krings et al., 1997; Ovchinnikov et al., 2000) and, thus, was an evolutionary dead-end. This would mean that modern humans replaced the Neandertal in Eurasia and Australasia.

Matthias Krings was the first to successfully study Neandertal DNA in 1997 (Krings et al., 1997). His analysis was of mtDNA of a Neandertal found in Feldhofer, Germany, which dated less then 100-ky old. For this study and all subsequent studies since mtDNA is used because, on average, per cell there are a significant number of copies of mtDNA, as opposed to the one nuclear copy of DNA (Krings et al., 1997; Ovchinnikov et al., 2000). Krings duplicated fragments of the mtDNA using PCR and obtained a 379bp fragment. The amplification process on ancient DNA (aDNA) has yet to be proven entirely reliable, but recent studies suggest that with the proper precautions a reliable sample can be obtained (Gilbert et al., 2003). Krings then proceeded to compare the sequence with that of modern humans and chimpanzees. He obtained an average difference of 8.0 ± 3.0 substitutions for a human-human comparison, 25.6 ± 2.2 substitutions for the human-Neandertal comparison, and 55.0 ± 3.0 for a human-chimpanzee relationship; this data is summarized in Figure 2. In addition, Krings found that the mtDNA of the Neandertal was no more closely related to the mtDNA of any regional group of modern humans and, thus, in disagreement with the Multiregional Model. This model predicts that European Neandertals should be closest in relation to modern Europeans, reflecting the assimilation of the Neandertal DNA by hybridization into local populations. Furthermore, he was able to reconstruct a phylogeny of humans, Neandertals, and chimps predicting a human-Neandertal split approximately 550-kya to 690-kya. He is careful to note that this phylogeny only compares one partial Neandertal mtDNA genome and, therefore, should be interpreted cautiously. (Krings et al., 1997)

Fig 2: Net Substitution Differences (Krings et al., 1997)



In 2000, Igor Ovchinnikov studied a second mtDNA sequence from a Neandertal, who dated at ~29-ky old (Ovchinnikov et al., 2000). Archeologists unearthed the Neandertal in the Mezmaiskaya cave in northern Caucasus – 2,500km from the Germen location, Feldhofer. Using the same precautions and general techniques of Krings, Ovchinnikov was able to extract a 345bp segment of mtDNA. Comparisons of this strand to the Feldhofer strand, modern humans, and chimpanzees revealed a parsimoniously phylogeny (with the lowest bootstrap number being 95/100), where Neandertals composed a separate species with a last common ancestor between 151-kya and 352-kya. This date, utilizing both the Feldhofer and Mezmaiskaya sample, is a more accurate prediction then the previous. As with Krings, Ovchinnikov stresses the same cautious interpretation of the data, but offers that the new evidence reinforces both the techniques and the conclusions of Krings. (Ovchinnikov et al., 2000)

David Caramelli has offers more evidence to support the conclusion that Neandertal and humans did not interbreed (Caramelli et al., 2003). Caramelli's research is unique to the previous studies because it utilizes ancient mtDNA from two Cro-Magnon men fossils (archaic *H. sapiens*) dating ~23-kya and ~24-kya and compares it with Neandertal mtDNA sequences. He found that the differences between the Cro-Magnon mtDNA and modern human DNA to be negligible: Cro-Magnon man falls well with in the range of modern human diversity. Moreover, Caramelli discovered that Neandertal mtDNA differed significantly from both the Cro-Magnon and modern human, so much so that unless a very abnormal selection event occurred these Neandertals never interbreed with neighboring Cro-Magnon and, thus, did not contribute to the modern gene pool. However, his data falls short of a proof given the nature of aDNA research. (Caramelli et al., 2003)

The process of extracting fifty-thousand year old DNA is no easy task. The complications and possible sources of contamination have been the main critics of the Neandertal finds (Gilbert et al., 2003). Only more research in the field will resolve the questions created by the research. However, the data suggests the there were at least two distinct species of hominids, suggesting a Replacement model pattern.

CONCLUSIONS:

The problem that lies in both models is the oversimplification of a complex evolutionary history. Understanding of selection pressures and varying mutations rates remains limited. Furthermore, neither of the two models in their present form can reconcile all the known data – both genetic and morphological. No one model has yet sufficiently explained the discrepancies between the various genetic results (mtDNA, Y-Chromosome, β-globin, etc.). The new Neandertal evidence seems to bolster Replacement, but given the available data, a more complex pattern replacement seems likely. Recent attempts to make models that are more complex have also fallen short (Eswaran, 2002; Excoffier, 2002). Erik Trinkaus, of Washington University, best summons up the current state of human origin theory:

> "I believe that it [Eswaran's model] suffers from a problem shared with the majority of the current and past models of modern human emergence: namely, it tries to explain too much of a geographically and temporally complex process with a single mechanism . . ." (Eswaran, 2002, 767).

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