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Article

Genesis and the Genome: Genomics Evidence for Human-Ape Common Ancestry and Ancestral Hominid Population Sizes

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The relatively new and rapidly expanding field of comparative genomics provides a wealth of data useful for testing the hypothesis that humans and other forms of life share common ancestry. Numerous independent lines of genomics evidence strongly support the hypothesis that our species shares a common ancestor with other primates. Additional lines of evidence also indicate that our species has maintained a population size of at least several thousand individuals since our speciation from the ancestors of other great apes. This article will provide an overview of genomics evidence for common ancestry and hominid population sizes, and briefly discuss the implications of these lines of evidence for scientific concordist approaches to the Genesis narratives.

Evolutionary theory has long proposed that humans and other great apes share common ancestors.[1] Evolutionary theory thus predicts that the genomes we observe in living primates (such as humans and chimpanzees) are, in fact, modified forms of an original genome present in the common ancestor of these species. This simple hypothesis can be readily tested using several independent lines of evidence derived from comparing the complete genomes of the two species.[2]

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The first line of evidence, and perhaps the one most widely discussed by Christian apologetics organizations, is that of gene sequence similarity. If, indeed, humans and chimpanzees are descended from a common ancestral species, then the individual gene sequences of these two species would be predicted to have a high degree of similarity due to inheritance from a common ancestor, or *homology*. Moreover, homology for individual genes should exist at two levels: the amino acid level (the functional sequence of a given gene's protein product), and at the nucleotide code level (the underlying DNA code for the required amino acid sequence). Since the nucleotide code has numerous coding options for a given amino acid sequence (i.e., the nucleotide code is *redundant*), genes in related organisms are predicted not only to share amino acid sequences but also nucleotide sequences,

despite a large number of possible coding options. Thus, related organisms should display homology at both levels of code.

A second, unrelated line of evidence is that of *synteny*. Synteny is a technical term for conservation of gene order along chromosomes between relatives. Put more simply, the hypothesis of common ancestry predicts that not only will related species have similar genes, but that they will also have these genes in a very similar spatial pattern.

A third line of evidence is that of *pseudogenes*. Pseudogenes (literally, "false genes") are the mutated remains of gene sequences that persist in the genome after their inactivation. Common ancestry predicts that related species should share pseudogenes that were present in the genome of their common ancestor. Moreover, these pseudogenes should be in the same genomic location in both descendant species (i.e., they should exhibit shared synteny) and retain gene sequence similarity (i.e., continue to exhibit homology) in spite of their inactivation.

The DNA sequence of the human genome was completed and published between 2001 and 2004.[3] Shortly thereafter, the genome sequence of the chimpanzee was completed.[4] The availability of complete genome sequences for both organisms allows for a comparison of homology, synteny, and shared pseudogenes at a genome-wide level for these two species. As such, these analyses function as independent tests of, and provide independent lines of evidence for, the hypothesis of human-chimpanzee common ancestry.

Sequence Similarities in Primate Genes: Evidence from Homology

Homology is defined as similarities derived from shared ancestry. It has long been known that humans and chimpanzees have nearly identical sequences for individual genes.[5] Complete genome sequencing has confirmed that this pattern of near identity is consistent across the genomes of both species. The human genome has approximately 3.0×10^9 nucleotides; of this number, 2.7×10^9 nucleotides match the chimpanzee genome with only a 1.23% difference between the species.[6]

In short, the vast majority of the human genome matches the chimpanzee genome with only rare differences. The inclusion of sequence alignment gaps between the two genomes that are thought to have arisen through either insertions or deletions (so-called "indel" mutations) drives the identity of the two genomes down to about 95%.[7] Restricting the comparison to the sequences responsible for coding for proteins raises the value to 99.4%.[8] By any measure, humans and chimpanzees have genomes that are highly homologous and readily interpreted as modified copies of an original ancestral genome.

Codon Usage in Homologous Genes: Evidence from Redundancy

The DNA code used to specify amino acids within proteins is based on nucleotide triplets, or "codons." Since there are four nucleotides (A, C, G, and T), there are 64 (i.e., 4^3) possible nucleotide triplets available; however, only twenty amino acids are present in biological proteins. Since three of the 64 codons are used as "stop" codons to halt the translation process, 61 codons are available for coding twenty amino acids. Thus, most amino acids can be encoded by more than one codon (i.e., the codon code is partially redundant). For example, a comparison of the nucleotide and amino acid sequences for insulin (a peptide hormone) of human, chimpanzee, gorilla, orangutan, a species of bat, and mouse is shown in figure 1.[9]

The unprocessed insulin peptide in all six species has 110 amino acids, the majority of which can be coded by alternate codons. This redundancy in the code means there are over 10^{19} different possible nucleotide sequences for human insulin that maintain the observed amino acid sequence. The sequence we observe, however, is one nearly identical to the nucleotide sequences seen in other mammals (figure 1A). The chimpanzee sequence differs by only six nucleotides; the gorilla, only by four. At the protein level, chimpanzees differ by two amino acids compared to humans, whereas the gorilla sequence is identical to ours (figure 1B). The amino acid and nucleotide homologies for other mammals become progressively less identical with the human sequence in a nested pattern that matches their phylogeny based on morphological criteria (figure 1C). While this is a very small sample (330 nucleotides), this pattern is representative: a genome-wide comparison of human and chimpanzee coding sequences reveals they are 99.4% identical across 1.85×10^7 nucleotides.[10]

Figure 1. Nucleotide and Amino Acid Homology for Insulin in Mammals

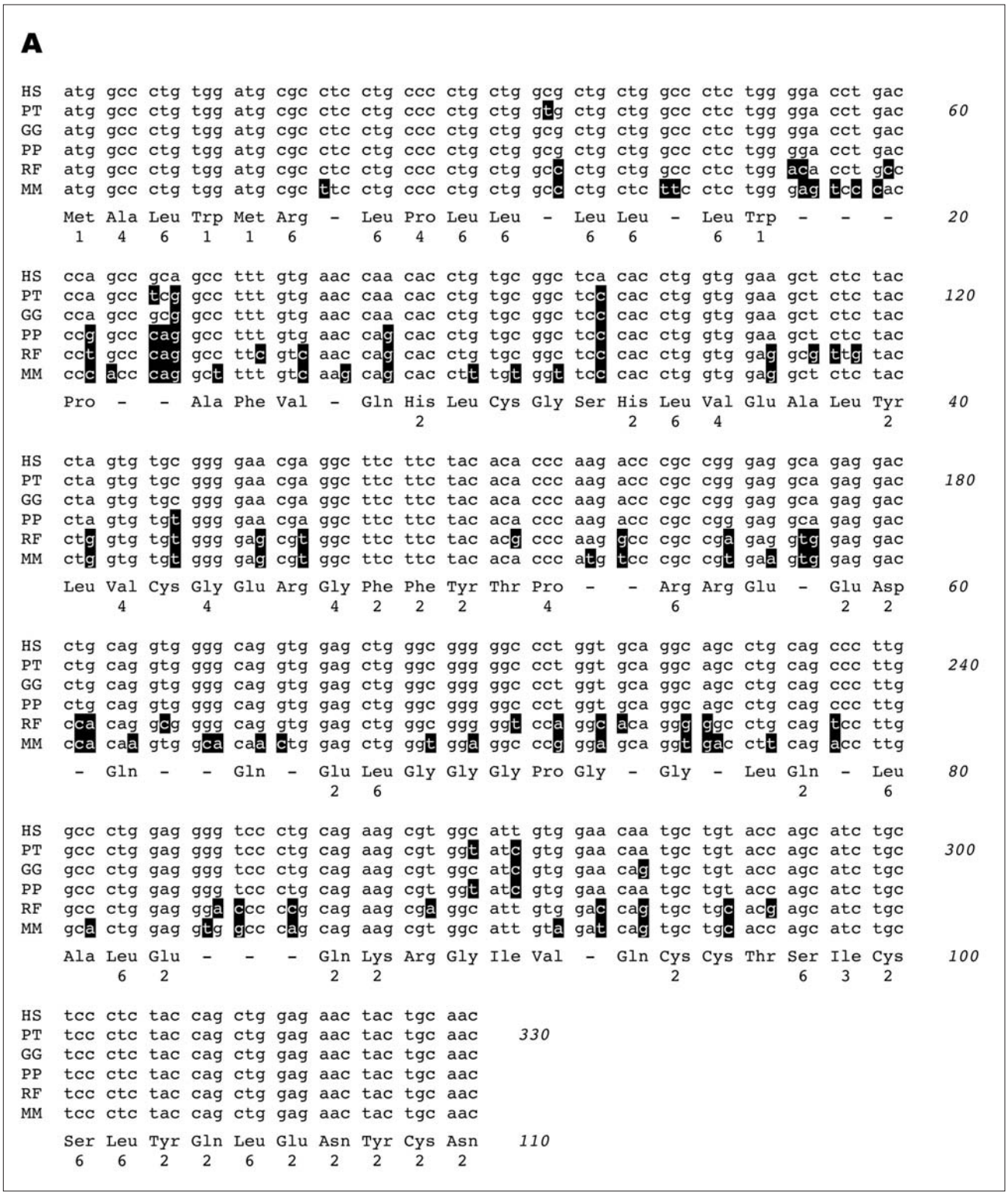

Figure 1A. The complete nucleotide coding sequence for pre-proinsulin aligned for four primate species (HS = *Homo sapiens* /human, PT = *Pan troglodytes*/chimpanzee, GG = *Gorilla gorilla*/gorilla, PP = *Pongo pygmaeus*/Bornean orangutan), one chiropteran (RF = *Rhinolophus ferrumequinum*/greater horseshoe bat) and one murid (MM = *Mus musculus*/mouse). Nucleotides that differ from the human sequence are shaded in black. Amino acids conserved in all six species are given below the nucleotide sequence. Numbers below codons conserved in all six species indicate the number of codon alternatives for that position.

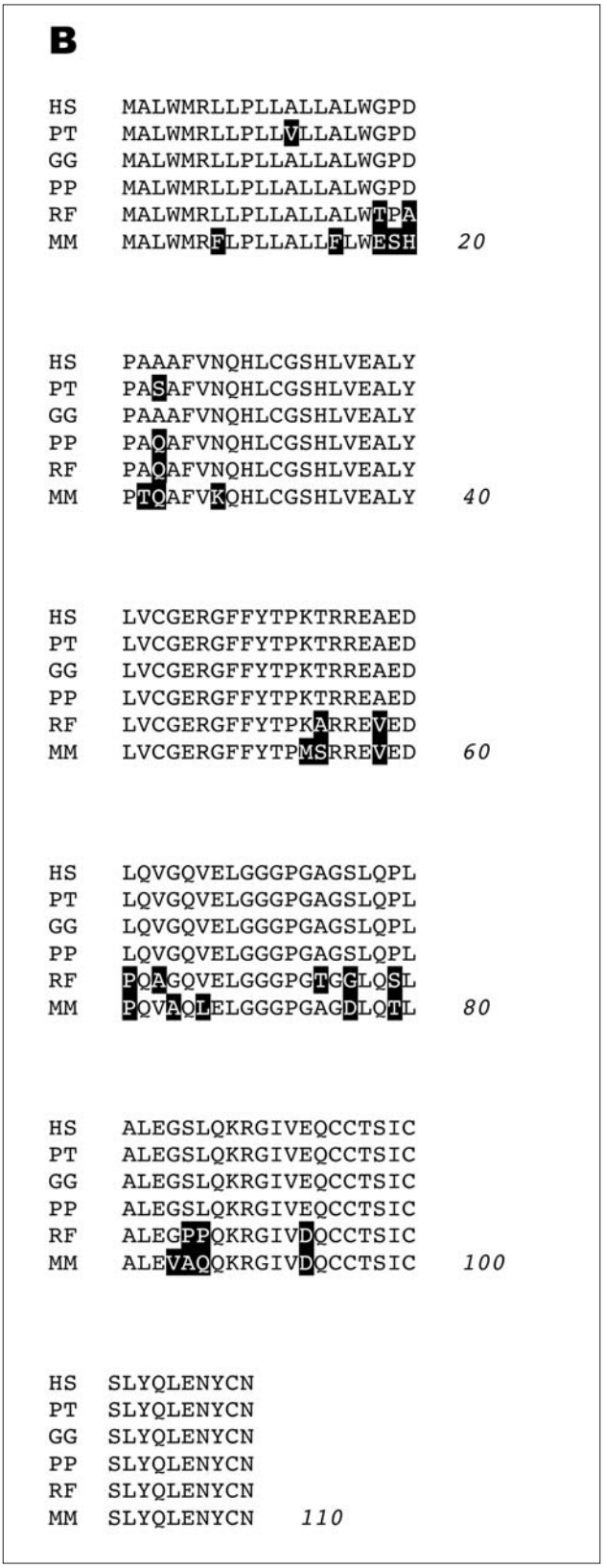

Figure 1B. The complete amino acid sequence of pre-proinsulin aligned for the same species as in (A). Amino acids that differ from the human sequence are shaded in black.

This argument can be extended to situations in which amino acid differences are observed in specific proteins between species. For example, the differences between human and chimpanzee insulin at the nucleic acid level are as small as possible despite the amino acid differences. The twelfth amino acid in chimpanzee insulin, for instance, is valine (codon GTG), whereas in the other mammals examined here (figures 1A, 1B), it is alanine (codons GCG or GCC). There are four codons that code for valine (GT followed by any of A, C, G, or T) and four that code for alanine (GC followed by any of A, C, G, or T). What we see when comparing this codon in humans and chimpanzees is the two closest possible codons despite the altered amino acid. Put another way, the nucleic acid code is consistent with only single nucleotide changes of a common ancestral sequence, even though there are multiple codon options for the different amino acids.

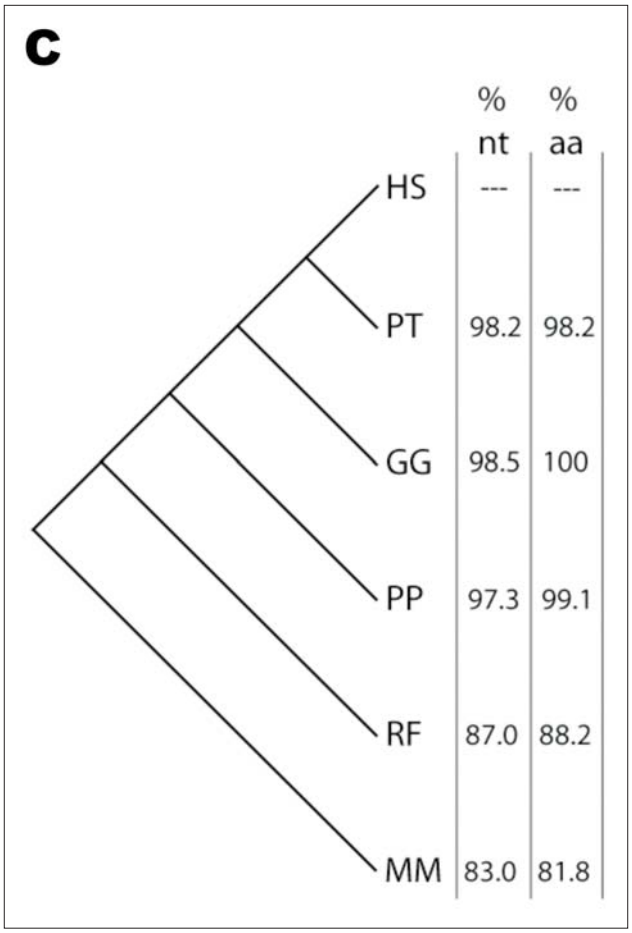

Figure 1C. Phylogeny for the same six species, with percent homology for pre-proinsulin relative to the human sequence shown for nucleotide (nt) and amino acid (aa) sequences.

Extending this type of analysis to other insulin sequences from organisms predicted to be less related to humans produces the same pattern: gorillas and orangutans use the same GCG codon for the alanine at the twelfth position, whereas bats and mice use a GCC codon for this alanine. This pattern persists across the entire coding sequence for insulin. Significant nucleic acid homology is retained despite the numerous options for the conserved amino acid sequence (figure 1C), and changes are highly consistent with single-nucleotide substitutions of an ancestral sequence (figure 1A). In summary, the observed pattern of gene homology across species is precisely what common ancestry predicts at two levels of code.

Genomic Spatial Organization: Evidence from Synteny

Synteny, in comparative genomics context, speaks to the observation that related organisms not only have high sequence homology for individual genes, but that the spatial organization of those genes is also similar. In short, organisms thought to be close evolutionary relatives have their genes in essentially the same order, with small differences arising from known mechanisms such as sequence inversions, translocations, and chromosome fusion events. As before, the hypothesis of common ancestry predicts such an outcome, since the two species in question are hypothesized to have once been a single species.

The fact that the human and chimpanzee genomes exhibit striking synteny with only subtle differences in genomic organization has been known for some time, based on chromosome staining and molecular hybridization techniques.[11] The main differences between human and chimpanzee chromosome sets are nine intrachromosomal inversions and one chromosome fusion.[12] These observations have now been confirmed at the molecular level by whole-genome sequencing of humans and chimpanzees.[13] Perhaps the best-known example of a difference between humans and chimpanzees with respect to genome organization is the telomere-to-telomere fusion resulting in human chromosome 2.[14] This chromosome corresponds to what are two separate chromosomes in chimpanzees and other great apes, suggesting that the human chromosome is the result of a fusion between what has persisted as two separate chromosomes in these other species. The evidence for the fusion event is based on synteny: the genes from the two ape chromosomes line up with the one human chromosome in the exact pattern one would expect from a tip-to-tip fusion event.

Synteny also predicts where certain byproducts of such a fusion event would be found. Chromosomes have special sequences called telomeres at their tips, as well as an internal sequence called a centromere that is used during cell division. Based on the two chromosomes we see in apes, we would predict *internal* telomere sequences where the human chromosome 2 sequence changes from aligning with the one ape chromosome to the other. We would also predict the presence of *two* centromeres that line up with the locations of those found in the ape chromosomes. In both cases, we find in human chromosome 2 exactly what common ancestry would predict: internal telomere sequences precisely at the expected fusion point, and the presence of two centromeres in their predicted locations, though one has been inactivated through accumulated mutations.[15]

In summary, when comparing the complete human and chimpanzee genomes, we observe that the spatial organization of genes in both species matches precisely what one would predict based on common ancestry: overwhelming similarity, with subtle differences arising since speciation.

Genomic Archaeology: Evidence from Pseudogenes

A third, and very compelling, line of evidence for common ancestry of humans and great apes comes from shared pseudogenes. *Pseudogenes* (literally, "false genes") are gene sequences that have been inactivated by mutation that persist in the genome as nonfunctional sequences. Pseudogenes remain recognizable for several reasons. First, only small changes are needed to inactivate a gene (for example, a change of one codon to an inappropriate "stop" codon, truncating protein translation). In such cases, the gene "remnants" are nearly identical to the functional gene and are readily identifiable by their homology. Secondly, comparative genomics allows us to identify pseudogenes not only by sequence homology to functional genes in other organisms, but also through synteny: pseudogenes retain their spatial orientation to neighboring functional genes after their inactivation. Thirdly, once inactivated, a pseudogene accumulates mutations only slowly,

because the proofreading mechanisms that govern DNA replication do not distinguish between functional and nonfunctional DNA sequences. These features allow for identification of pseudogenes in various states of disrepair as they are slowly mutated beyond recognition over millions of generations.[16]

Common ancestry also predicts that, beyond human-chimpanzee common ancestry, the common primate ancestor also shares ancestry with other vertebrates in the more distant past. For example, evolutionary theory predicts that humans, like all vertebrates, are descended from egg-laying ancestors.[17] As with all placental mammals, humans do not use egg yolk as a source of nutrition for their embryos. Other vertebrates such as fish and birds do employ egg yolk, as do a small number of extant mammals such as the platypus.

One protein used as a yolk component in egg-laying vertebrates is the product of the *vitellogenin* gene.[18] Since placental mammals are proposed to be descended from egg-laying ancestors, researchers recently investigated whether humans retained the remnants of the *vitellogenin* gene sequence in pseudogene form. To assist in their search, this group determined the location of the functional *vitellogenin* gene in the chicken genome, noted the identity of the genes flanking the *vitellogenin* sequence, and located these genes in the human genome. They found that these genes were present side-by-side and functional in the human genome; then they performed an examination of human sequence between them. As expected, the heavily mutated, pseudogenized sequence of the *vitellogenin* gene was present in the human genome at this precise location.[19] The human genome thus contains the mutated remains of a gene devoted to egg yolk formation in egg-laying vertebrates at the precise location predicted by shared synteny derived from common ancestry.

While the *vitellogenin* pseudogene is compelling, it is but one example of thousands that could be given.[20] For example, there are hundreds of genes used for the sense of smell (olfactory receptor genes) in the human genome that have become pseudogenes.[21] Moreover, many of these pseudogenes have identical inactivating mutations shared among humans, chimpanzees, and gorillas.[22] Furthermore, determining degrees of relatedness solely based on genomes that share identical inactivating mutations in olfactory receptor pseudogenes, independently arranges humans as most closely related to chimpanzees (most errors in common), and less so with gorillas (fewer errors in common), and even less with orangutans (fewer still errors in common).[23] Additionally, no "out of place" pseudogenes were found in this study: pseudogenes with identical inactivating mutations common to humans and gorillas were also present with the identical mutation in chimpanzees; mutations common to humans and orangutans were present in chimpanzees and gorillas.

This pattern is precisely what common ancestry predicts for these species, since an identical mutation present in two species is most readily explained by its presence in the common ancestor of both species. The common ancestor of humans and gorillas is also the common ancestor of chimpanzees, hence inactivating mutations present in humans and gorillas are also predicted to be present in chimpanzees. In short, the existence of shared pseudogenes between primate genomes, their syntenic locations, and their patterns of inactivation and distribution all coherently support the same model of common ancestry based on comparative sequence homology criteria alone.

Comparative Genomics: Evidence for Common Descent or Common Design?

While genomics evidence from homology, synteny, and pseudogeny independently supports the hypothesis that humans and chimpanzees share a common ancestor, it is also possible to assess these lines of evidence from an anticommon descent perspective such as intelligent design (ID). While it is true that a few individuals within the ID movement accept human-chimpanzee common ancestry,[24] this position appears to be a minority in the movement as a whole, which prefers an explanation of common design in lieu of common descent.[25] While a more complete treatment of these issues is beyond the scope of this article, a brief overview of genomics evidence from an anticommon descent framework is instructive in investigating the relative strengths and weaknesses of anticommon descent ID and standard evolutionary common ancestry as explanatory frameworks for primate comparative genomics data.

Homology, Redundancy, and Common Design

> Why couldn't the designer use some similar DNA and body structure for different organisms as well? Genetic similarity between chimps and humans does make sense from an evolutionary standpoint, but it is also consistent with intelligent design.[26]
>
> … designers often reuse part designs for different applications. If a designer wanted to generate a species similar to humans, it naturally follows that the designer would redeploy many of the same genes.[27]

It is perhaps reasonable to conclude that a designer may reuse parts to accomplish a similar design implementation (i.e., special creation) event. What we observe, however, is that human and chimpanzee genes match one another at the amino acid (i.e., functional) level as well as in their underlying nucleotide codes.

As we have seen, there is a vast array of nucleotide sequences available to a designer to encode a given amino acid sequence. Even if a designer were constrained by amino acid sequence in order to achieve protein functionality in similar organisms (which, in itself, is questionable, since nonhomologous enzymes can perform the same reaction), it would be easy for such a designer to choose alternate nucleotide codes to avoid the appearance of common ancestry. Yet what we observe, time and again, is that genetic codes in organisms thought to be close evolutionary relatives based on nongenetic criteria, match at the nucleotide as well as at the amino acid levels. This is precisely what common ancestry predicts, since the hypothesis is that similar organisms once were the same species with identical genomes. From an anticommon ancestry design perspective, this pattern is problematic. It suggests that the designer was unwilling (or worse, unable) to avoid the overwhelming appearance of shared ancestry when implementing design for what, in fact, are separately created organisms.

Synteny and Common Design

Discussions of synteny in the ID literature are few and unsubstantial. One example, in an attempt to rebut the conclusion that the signs of chromosomal fusion in human chromosome 2 support common ancestry, displays the basic arguments:

> … chromosomal fusion evidence simply strengthens the evidence for genetic similarity between chimps and humans. Since similarity could have been expected apart from Darwinism and common ancestry, similarities between organisms may just as easily be the result of functional requirements implemented via common design.[28]

This argument, as we have seen, evades the issue that synteny and homology are not necessarily to be expected together from a common design viewpoint.

Additionally, the ID literature does not mention that this prediction of a "shared synteny requirement" is not supported by evidence when comparing the genomes of other groups of highly similar organisms. For example, complete genome sequences of twelve fruit fly (Drosophilid) species are now available[29] and their genomic organizations have been compared.[30] The results of these analyses demonstrate that the Drosophilid body plan and biochemistry are well served by a wide array of synteny arrangements, with chromosomal rearrangements greatly more diverse in this group than that observed between humans and chimpanzees. Moreover, the size of genes held together in syntenic blocks between Drosophilid species is a function of their time since speciation based on molecular clocks. The more divergent the individual gene sequences are between two species, the fewer genes are retained in syntenic groups.[31] Put more simply, the designer seems to have employed a wide array of different genomic organizations for fruit flies, all of which provide appropriate biological function and Drosophilid morphology. The pattern of decreasing synteny matches the pattern of decreasing gene sequence homology as predicted by common descent. Therefore, it is easier to argue that various Drosophilid species are separate, independent designs than it is to argue that humans and chimpanzees are separate, independent designs, despite the fact that the fly species in question are difficult for a nonspecialist to distinguish by eye.

The problem with this line of ID argumentation is similar to what we have seen with redundancy. There is no a priori reason to expect a pattern of similar genomic organization (i.e., shared synteny) for humans and chimpanzees based on an anticommon descent design perspective. Moreover, there is every reason to predict a *very different* pattern, suggestive of independent special creation. Once again, synteny

evidence is not only strongly supportive of human-chimpanzee common ancestry, but also highly problematic for anticommon descent interpretations.

Pseudogenes and Common Design

Anticommon descent ID literature displays three common features with regard to pseudogenes: (1) conflation of pseudogenes with all noncoding DNA under the rubric "junk DNA"; (2) no discussion of the observation that pseudogenes with identical inactivating mutations are shared among organisms in the precise pattern predicted by common ancestry; and (3) the suggestion that pseudogenes have an as-of-yet undetermined function that explains their presence as the result of deliberate design.[32] The one positive argument, that of undetermined pseudogene function, does not address the many instances where a function is known for a given gene product. For example, the function of the *vitellogenin* gene in amniotic organisms is known, as is the function of the numerous olfactory receptors we observe as pseudogenes in humans and other primates.

Moreover, the ID literature does not address the fact that we observe these pseudogenes in the precise syntenic arrangement predicted by common ancestry. To accept the ID argument is to hold that the designer placed these sequences into the human genome in the precise syntenic location where we observe functional versions of these genes in other organisms, with highly homologous sequences that share apparent mutations in a nested hierarchy that matches phylogenies based on independent criteria, to perform an unrelated, as-of-yet unknown function. While such a possibility can never be absolutely ruled out, one wonders why the designer would choose a method of design that would give such a strong impression of common ancestry.

Common Design: A Theory in Crisis

In summary, homology, redundancy, synteny, and shared pseudogenes are independent lines of genomics-based evidence that converge on a single conclusion: humans are not biologically independent, *de novo* creations, but share common ancestry with other forms of life. Moreover, attempts to account for genomics evidence from an anticommon-ancestry ID, common-design viewpoint are enormously strained and severely ad hoc. While each line of evidence is individually problematic from an anticommon-descent, common-design standpoint, their combined, cohesive pattern is devastating.

Genomics and Ancestral Hominid Population Sizes: The Question of Adam and Eve

While much attention has focused on the implications of the human genome project for common ancestry with other primates, other advances in comparative human genomics have provided insight into other aspects of our biological past. One such area is the use of modern-day human genetic variation to estimate effective ancestral human population sizes at several time points in our evolutionary history.

The process for estimating population sizes from comparative genomics data is quantitative in nature,[33] and as such, it is less accessible to a nonspecialist audience. It is, however, possible to appreciate this data qualitatively as well as quantitatively. For example, a small, but significant, fraction of the human genome is more similar to the modern gorilla genome than to the chimpanzee genome.[34] For this subset of sequences, our *species* tree does not match the *gene* tree (figure 2).[35] This discordance is expected for closely related species that have diverged from each other in a short amount of time.[36] Put another way, the reason our genome is overwhelmingly more similar to the chimpanzee genome is that we most recently shared a common ancestor with chimpanzees. Yet, in spite of this, we retain some regions of our genome that are more closely related to gorillas. This situation arises because the population that gave rise to the human-chimpanzee common ancestor was large enough, and genetically diverse enough, to transmit this variation to us without passing it on to chimpanzees. Chimpanzees and humans are thus separate genomic samplings of a diverse ancestral population. Had this pool been small, the human-chimpanzee gene trees would match the species tree in almost every case. The proportion of gene trees that do not match the species tree can therefore be used to estimate the population size of the ancestral population.[37]

Early studies, using limited data sets, consistently estimated that the effective ancestral population size for *Homo sapiens* was in the range of 10,000 individuals, with the lower bound of the 90% confidence

interval in the 6,000 range.[38] This value, because it uses chimpanzees and/or gorillas as a comparison, is a measure of the effective population size of our lineage since speciation with chimpanzees (~4–6 million years ago) or gorillas (~6–9 million years ago).[39] The availability of the complete chimpanzee genome, as well as extensive sequences available from the ongoing gorilla genome project, have allowed for these ancestral population estimates to be made with increasing precision. Consistent with the older work, newer studies have returned estimates in the 8,000–10,000 range using very large data sets.[40]

Perhaps the most sophisticated study to date uses the completed human and chimpanzee genome sequences to assess alternative gene trees for sequences in situ within their human chromosome context (i.e., incorporating synteny).[41] This study, while agreeing with previous estimates, also shows that sequences with the alternative tree (i.e., human and gorilla sequences coalescing before human and chimpanzee) are grouped together in small blocks of synteny, as expected.[42]

Recent progress in examining genetic diversity solely *within* our species has provided a complementary means to estimate our ancestral effective population size, using assumptions independent of those used for cross-species, comparative-genomics approaches. The International HapMap Project is a large-scale effort to map and catalog human single nucleotide polymorphisms (SNPs).[43] While SNPs are like any other source of genetic variation when considered individually, when examined in groups linked together on the same chromosome, they can be used to estimate ancestral population dynamics using an effect called Linkage Disequilibrium (LD).[44]

SNPs linked far apart recombine easily during meiosis, but SNPs linked closely do not, and they tend to be inherited together. Comparing the frequency of individual SNP alleles with their patterns of linkage to other SNPs in the same population reveals that many SNP pairs are in LD: they show up linked to other SNP alleles more frequently than would be expected, based on a random distribution. The biological basis for LD is that SNP pairs are inherited from ancestors and spread through a population without being broken up: closely linked ones stay together longer, and more widely separated ones recombine at a faster rate. Thus, known recombination frequencies between SNPs and the distribution and proportions of SNP pairs in a population can be used to estimate population sizes.[45] Since recombination frequency is determined by the physical distance between SNP pairs, LD studies can be used to estimate population sizes over time in a way

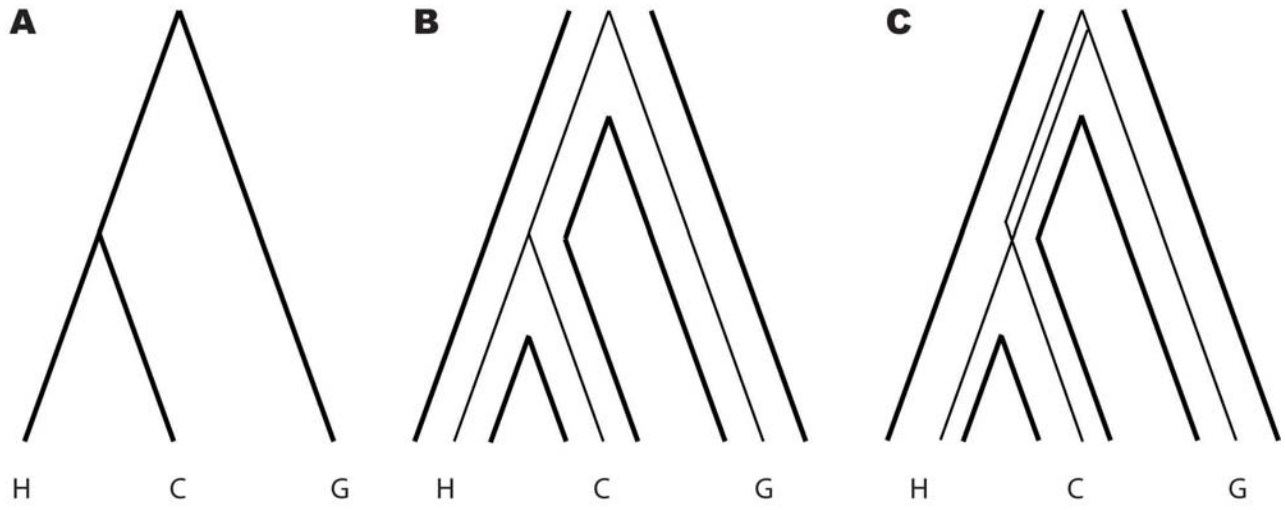

Figure 2. Species and Gene Trees for Human, Chimpanzee, and Gorilla

A. Comparative primate genomics strongly supports a primate species tree that groups humans (H) and chimpanzees (C) as more recently diverged relative to gorilla (G). Most genes in humans and chimpanzees coalesce before coalescence with gorilla (**B**); however, a minority coalesce first with gorilla (**C**). This alternative gene tree arises when variants of these genes were maintained in the human-chimpanzee common ancestral population after gorillas branch off (**C**). Accordingly, the proportion of genes in humans with a gene tree discordant with the species tree can be used to infer the effective population size of the lineage leading to humans from the present to the point of divergence with gorilla. See text for details.

that mutation-based estimates cannot. Selection of tightly linked markers allows for estimates in the deeper past, while more distantly linked SNPs (with their accordingly faster rates of recombination) are useful for more recent estimates. Also, since there are many thousands of SNP pairs to examine in the human genome, any sampled human population provides a multitude of data points for LD-based methods.

Studies based on SNP/LD approaches have now estimated ancestral population dynamics for various human groups over time in more detail than is possible with mutation-based estimates. African groups have a higher effective population size (~7,000) than do non-African groups (~3,000) over the last 200,000 years.[46] This approach, though based on methods and assumptions independent of previous work, nonetheless continues to support the conclusion that humans, as a species, are descended from an ancestral population of at least several thousand individuals. More importantly, the scalability of this approach reveals that there was no significant change in human population size at the time modern humans appeared in the fossil record (~200,000 years ago), or at the time of significant cultural and religious development at ~50,000 years ago.[47]

Taken individually and collectively, population genomics studies strongly suggest that our lineage has not experienced an extreme population bottleneck in the last nine million years or more (and thus not in any hominid, nor even an australopithecine species), and that any bottlenecks our lineage did experience were a reduction only to a population of several thousand breeding individuals. As such, the hypothesis that humans are genetically derived from a single ancestral pair in the recent past has no support from a genomics perspective, and, indeed, is counter to a large body of evidence.

What about Mitochondrial Eve and Y-Chromosomal Adam?

The genomics data presented above may appear to be at odds with the observation that human mitochondrial DNA coalesces to a common ancestor in the recent past (~170,000 years ago), and that human Y-chromosome sequences also coalesce to a common ancestor even more recently (~50,000 years ago).[48] This appearance of conflict, while commonly exploited in antievolutionary literature,[49] is in error. The reason for the rapid coalescence of mitochondrial and Y-chromosome sequences is that these DNA sequences are inherited in a manner distinct from (non-Y) chromosomal DNA. Mitochondrial DNA is passed only through mothers; Y chromosomes are passed only from father to son. As such, mitochondrial DNA lineages end abruptly if a mother has only sons; similarly, Y-chromosome lineages end abruptly if a father has only daughters. In both cases, however, non-Y chromosomal DNA lineages continue (i.e., fathers and mothers pass chromosomes to offspring of both genders).

Consider an extended family (figure 3). In this example, all females in the third generation derive their mitochondrial DNA from one common female ancestor in the first generation. Examining the females in generation three would produce the following results: their mitochondrial lineage would coalesce rapidly, but their chromosomal DNA lineage would not, since it is in part (50%) derived from two individuals in the second generation who are unrelated to the source of their mitochondrial DNA. Accordingly, variation in their genomic sequences would indicate that they are derived from a larger population that did not pass on its mitochondrial DNA to the present. In other words, it would be inappropriate to conclude that their matrilineal ancestor in the first generation was the only female present at that time, or that she lived at a time of a severe population bottleneck.

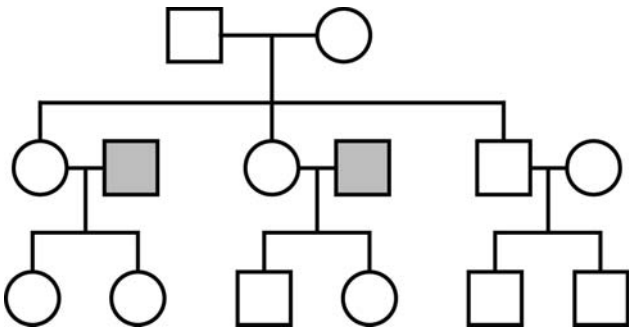

Figure 3. Mitochondrial and Chromosomal Inheritance in Humans

Squares indicate males, circles represent females. All females in the third generation have inherited their mitochondrial DNA from their common grandmother; however, they have inherited chromosomal DNA from their fathers as well (grey squares). As such, variation in their chromosomal DNA is the appropriate basis for estimating their population size.

So, too, for modern human populations. Though our mitochondrial DNA lineage coalesces to "Mitochondrial Eve" in the relatively recent past, present-day variation of human chromosomal DNA indicates that she was but one member of a substantial breeding population. The same logic, *mutatis mutandis*, applies to the inheritance of the Y-chromosome and the coalescence of human Y-chromosome variation to a single "Adam" in the recent past. While the rapid coalescence of these specially inherited DNA sequences is interesting in its own right, such sequences are not useful measures of ancestral human population sizes because of their unique modes of inheritance.[50]

Genesis and the Genome: "Ratcheting Concordism" or Divine Accommodation?

In summary, the expectation that the Genesis narratives provide scientific biological details of human ancestry fails in light of human genomics evidence on two fronts: humans share ancestry with other forms of life; and our speciation was through an interbreeding population, not an ancestral pair. As such, Christian "scientific concordist" approaches to Genesis are now under pressure from these lines of evidence.[51] The expectation that Genesis offers—at least at some level—scientific information, coupled with a view that science is a valid enterprise that provides an increasingly reliable understanding about the created order, produces a phenomenon I refer to as "ratcheting concordism." This approach is recognizable in that those who employ it, at first, resist the implications of new research that conflict with their concordist expectations, often deferring a decision on the claim of insufficient evidence. However, if contrary evidence continues to mount against their position, eventually such an individual may concede the point, discard the specific concordist expectation in question, and "ratchet" over to the next available position that retains the balance of their expectations. Considering the evidence presented here, one example might be a shift from denying common ancestry to its acceptance, while still retaining the expectation that our common ancestry was derived biologically through a single pair in the recent past.[52]

In contrast to a ratcheting concordist approach, an Evolutionary Creationist framework, such as that advanced recently in the works of Denis Lamoureux,[53] readily accepts and incorporates new scientific information. This view, in that it approaches the science of the Genesis narratives as divine accommodation to an Ancient Near-Eastern culture, has no expectation that Genesis will be in concord with modern science. While such a view may be criticized as a "low view" of Scripture, a ratcheting concordist approach is open to the same criticism, in that it postulates that only a *subset* of Genesis contains reliable scientific information. The implication for this approach, therefore, is that while Genesis is intended to convey scientific information, certain scientific features of Genesis are inaccurate or obscured due to accommodation. Evolutionary Creationism, in contrast, views the Genesis narratives as seamless documents of divine accommodation to their original audience, narratives that are written without intent to address modern scientific concerns. ◈

Notes

[1]C. Darwin, *The Descent of Man, and Selection in Relation to Sex* (New York: D. Appleton and Company, 1871).

[2]The Chimpanzee Sequencing and Analysis Consortium, "Initial Sequence of the Chimpanzee Genome and Comparison with the Human Genome," *Nature* 437 (2005): 69–87.

[3]International Human Genome Sequencing Consortium, "Initial Sequencing and Analysis of the Human Genome," *Nature* 409 (2001): 860–920; International Human Genome Sequencing Consortium, "Finishing the Euchromatic Sequence of the Human Genome," *Nature* 431 (2004): 931–45.

[4]The Chimpanzee Sequencing and Analysis Consortium, "Initial Sequence of the Chimpanzee Genome and Comparison with the Human Genome."

[5]M. C. King and A. C. Wilson, "Evolution at Two Levels in Humans and Chimpanzees," *Science* 188 (1975): 107–16.

[6]The Chimpanzee Sequencing and Analysis Consortium, "Initial Sequence of the Chimpanzee Genome and Comparison with the Human Genome"; R. J. Britten, "Divergence between Samples of Chimpanzee and Human DNA Sequences Is 5%, Counting Indels," *Proceedings of the National Academy of Sciences of the USA* 99 (2002): 13633–5.

[7]Ibid.

[8]R. Nielsen, C. Bustamante, A. G. Clark et al., "A Scan for Positively Selected Genes in the Genomes of Humans and Chimpanzees," *PLoS Biology* 3 (2005): e170.

[9]Tetrapod insulin sequences in Figure 1 were assembled from data accessed from National Center for Biotechnology Information public genome databases using BLAST searches (http://blast.ncbi.nlm.nih.gov/Blast.cgi).

[10]Nielsen, Bustamante, Clark et al., "A Scan for Positively Selected Genes in the Genomes of Humans and Chimpanzees."

[11]King and Wilson, "Evolution at Two Levels in Humans and Chimpanzees"; J. W. Ijdo, A. Baldini, D. C. Ward et al., "Origin of Human Chromosome 2: An Ancestral Telomere-

Telomere Fusion," *Proceedings of the National Academy of Sciences of the USA* 88 (1991): 9051–5; T. Ried, N. Arnold, D. C. Ward, and J. Wienberg, "Comparative High-Resolution Mapping of Human and Primate Chromosomes by Fluorescence in Situ Hybridization," *Genomics* 18 (1993): 381–6.

[12]H. Kerher-Sawatzki, B. Schreiner, S. Tanzer et al., "Molecular Characterization of the Pericentric Inversion That Causes Differences between Chimpanzee Chromosome 19 and Human Chromosome 17," *American Journal of Human Genetics* 71 (2002): 375–88; see also references therein.

[13]L. W. Hillier, T. A. Graves, R. S. Fulton et al., "Generation and Annotation of the DNA Sequences of Human Chromosomes 2 and 4," *Nature* 434 (2005): 724–31; The Chimpanzee Sequencing and Analysis Consortium, "Initial Sequence of the Chimpanzee Genome and Comparison with the Human Genome"; L. Feuk, J. R. MacDonald, T. Tang et al., "Discovery of Human Inversion Polymorphisms by Comparative Analysis of Human and Chimpanzee DNA Sequence Assemblies," *PLoS Genetics* 1 (2005): e56.

[14]Ijdo, Baldini, Ward et al., "Origin of Human Chromosome 2"; Hillier, Graves, Fulton et al., "Generation and Annotation of the DNA Sequences of Human Chromosomes 2 and 4"; The Chimpanzee Sequencing and Analysis Consortium, "Initial Sequence of the Chimpanzee Genome and Comparison with the Human Genome."

[15]Hillier, Graves, Fulton et al., "Generation and Annotation of the DNA Sequences of Human Chromosomes 2 and 4."

[16]While the mutation rate within pseudogenes *appears* faster than that observed in functional sequences (because purifying selection removes mutations from the population), it is, in fact, slow, in an absolute sense, due to proofreading by DNA polymerases during chromosome replication.

[17]D. Brawand, W. Wali, and H. Kaessmann, "Loss of Egg Yolk Genes in Mammals and the Origin of Lactation and Placentation," *PLoS Biology* 6 (2006): 0507–17.

[18]Ibid.

[19]Ibid.

[20]This article has limited the discussion of pseudogenes to *unitary* pseudogenes: sequences that lack a homologous sequence within the same genome, but are present at the expected syntenic area in functional form in other organisms. Indeed, if one considers repetitive elements, endogenous retrovirus insertions, processed pseudogenes, and so forth, examples could be multiplied many times over.

[21]T. Olender, D. Lancet, and D. W. Nebert, "Update on the Olfactory Receptor (OR) Gene Superfamily," *Human Genomics* 3 (2008): 87–97.

[22]Y. Gilad, O. Man, S. Paabo, and D. Lancet, "Human Specific Loss of Olfactory Receptor Genes," *Proceedings of the National Academy of Sciences of the USA* 100 (2003): 3324–7.

[23]Ibid.

[24]M. Behe, *The Edge of Evolution: The Search for the Limits of Darwinism* (New York: Free Press, 2007).

[25]For example, two recent, popular-level ID books attempt to cast doubt on human-chimpanzee common ancestry, and conflate common ancestry with "Darwinism": see W. A. Dembski and S. McDowell, *Understanding Intelligent Design: Everything You Need to Know in Plain Language* (Eugene, OR: Harvest House, 2008), 55–7 and C. Luskin and L. P. Gage, "A Reply to Francis Collins's Darwinian Arguments for Common Ancestry of Apes and Humans," in *Intelligent Design 101: Leading Experts Explain the Key Issues*, ed. H. W. House (Grand Rapids, MI: Kregel Publications, 2008), 215–35.

[26]Dembski and McDowell, *Understanding Intelligent Design: Everything You Need to Know in Plain Language*.

[27]C. Luskin, "Finding Intelligent Design in Nature," in *Intelligent Design 101: Leading Experts Explain the Key Issues*, 90.

[28]Luskin and Gage, "A Reply to Francis Collins's Darwinian Arguments for Common Ancestry of Apes and Humans," 221.

[29]*Drosophila* 12 Genomes Consortium, "Evolution of Genes and Genomes on the Drosophila Phylogeny," *Nature* 450 (2007): 203-18.

[30]S. W. Schaeffer, A. Bhutkar, B. F. McAllister et al., "Polytene Chromosomal Maps of 11 Drosophila Species: The Order of Genomic Scaffolds Inferred from Genetic and Physical Maps," *Genetics* 179 (2008): 1601–55; A. Bhutkar, S. W. Schaeffer, S. M. Russo et al., "Chromosomal Rearrangement Inferred from Comparisons of 12 Drosophila Genomes," *Genetics* 179 (2008): 1657–80.

[31]Ibid.

[32]For an example that features the key ID approaches, see Luskin and Gage, "A Reply to Francis Collins's Darwinian Arguments for Common Ancestry of Apes and Humans," 224–31.

[33]For a review, see N. A. Rosenberg and M. Nordborg, "Genealogical Trees, Coalescent Theory and the Analysis of Genetic Polymorphisms," *Nature Reviews Genetics* 3 (2002): 380–90.

[34]A. Hobolth, O. F. Christensen, T. Mailund, and M. H. Schierup, "Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model," *PLoS Genetics* 3 (2007): e7.

[35]Additional gene trees are possible, for example, where gene divergence occurs within an ancestral population prior to speciation. Also, additional factors such as hypermutability must be accounted for when estimating population sizes from discordant gene/species trees. Figure 2 is in part an adaptation and condensation of figure 1 in Holbolth, Christensen, Mailund, and Schierup, "Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model." For a more thorough discussion of these issues, see the complete article; and Rosenberg and Nordborg, "Genealogical Trees, Coalescent Theory and the Analysis of Genetic Polymorphisms."

[36]Rosenberg and Nordborg, "Genealogical Trees, Coalescent Theory and the Analysis of Genetic Polymorphisms"; Hobolth, Christensen, Mailund, and Schierup, "Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model."

[37]Ibid.

[38]For an example, see W. Li and L. A. Sadler, "Low Nucleotide Diversity in Man," *Genetics* 129 (1991): 513–23.

[39]Hobolth, Christensen, Mailund, and Schierup, "Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model."

[40]F. C. Chen and W. H. Li, "Genomic Divergences between Humans and Other Hominoids and the Effective Population Size of the Common Ancestor of Humans and Chimpanzees," *American Journal of Human Genetics* 68 (2001): 444–56; Z. H. Yang, "Likelihood and Bayes Estimation of Ancestral Population Sizes in Hominoids Using Data from Multiple Loci," *Genetics* 162 (2002): 1811–23; Z. Zhao, L. Jin, Y. Fu et al., "Worldwide DNA Sequence Variation in a 10-kilobase Noncoding Region on Human Chromosome 22," *Proceedings of the National Academy of Sciences of the USA* 97 (2000): 11354–8.

[41]Hobolth, Christensen, Mailund, and Schierup, "Genomic Relationships and Speciation Times of Human, Chimpanzee, and Gorilla Inferred from a Coalescent Hidden Markov Model."

[42]Ibid.

[43]See www.hapmap.org

[44]A. Tenesa, P. Navarro, B. J. Hayes et al. "Recent Human Effective Population Size Estimated from Linkage Disequilibrium," *Genome Research* 17 (2007): 520–6.

[45]Ibid.

[46]Ibid.

[47]Reasons to Believe maintains a literal Adam and Eve as progenitors of the entire human race and places them at ~50,000 years ago.

[48]M. Ingman, H. Kaessmann, S. Paabo, and U. Gyllensten, "Mitochondrial Genome Variation and the Origin of Modern Humans," *Nature* 408 (2000): 708–13; R. Thomson, J. K. Pritchard, P. Shen et al., "Recent Common Ancestry of Human Y Chromosomes: Evidence from DNA Sequence Data," *Proceedings of the National Academy of Sciences of the USA* 97 (2000): 7360–5.

[49]For example, see F. Rana and H. Ross, *Who Was Adam?* (Colorado Springs: Navpress, 2005), 123–31.

[50]F. Ayala, A. Escalante, C. O'Huigin, and J. Klein, "Molecular Genetics of Speciation and Human Origins," *Proceedings of the National Academy of Sciences of the USA* 91 (1994): 6787–94.

[51]D. Lamoureux, *Evolutionary Creation: A Christian Approach to Evolution* (Eugene, OR: Wipf & Stock, 2008).

[52]Denis Lamoureux (personal communication) has also collected anecdotal evidence of individuals ratcheting from one concordist position to another in light of new evidence. In his experience, the most prominent "positions" on the ratchet are Young-Earth Creationism; Old-Earth Creationism; Evolutionary Creationism that retains a literal Adam and Eve as biological progenitors of humanity (evolutionary monogenism); and Evolutionary Creationism proper (with no scientific concordist expectations of Genesis remaining). Further gradations are, of course, possible.

[53]D. Lamoureux, *Evolutionary Creation: A Christian Approach to Evolution*; ____, "Lessons From the Heavens: On Scripture, Science and Inerrancy," *Perspectives on Science and Christian Faith* 60 (2008): 4–15.