

HUMAN CHROMOSOME 2 FUSION FROM TWO CHIMPANZEE CHROMOSOMES: *DOES THE EVIDENCE SUPPORT THE CLAIM?*

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ABSTRACT

Since the discovery of chromosomes in the early twentieth century, evolutionary scientists have proposed that a major event in ape-to-human evolution occurred less than 10 million years ago through the telomere-to-telomere fusion of two ancestral ape chromosomes to form human chromosome 2. Although the idea that living organisms acquired their anatomical structures through natural processes dates back to Aristotle, Charles Darwin articulated the central tenets of evolutionary theory. He proposed that natural selection directed changes within species and that anatomical similarities between humans and animals provided evidence that humans descended from a lower form of life, with the apes considered their closest living relatives because of their anatomical similarities to humans. Using chromosome-banding techniques, researchers compared human and ape chromosomes and observed that chimpanzee chromosomes 12 and 13 displayed banding patterns similar to those of human chromosome 2 when arranged end to end. These observations were presented as additional evidence that humans and chimpanzees share a common ancestor. However, subsequent genetic sequencing and comparative analyses of human and ape chromosomes have raised questions about this hypothesis. The genomic evidence examined in this paper challenges the claim that human chromosome 2 originated through the fusion of two ancestral ape chromosomes and permits alternative explanations of human origins to be considered.

Keywords: human chromosome 2, evolution, chimpanzee, ape, banding, telomere.

Introduction

Modern evolutionary consensus holds that chimpanzees are the closest living relatives of humans, based partly on similarities in their karyotypes. Before the role of chromosomes in heredity was understood, evolutionary scientists relied heavily on homologous anatomical structures as evidence of a relationship between humans and apes. Human chromosome-banding techniques were standardized during the Paris Conference of 1971, enabling researchers to compare human chromosomes more systematically with those of other primates (Hamerton et al., 1972; Miller, 1977).

Using these banding techniques, researchers proposed that two ancestral acrocentric chromosomes corresponding to chimpanzee chromosomes 2A and 2B fused to form human chromosome 2. Under this hypothesis, the ancestral chromosome number was reduced from 48 to 46, and the fused chromosome subsequently became fixed in the human population through a transition from the heterozygous to the homozygous state (Fan et al., 2002; Miller, 1977; Stankiewicz, 2016; Yunis et al., 1980).

Miller (1977) proposed that speciation among primates, including humans, occurred over long time intervals through a series of inversions or translocations involving chromosomal satellite DNA. Yunis et al. (1980) analyzed human and chimpanzee chromosomes using standardized G-banding techniques and concluded that the chromosomes of the two species exhibited a high degree of homology, as had previously been suspected.

Yunis and Prakash (1982) extended these comparisons to the chromosomes of humans, chimpanzees, gorillas, and orangutans by photographing G-banded chromosomes during late prophase in each of the four species. They concluded that humans, chimpanzees, and gorillas shared a common ancestor, whereas orangutans represented an earlier-diverging lineage. The proposed chromosome 2 fusion has been estimated to have occurred approximately 0.9 million years ago (MYA), although other researchers have placed the event at approximately 5–6 MYA (Poszewiecka et al., 2022; Yoo et al., 2025).

Background of Evolution

Aristotle (ca. 350 B.C.) proposed that body parts were not formed for specific functions but arose accidentally through natural processes, with advantageous structures being preserved (Darwin, 1859). Organisms whose structures were not preserved in this manner perished. Darwin later described this principle as “natural selection,” thereby articulating the theory of evolution in a systematic form. In the introduction to his influential work, *On the Origin of Species by Means of Natural Selection, or the Preservation of Favoured Races in the Struggle for Life*, Darwin argued that the study of domesticated animals and cultivated plants demonstrated the possibility of substantial hereditary modification through the accumulation of slight variations under artificial selection. He reasoned that comparable modifications could also arise through natural processes without human intervention (Darwin, 1859).

In his subsequent volume, *The Descent of Man, and Selection in Relation to Sex*, Darwin argued that evidence for human descent from lower forms of life included homologous structures shared by humans and other species. These similarities included patterns of development, rudimentary structures, muscles, sensory organs, hair, bones, and reproductive organs. He further proposed that, just as other species had developed subspecies through natural selection, human populations had developed what he described as subraces through similar mechanisms (Darwin, 1871).

The principal evidence advanced for the ape-to-human theory was that the great apes were considered anatomically more similar to humans than were other animals, despite the substantial differences identified by anatomists during Darwin’s lifetime. These distinctively human features were presumed to have arisen gradually, with upright posture and bipedal locomotion regarded as especially significant. Following Darwin’s ideas, twentieth-century biologists interpreted similarities among fossils classified as Neanderthals, *Australopithecus*, *Homo habilis*, *Homo erectus*, and *Homo sapiens* as evidence of an evolutionary relationship between humans and the great apes (Simpson, 1966).

Simpson (1966) theorized that humans shared a common ancestor with chimpanzees and gorillas and that the human lineage became distinct through changes involving bipedalism, upright posture, and dentition. Other major characteristics associated with human evolution included tool

manufacture and language. Humans constitute a single biological species and can reproduce with other humans regardless of population or racial classification (Simpson, 1966).

During the late twentieth and early twenty-first centuries, additional evolutionary frameworks were developed or expanded to explain patterns and mechanisms of biological change. Two prominent frameworks are the modern synthesis and punctuated equilibrium. The modern synthesis, also known as neo-Darwinism or the evolutionary synthesis, integrates Darwinian natural selection with Mendelian genetics. It explains evolutionary change through processes that include mutation, recombination, genetic drift, gene flow, and natural selection acting within populations over successive generations.

Punctuated equilibrium proposes that species may remain relatively stable for long intervals, with substantial evolutionary change occurring during comparatively brief periods associated with speciation. This framework was developed partly to explain apparent patterns of prolonged morphological stasis and comparatively abrupt change in the fossil record (Hancock et al., 2021).

Human chromosomes were first observed microscopically during the late nineteenth century. Decades later, improved cytological techniques established that humans normally possess 46 chromosomes. Subsequent technical advances, including chromosome-banding methods and improved analysis of metaphase chromosomes, enabled researchers to compare human chromosomes with those of other primates in greater detail (Liehr, 2014; Miller, 1977).

The Data

Early chromosome-banding studies using metaphase chromosomes suggested that humans were more closely related to gorillas than to chimpanzees. Later high-resolution studies of late-prophase chromosomes, however, supported a closer evolutionary relationship between humans and chimpanzees (Miller, 1977; Yunis & Prakash, 1982).

Yunis et al. (1980) conducted a side-by-side comparative analysis of G-banded chromosomes obtained during late prophase, prometaphase, and early metaphase from six chimpanzees—four females and two males—and 24 humans—seven females and 17 males. Using high-resolution microscopy at $\times 1,600$ magnification, the researchers compared banding patterns, band thickness, and staining intensity. They reported striking homology between the two species in virtually all nonheterochromatic subbands. Their technique also permitted the precise localization of observed structural differences and revealed two previously unidentified centromeric inversions: a large inversion involving chromosome 9 and a smaller inversion involving chromosome 1. The researchers concluded that most chromosomal differences were confined to heterochromatic regions and were of little apparent consequence (Yunis et al., 1980).

In a subsequent study, Yunis and Prakash (1982) compared high-resolution, G-banded late-prophase chromosomes from humans, chimpanzees, gorillas, and orangutans using a methotrexate-based cell-synchronization technique. They proposed a sequence of evolutionary divergence among these presumed descendants of a common hominoid ancestor. Their samples consisted of cultured lymphocytes from five orangutans—two males and three females—five gorillas—one male and four females—nine chimpanzees—four males and five females—and 31 humans—10 females and 21 males.

Through side-by-side comparisons, the researchers reported remarkable similarities in chromosome-banding patterns among the four species. They attributed the observed differences primarily to inversions of chromosomal segments and variations in heterochromatin. They also compared the chromosomes of each species with a reconstructed “common hominoid ancestor” and reported that 18 pairs of modern human chromosomes resembled the proposed ancestral arrangement, whereas 15 chromosome pairs in chimpanzees, gorillas, and orangutans resembled that arrangement (Yunis & Prakash, 1982, p. 1528).

The researchers interpreted these comparisons as evidence of general chromosomal homology. They proposed that the orangutan lineage diverged first, forming the subfamily Ponginae, while gorillas, chimpanzees, and humans remained within the subfamily Homininae. Within the latter group, they hypothesized that the gorilla lineage diverged first, followed by the chimpanzee and human lineages (Yunis & Prakash, 1982).

Yunis and Prakash (1982) further stated that the observed chromosomal differences could best be explained through a series of “obligatory” intermediate steps, although those intermediate stages were not described in detail (p. 1528). Both Yunis et al. (1980) and Yunis and Prakash (1982) presented schematic, side-by-side representations of human and ape chromosomes. In these illustrations, one of the two ape chromosomes proposed to correspond to human chromosome 2 was shown in an inverted orientation, with its p arm directed downward (Yunis et al., 1980; Yunis & Prakash, 1982).

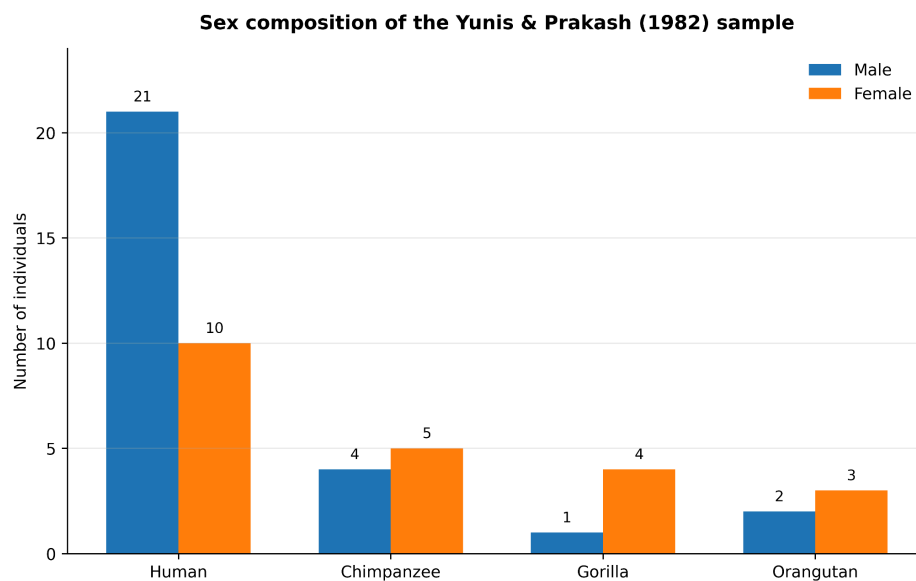


Figure 1. Comparative sample sizes reported in two high-resolution cytogenetic studies. Yunis and Sawyer (1980) examined 24 humans and six chimpanzees. Yunis and Prakash (1982) examined 31 humans, nine chimpanzees, five gorillas, and five orangutans.

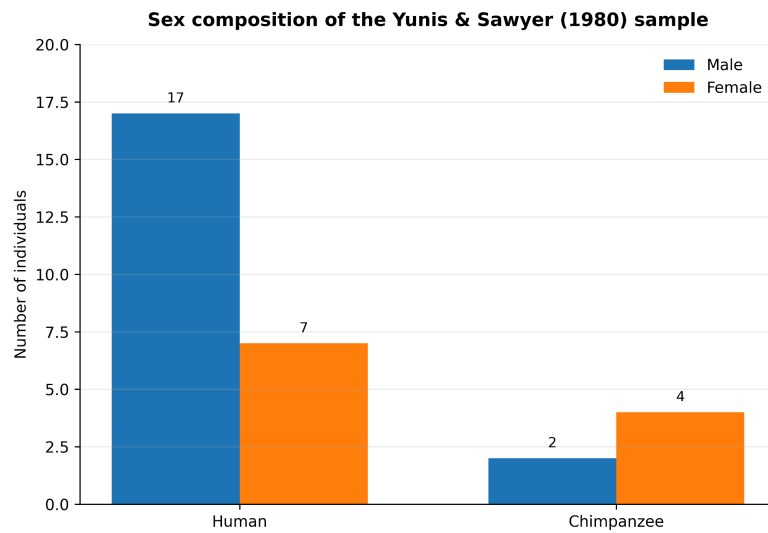


Figure 2. Sex composition of the Yunis and Sawyer (1980) study sample. The sample included 17 male and seven female humans, and two male and four female Chimpanzees.

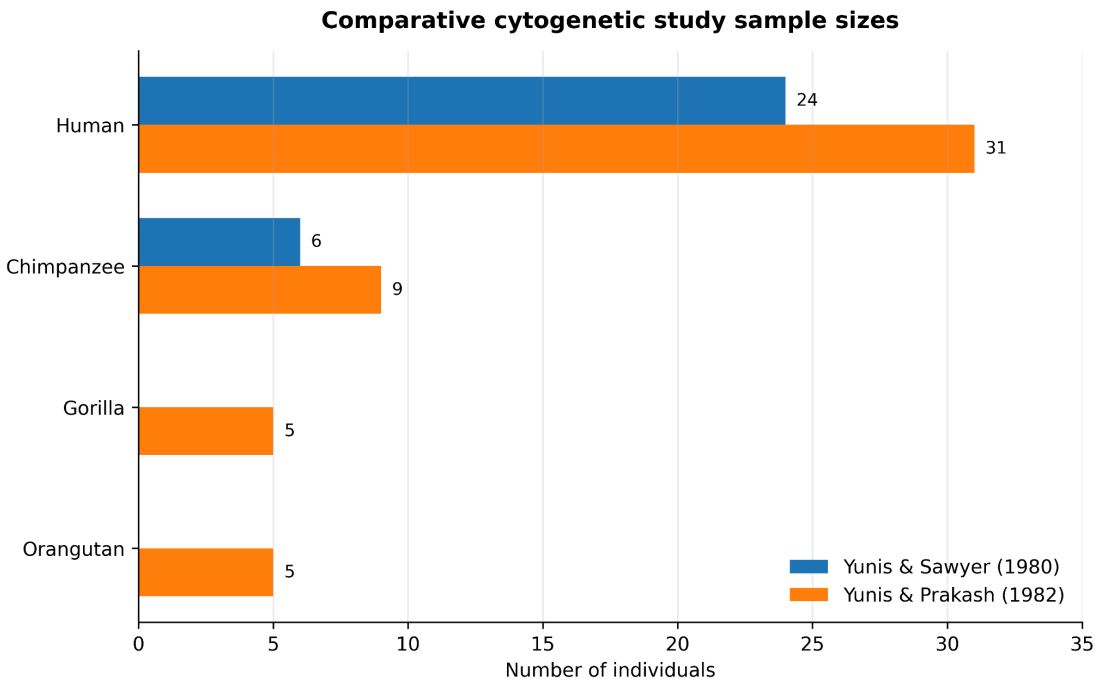
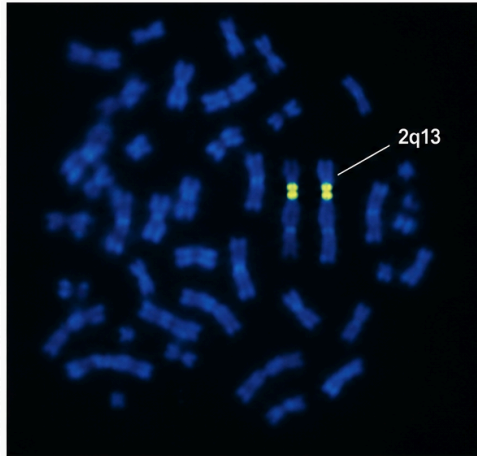


Figure 3. Sex composition of the Yunis and Prakash (1982) study sample. The sample included 21 male and ten female humans, four male and five female chimpanzees, one male and four female gorillas, and two male and three female orangutans.

Ijdo et al. (1991) studied telomeric repeat sequences in human chromosome 2 at band 2q13 using fluorescence in situ hybridization (FISH) on chromosome samples from six unrelated individuals. The researchers identified a degenerate head-to-head arrangement involving two cloned fragments in this region. However, they acknowledged that telomere fusion is a rare occurrence and is usually associated with certain tumors. They identified only 269 bp of tandem TTAGGG and CCCTAA repeat sequences. The researchers reported that telomeres normally form a protective buffer that prevents chromosomes from fusing, but they suggested that a small duplication and inversion event may have arisen by chance (Ijdo et al., 1991).

Ijdo et al. (1991): Telomeric Repeat Sequences at Human Chromosome 2q13

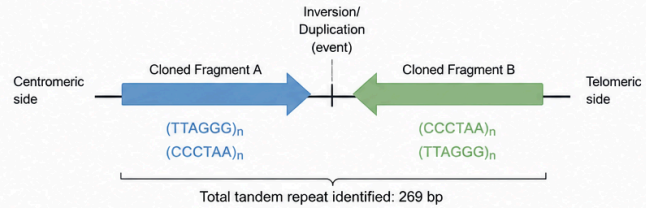
A FISH localization of telomeric repeats at chromosome 2 band 2q13



■ Telomeric repeat probe (TTAGGG)_n
■ Chromosomes (DAPI)

FISH analysis of metaphase chromosomes from six unrelated individuals showed hybridization of telomeric repeat probes at band 2q13 on chromosome 2.

B Degenerate head-to-head arrangement of cloned fragments at 2q13



C Key findings and interpretations

Aspect	Findings / Interpretation
Samples	Chromosome samples from six unrelated individuals
Method	Fluorescence in situ hybridization (FISH)
Location	Human chromosome 2, band 2q13
Arrangement	Degenerate head-to-head arrangement involving two cloned fragments
Tandem repeats identified	269 bp of tandem TTAGGG and CCCTAA repeat sequences
Interpretation	Telomeres normally form a protective buffer that prevents chromosomes from fusing. Telomere fusion is rare and usually associated with certain tumors.
Proposed mechanism	A small duplication and inversion event may have arisen by chance.

Note: Telomere fusion is a rare occurrence and is usually associated with certain tumors.

Another study found that head-to-head repeat arrays at fusion sites were highly degenerate compared with the intact TTAGGG repeats found at normal telomeres. These arrays were similar to interstitial arrays of degenerate telomeric repeats commonly found in subtelomeric regions of the human genome (Fan et al., 2002). The median telomere length in the diploid HG002 cell line, a high-quality human reference genome, is approximately 4.6 kb (Karimian et al., 2024). Because the proposed fusion of human chromosome 2 is unique to humans and fixed in the human population, Fan et al. (2002) theorized that the fusion must have occurred after the human–chimpanzee split but before modern humans dispersed around the world.

Poszewiecka et al. (2022) proposed a revised estimate for the timing of the hypothetical fusion event. Using genomic data from the Unexpected Bias Clustered Substitutions (UBCS) Genome Browser, the researchers compared modern human and chimpanzee genomes. They estimated that the fusion occurred approximately 0.9 million years ago (MYA), with a 95% confidence interval of 0.4–1.5 MYA. To obtain this estimate, the researchers assumed a constant evolutionary process leading to the accumulation of biased clustered substitutions near the telomeres of both species (Poszewiecka et al., 2022).

Because of the high structural complexity of the genomic region flanking the proposed fusion site, Stankiewicz (2016) suggested that human chromosome 2 “(HSA2) arose only once likely in one early modern human male and was subsequently transmitted and accumulated as a heterozygous event, converted to the homozygous state due to inbreeding in a small ‘bottleneck’ polygamous clan..., and spread in the succeeding populations” (p. 2). Of note, repetitive satellite DNA usually found in fused chromosomes is absent from the human genome (Stankiewicz, 2016).

Fan et al. (2002) conducted FISH analyses of human, chimpanzee, gorilla, and orangutan chromosomes using the bacterial artificial chromosome (BAC) clones RP11-480C16, RP11-395L14, and RP11-432G15, which were derived from the human chromosome 2 fusion region at 2q13–q14.1, referred to as 2qFus. Their findings confirmed earlier work demonstrating the centromere-to-telomere orientation of the constituent 2qFus clones on chimpanzee chromosomes. As Fan et al. (2002) stated, “Chimpanzee chromosomes 12 and 13 are homologous to human 2p and 2q, respectively” (p. 1656). The average guanine–cytosine (GC) content of the 614-kb sequence within the 2q fusion region is 44%. Both the GC content and the density and composition of repetitive elements vary across the sequence, with interspersed repeats occupying approximately 40% of the region (Fan et al., 2002).

In a review of the literature, Suntsova and Buzdin (2020) identified numerous differences between human and chimpanzee chromosomes. In earlier studies, observations of banding patterns in human and ape chromosomes were used to hypothesize that human chromosome 2 originated through the fusion of two acrocentric chromosomes found separately in chimpanzees (Suntsova & Buzdin, 2020). Fan et al. (2002) compared the fusion region in BAC RP11-395L14 with a 1,873-bp sequence obtained from a different individual and previously reported by Ijdo et al. (1991). They found that perfect TTAGGG or TTGGGG telomeric repeats were distributed intermittently across the fusion region.

Later studies employing methods such as genome sequencing and DNA hybridization arrays reported notable differences between human and chimpanzee genomes. For example, most chimpanzee chromosomes contain subterminal constitutive heterochromatin blocks (SCBs) composed of subterminal satellite (StSat) repeats that are absent from human chromosomes (Suntsova & Buzdin, 2020). Satellite DNAs (satDNAs), also known as tandem repeats, are major components of centromeric, pericentromeric, and telomeric regions in eukaryotic genomes. Cellular processes such as the cell cycle, gene expression, and genome stability are influenced by the organization of satDNA sequences (Lopes et al., 2021).

Three families of satDNA are present in all human acrocentric chromosomes, whose centromeres are located near one end of the chromosome, chromosomes 13, 14, 15, 21, and 22, as well as in chromosomes 3 and 4 (Liehr, 2014; Lopes et al., 2021). SatDNA II repeats are also present to a limited extent in the pericentromeric regions of chromosomes 2 and 10 (Lopes et al., 2021).

Ventura et al. (2012) performed a quantitative analysis of copy-number variation in the subterminal cap regions of chimpanzee, gorilla, and human chromosomes using read-depth-based copy-number predictions. They estimated the median copy number of hyperexpanded segments within the subterminal caps to be 798 in chimpanzees for chromosomes 2a and 2b, 1,092 in gorillas for chromosome 10, and fewer than 14 copies in humans for chromosomes 2 and 10 (Ventura et al., 2012). Martin et al. (2002) conducted a FISH analysis of subtelomeric regions in human, chimpanzee, gorilla, orangutan, macaque, and baboon chromosomes. They tested human probes derived from chromosomes 9p and 22q on primate metaphase cells and observed strong hybridization signals from BAC CITB-HSP-305J7 at the 9p telomere and the pericentromeric region of chromosome 9, as well as a weaker hybridization signal at the 2q13 region (Martin et al., 2002). In Figure 4 of that article, which is not reproduced in the present paper, the researchers identified 2q13 as an ancestral subtelomeric region; however, the region pictured appears to be centromeric.

Robertsonian translocations are fusions between two acrocentric chromosomes and most commonly involve human chromosomes 13, 14, 15, 21, and 22. These translocations occur when breaks arise near the centromeres of two chromosomes and their long arms fuse to form a single chromosome with one functional centromere. The short arms may also fuse to form a smaller reciprocal chromosome that is subsequently lost during cell division, thereby reducing the total chromosome number. Robertsonian translocations occur in approximately 1 in 1,000 newborns. Because the lost chromosome may contain relatively few essential genes, the translocation may produce no adverse effects in the carrier (Song et al., 2016).

Hackett et al. (2001) examined the mutation rate of the *CAN1* gene in *Saccharomyces cerevisiae* in the absence of telomerase, a ribonucleoprotein enzyme that maintains telomere length. The researchers compared *est1Δ* strains with nearly identical wild-type strains and found that the early growth of both cultures on day 4 was similar. By day 7, however, the frequency of *CAN1* deletions in the *est1Δ* strains was approximately 10 times higher than that observed in the wild-type strains. The absence of telomerase caused progressive telomere shortening, resulting in dysfunctional telomeres as cell division continued (Hackett et al., 2001).

Haploid cells of *S. cerevisiae* contain 16 chromosomes and have a genome of approximately 12 Mb. Shao et al. (2018) successively fused these 16 chromosomes into a single chromosome by deleting one centromere and two telomeres during each round of fusion, thereby preventing the formation of dicentric chromosomes. The researchers also removed redundant copies of telomere-associated long repetitive sequences from chromosomes not selected for fusion to prevent homologous recombination at unintended sites. The resulting single-chromosome *S. cerevisiae* strain remained biologically functional but exhibited a markedly altered overall three-dimensional chromosomal structure. Its single point centromere was only 125 bp long, and the engineered chromosome was approximately eight times larger than the longest native chromosome (Shao et al., 2018).

Discussion

During the early years of DNA and molecular biology research, Simpson (1966) pointed out that what had been learned about DNA had not yet explained the inheritance of traits, how those traits are expressed in a developing individual, or how they affect the nature of humankind. He described striking anatomical differences between humans and apes, including upright posture, legs that are longer than the arms, an S-shaped curvature of the vertebral column, the positioning of the skull–neck joint near the center of the skull, and several other features unique to humans (Simpson, 1966).

In addition to these anatomical distinctions, significant chromosomal differences exist between humans and the great apes. Humans typically have 46 chromosomes, whereas gorillas, chimpanzees, and orangutans typically have 48. Under the evolutionary model, the reduction from 48 to 46 chromosomes would have required the fusion of two ancestral acrocentric chromosomes, possibly accompanied by additional chromosomal rearrangements (Miller, 1977). Although chromosome fusion is relatively rare, Robertsonian translocations have been cited as evidence that such a fusion could have occurred in the ancestry of human chromosome 2 and as a potential mechanism of speciation in humans (Dutta et al., 2024; Song et al., 2016).

However, carriers of Robertsonian translocations have an increased risk of miscarriage and other adverse reproductive outcomes because of improper segregation of the translocated chromosome during meiosis and the resulting transmission of chromosomally unbalanced gametes. This process can increase the risk of conceiving a fetus with an unbalanced chromosomal complement (Dutta et al., 2024; Song et al., 2016; Stankiewicz, 2016). No chromosomal fusion has been directly observed or documented to produce the extensive anatomical and phylogenetic differences that exist between humans and apes. The proposed formation of human chromosome 2 through the fusion of two ancestral ape chromosomes is therefore presented as a post hoc explanation based on the prior assumption that humans and apes share a common evolutionary ancestry.

Heterochromatic bands on chromosomes 1, 9, and 16, as well as those on all human acrocentric chromosomes, contain satellite DNA (satDNA) repeats that are not present in the corresponding regions of human chromosome 2 (Lopes et al., 2021). Another major difference between human and chimpanzee chromosomes is that approximately half of chimpanzee chromosome ends are capped with AT-rich, DAPI-bright bands, whereas these bands are absent from human chromosomes (Fan et al., 2002). Normal telomeres contain approximately 5,000–15,000 bp of telomeric repeats, whereas only approximately 798 bp of telomere-like sequence has been identified at the proposed fusion site of human chromosome 2. A head-to-head fusion of two intact telomeres would therefore be expected to contain approximately 10,000–30,000 bp of telomeric sequence. Researchers recognize this discrepancy and describe the proposed fusion region as severely degenerate (Fan et al., 2002; Tomkins, 2017).

Telomere-to-telomere fusions can result in pathogenic genomic instability or cellular death (Stroik & Hendrickson, 2020). These end-to-end chromosome fusions can produce dicentric chromosomes, which contain two centromeres rather than the usual one. When dicentric chromosomes break during cell division, their broken ends can undergo additional fusion events, perpetuating a breakage–fusion–bridge (BFB) cycle and increasing genomic instability and mutation rates (Hackett et al., 2001). Increased mutation rates have been associated with decreased resistance to infection in mice and with the development of many monogenic diseases in humans (Bach, 2019).

Common Ancestry is Not a Necessary Conclusion

Most scientists accept that a chromosomal fusion event may have occurred in the distant past. However, a small number of researchers argue that humans and apes were created separately and that a fusion event occurred within human history. One line of evidence cited in support of this hypothesis is that the fused chromosome is found only in humans and not in apes, making it an apomorphic feature (Luskin, 2022). According to the online Oxford Dictionary, an apomorphy is “a novel evolutionary trait that is unique to a particular species and all its descendants.”

Under the common-ancestry model, an initial offspring carrying the fused chromosome would have had to reproduce with individuals possessing 48 chromosomes. Luskin (2022) argued that such a chromosomal difference could result in sterility or nonviable offspring. Alternatively, the individual carrying the fusion would eventually have had to reproduce with another individual possessing the same homologous fusion within the same population and general period, a scenario that appears highly improbable.

Fan et al. (2002) described the chromosome 2 fusion as unique to humans and fixed within the human population; therefore, “the fusion must have occurred after the human–chimpanzee split, but before modern humans spread around the world” (p. 1657). The researchers also stated that the offspring carrying this fusion “would have had reduced fertility because of the risk of unbalanced segregation of chromosomes during meiosis” (Fan et al., 2002, p. 1657).

Additional evidence cited against the proposed formation of human chromosome 2 through the fusion of two ancestral acrocentric chromosomes concerns subtelomeric rearrangements. Knight et al. (1999, as cited in Martin et al., 2002) reported that such rearrangements accounted for 7.4% of cases involving moderate-to-severe intellectual disability. Reduced fertility and serious developmental abnormalities would not generally be expected to promote the successful transmission and eventual fixation of a novel chromosomal arrangement.

In many of the studies discussed in this paper, researchers began with the assumption that human chromosome 2 originated through the inversion of one ancestral chromosome and the simultaneous telomere-to-telomere fusion of two ancestral chromosomes corresponding to chimpanzee chromosomes 2A and 2B. This interpretation was based largely on G-banded chromosome studies published by Yunis and Prakash (1982) and the molecular analysis conducted by Ijdo et al. (1991). Initial staining and hybridization techniques, however, provided only estimates of sequence similarity (Bergman & Tomkins, 2011).

Studies involving engineered yeast chromosomes likewise did not demonstrate that a naturally occurring reduction in chromosome number through fusion would produce morphologically different offspring. The transcriptome of the engineered single-chromosome yeast was nearly identical to that of its wild-type parent strain, whose genome was approximately 11.8 Mb. Although Shao et al. (2018) demonstrated that extensive chromosomal fusion could remain biologically functional in yeast, the researchers employed a tightly controlled laboratory procedure, and the fitness of the single-chromosome strain was significantly lower than that of the wild-type strain.

Fan et al. (2002) reported that the telomeric repeat arrays at the proposed fusion site were approximately 14% diverged, compared with less than 1.5% divergence in other noncoding sequences of the human and chimpanzee genomes under the inferred six million years of evolutionary separation. A head-to-head fusion of two intact telomeres would be expected to contain at least 5,000 bp of tandem TTAGGG repeats from one chromosome arm and approximately 5,000 bp of oppositely oriented CCCTAA repeats from the other. The absence of long tandem arrays of these oppositely oriented telomeric sequences at the proposed fusion site has therefore been presented as a significant finding from genomic sequencing studies (Bergman & Tomkins, 2011).

Tomkins (2017) stated, “However, the strongest evidence for negation of fusion [is] ... that the purported fusion site (read in the minus-strand orientation) is a functional DNA-binding domain inside the first intron of the DDX11L2 noncoding RNA helicase gene acting as a second promoter” (p. 45).

A More Plausible Theory of Human Origins

“First and most important is the fact that mankind is a kind, a definite and single species” (Simpson, 1966, p. 152). Charles Darwin (1871) proposed that humankind descended from a lower form of life based partly on similarities in skeletal structure between humans and mammals such as monkeys, bats, and seals. However, closer examination of human anatomy and physiology reveals substantial differences between human and animal musculoskeletal structures and mental capacities. Humans are the only obligately bipedal species and the only species capable of producing complex technologies, works of art, and musical and literary compositions.

Humans reproduce exclusively with other humans to produce fertile offspring who share the fundamental anatomical characteristics of humankind, except when genetic mutations or chromosomal abnormalities alter particular features (Hackett et al., 2001; Simpson, 1966; Song et al., 2016). Regardless of these abnormalities, affected offspring remain fully and functionally human.

Stankiewicz (2016) proposed that the end-to-end telomeric fusion associated with human chromosome 2 may have created a reproductive barrier between humans and the great apes. However, because telomeres function to stabilize chromosome ends and prevent chromosomal fusion, their protective role raises questions about how such a fusion could have occurred naturally. In a study of chromosomal rearrangements involving terminal deletions, translocations, and ring chromosomes, Fortin et al. (2009) found that terminal deletions were subject to chromosome healing after chromosomal breakage. The inheritance of some of these deletions from a parent further indicated that the healed chromosome ends were stable. Had breaks occurred in two ancestral ape chromosomes corresponding to the chromosomes proposed to have formed human chromosome 2, one or both broken ends may have undergone chromosome healing, thereby preventing the proposed fusion event.

A more plausible explanation of human origins is that fully formed male and female humans reproduced with one another to establish the human lineage. Every organism’s genome comprises an integrated body of genetic information specific to that form of life, compactly organized within the cell nucleus (Sanford, 2014). Consequently, each life form possesses a distinctive genomic organization.

To this author’s knowledge, the scientific literature has not demonstrated the natural presence of specifically human cells within nonhuman organisms. Likewise, studies of chromosomal translocations, inversions, and fusions have not demonstrated the production of fundamentally new kinds of organisms. As the literature discussed in this paper indicates, chromosomal abnormalities commonly result in reduced viability, impaired health, or decreased fertility.

These observations further challenge the proposition that the fusion of two ancestral ape chromosomes produced fertile human offspring. Based on the evidence presented, chromosome 2 fusion should be rejected as an explanation for ape-to-human evolution and the origin of humankind.

Conclusion

Empirical genomic data increasingly challenge the long-standing assumption that human chromosome 2 arose through a telomere-to-telomere fusion of two acrocentric chimpanzee chromosomes. High-resolution sequencing indicates that the proposed fusion region contains only approximately 798 bp of telomere-like repeats, far fewer than the 10,000–30,000 bp expected from a true head-to-head fusion. Furthermore, these repeats are highly degenerate, interspersed, and structurally inconsistent with normal telomeric architecture. Comparative analyses also reveal extensive differences between humans and great apes in satellite DNA organization, subtelomeric heterochromatin, centromeric higher-order repeat arrays, and gene regulation (Fan et al., 2002; Ventura et al., 2012; Yoo et al., 2025).

Moreover, documented telomere fusions in living organisms are associated with genomic instability, dicentric chromosomes, breakage–fusion–bridge cycles, and reduced viability (Hackett et al., 2001; Stroik & Hendrickson, 2020), rather than with the emergence of anatomically and cognitively distinct species. Collectively, these findings indicate that the chromosome 2 fusion hypothesis is biologically improbable and unsupported by direct molecular evidence.

When the genetic findings are considered on their own terms, a different picture emerges—one consistent with humans possessing a unique and internally coherent genome rather than a remodeled ape karyotype. The absence of functional or structural intermediates, the lack of a demonstrated evolutionary pathway capable of producing fertile offspring following such a fusion, and the distinct genome-wide architecture of human DNA all point toward humans originating as a separate, fully formed kind rather than as modified descendants of an ape-like ancestor.

This scientific conclusion is consistent with the biblical teaching in Genesis 1:26–27 that humans were created in the image of God and endowed from the beginning with the genetic capacity necessary for rationality, creativity, moral agency, and relational life. Rather than conflicting with the empirical evidence, the creation framework provides a coherent explanatory model: a distinctive human genome is precisely what would be expected if humans were intentionally formed as a unique biological and spiritual kind rather than produced through random chromosomal accidents.

In summary, the cumulative genomic evidence does not validate a chromosomal fusion event as the origin of human chromosome 2, nor does it support shared ancestry between humans and the great apes. Instead, the evidence is more consistent with a model in which humans originated as a separate, specially created kind, reflecting both their biological distinctiveness and their theological status as bearers of the image of God.

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