

Application of a Hypothesis to Speciation in Hominidae

George E. Parris

9601 Warfield Road, Gaithersburg, MD 20882, USA. Email: antimony_121@hotmail.com

Abstract

I have previously hypothesized that biological species are not the result of gradual changes in the genome or morphology as suggested by Darwin, but rather the result of punctuated major pericentric or paracentric inversions or other rearrangements (e.g., chromosome fusions) that prevent reproductive compatibility with the parent group. Following the rearrangement, a new nascent species can be formed through inbreeding within two generations consistent with the views of Goldschmidt. Applying this hypothesis to speciation in Hominidae (the great apes) suggests that (i) orangutans are close to the common ancestor of Hominidae; (ii) humans are close to the common ancestor of Hominoids, which was adapted for efficient all-terrain locomotion; (iii) gorillas and chimpanzees have passed through more species as they have adapted to a very specialized ecological niche in the tropical forest, and (iv) speciation events in *Homo* facilitated evolution of the human brain.

Key words

Evolution, Chimpanzee, Human, Species, Pericentric Inversion, Brain

Introduction

In a recent paper in this journal [1], I argue that biological speciation (i.e., reproductive isolation) is the result of common pericentric or paracentric inversions or rarer large scale chromosomal rearrangements (e.g., chromosome fusions or fractions) that have little or no effect on phenotype. Mutations of these types and magnitude are believed to adversely affect the metaphase alignment and/or anaphase segregation of chromosomes during the first few divisions of the fertilized egg [2-5]. Note that there are many other smaller or less strategically placed inversions in human chromosomes that probably do not interfere with reproduction or cause speciation [6]. A nascent new species is formed by inbreeding of the heterozygous clan members, leading to mutually fertile, homozygous offspring within two generations.

The nascent species resulting from these rearrangements then acquire minor mutations that vary the phenotype from the coexisting parental species [7]. Natural selection (as proposed by Wallace [8]) acting on the populations then selects

the successful species for the specific habitat. The fittest phenotype eventually dominates in each habitat/ecosystem. It should be noted that the modern synthesis of evolutionary theory [9] does not recognize these inversions and fusions as speciation events, but rather interprets them as becoming fixed in species during Darwinian evolution (i.e., species created by progressive natural selection as implied by Darwin's *On the Origin of Species: By Means of Natural Selection*) [10]. Without preliminary reproductive isolation, changes in morphology are likely very slow and easily reversible.

My hypothesis [1] draws a line between what a biologist might consider to be a species (based on reproductive compatibility) and what an anthropologist/paleontologist might consider to be a species based on morphology of fossils. It calls attention to the fact that within a biological species there may be substantial anatomical differences, while between species their might be almost no anatomical difference. The hypothesis might provide a framework for interpretation of paleontological observations.

An application of the hypothesis to Hominidae

Keep in mind that we only have access to the genomes of surviving species, but we can deduce the gross features of the genome of their most recent common ancestors, when there are two surviving species. Note that the frequency of the major mutations and formation of nascent species is much greater than the frequency of replacement of the parent species through natural selection. In fact, in a large population there are always nascent species [1]. Because reduced fertility with the parent group leading to speciation appears to be associated only with major mismatches in chromosome parity [1], the results of Yunis and Prakash [11-13] are adequate to identify the most likely speciation events in the Hominidae line. They identify 5 events (usually pericentric or paracentric inversions) in the ancestral line leading to Hominidae (>16 Ma). The orangutan (*Pongo*) line split first and has retained only 6 major events (in chromosomes 2q, 4, 8, 11, 17 and 20) in about 16 million years. Then, in the line leading to the surviving African apes, each chromosome except 6, 13, 19, 21, 22, and X was replaced by its mutant before the split of the gorillas (*Gorilla*). This implies a high rate of selective adaptation (i.e., a succession of 19 species in 7 million years during the Miocene radiation [14], 15 to 8 Ma). The gorilla line has retained 9 major events (chromosomes 1, 4, 5, 8, 10, 12, 14, 16 and 17) over the last 6-8 million years. During the two to three million years immediately after the gorillas split [14, 15], three events (chromosomes 2p, 7 and 9) formed the common ancestor of chimpanzees (*Pan*) and humans (*Homo*). The chimpanzee line retained 7 major events (all pericentromeric inversions in chromosomes 4, 5, 9, 12, 15, 16 and 17) over the last 4 to 6 million years [15]. In the same period, the human line has retained only three major events (pericentric inversions in chromosomes 1 and 18 and the fusion of 2p and 2q to form human chromosome 2). I interpret these events (i.e., major mutations followed by slower replacement of one chromosome by its mutant in the surviving population) as the boundaries between successive (reproductively isolated) species.

The African apes have diverged genomically and anatomically from their most recent common ancestor. If we look at the last common ancestor of the African apes that lived about 8 Ma [15], gorillas have gone through 9 major events, and chimpanzees have gone through 10 major events, but humans have only gone through 6 major events. I would argue that

the adaptation of chimpanzees and gorillas to thrive in the dense tropical forest habitat was a radical departure from the primate line that leads to *Homo sapiens*; and, thus, required more speciation events to accomplish the deed. The common ancestor of the African apes appears to have been a world-traveling generalist with relatively upright stance and bipedal locomotion capable of crossing all sorts of land forms (across Europe, Asia and Africa during the Miocene [16, 17]) efficiently [18-21]. *Homo sapiens* have built on those ancestral traits, while the gorillas and chimpanzees have adapted to living exclusively in tropical rain forest. While *Homo* developed a larger brain, optimized all-terrain locomotion and adapted for cooling during high energy output by loss of hair [22]; the forest apes [23] have undergone convergent evolution reminiscent of the much earlier speciation of monkeys [24] more than 20 million years ago.

Tree-dwelling became an attractive alternative to plains-dwelling because of the escape from most large predators, available prey in the form of monkeys and, most important of all, the availability of high carbohydrate and vitamin-rich fruits. The primary down-side of moving into the jungle was insect-borne diseases, which were thwarted by hair over most of the forest ape's body [25] and nesting high in insect-repellent trees [26]. The locomotion capabilities of *Gorilla* and *Pan* appear to have become specialized to the point where chimpanzees and bonobos have been isolated for millions of years by the Congo River in comparison to their ancestors who traversed the world.

Speciation and the human brain

Advanced intelligence is one of the hallmarks of our species not shared with other primates. It is clear that the human brain is much larger than that of chimpanzees or other members of Hominidae [27], and that even in very distant species, large brains seem to be correlated with intelligence [28]. There are two genes that seem to be closely associated with brain size [29]. The *microcephalin* group (located at 8p23.1) appears to have evolved during the Miocene (before the last common ancestor of the African apes) [29, 30], but it did not have much effect on brain size. These gene products (MCPH-1 to -4) appear to be necessary, but not sufficient for brain growth without producing tumors [31, 32]. Mutations in *MCPH* genes of humans result in brains the size of chimpanzees and/or dispose humans to various proliferation diseases by deregulation of the G2/M checkpoint [33-36].

The other gene associated with developmental expansion of the brain tissue is known as *ASPM* (located at 1q31.2-32.1). *ASPM*'s function appears to be related to cell proliferation, especially of neurons during development [37, 38]. This region of chromosome 1 was involved in a complex series of rearrangements culminating in a pericentric inversion in the *Homo* line about 3 Ma [39]. As described by Szamalek et al. [39] a duplication was followed by a transposition from 1q32 to 1q21; there was an inverted duplication in the p-arm; and finally these segments were involved in the major (putative speciation event) pericentric inversion between 1p11.2 and 1q21.1. The hypothesis applied here implies that these rearrangements isolated *ASPM* in a clan, which quickly formed a nascent species that allowed positive selection of *ASPM*. Interestingly, a similar chromosome 1 pericentric inversion is found in the *Gorilla* lineage [11], which also experienced rapid evolution of *ASPM* [40]. Once the *ASPM* gene was isolated in a nascent species, it is believed to have taken about 2 million years to bring the gene to its modern form [40].

However, expansion of the brain cannot occur without changes in the skull and the female pelvis. These changes in development and/or adult phenotype were likely accomplished in other speciation events (chromosome 2 fusion and 18 inversion) [11]. According to Rightmire et al. [41] the skull changed slowly until 700,000 years ago (coinciding with perfection of the *ASPM* gene [40]) and modern human skull structure was not achieved until about 200,000 years ago. The skull was refined to a modern appearance a mere 35,000 years ago [41].

References

- [1] Parris, G. E. The hopeful monster finds a mate and founds a new species. *Hypoth Life Sci*, (2011) 1(2), 32-37.
- [2] Bajnoczky, K., Mehes, K. Parental centromere separation sequence and aneuploidy in the offspring. *Hum Genet*, (1988) 78(3), 286-288.
- [3] Vig, B. K. Sequence of centromere separation: occurrence, possible significance, and control. *Cancer Genet Cytogenet*, (1983) 8(3), 249-274.
- [4] Fernius, J., Marston, A. L. Establishment of cohesion at the pericentromere by the Ctf19 kinetochore subcomplex and the replication fork-associated factor, Csm3. *PLoS Genet*, (2009) 5(9), e1000629.
- [5] Hawes, S. M., Gie Chung, Y., Latham, K. E. Genetic and epigenetic factors affecting blastomere fragmentation in two-cell stage mouse embryos. *Biol Reprod*, (2001) 65(4), 1050-1056.
- [6] Feuk, L., MacDonald, J. R., Tang, T., Carson, A. R., Li, M., Rao, G., Khaja, R., Scherer, S. W. Discovery of human inversion polymorphisms by comparative analysis of human and chimpanzee DNA sequence assemblies. *PLoS Genet*, (2005) 1(4), e56.
- [7] Bush, G. L., Case, S. M., Wilson, A. C., Patton, J. L. Rapid speciation and chromosomal evolution in mammals. *Proc Natl Acad Sci U S A*, (1977) 74(9), 3942-3946.
- [8] Bulmer, M. The theory of natural selection of Alfred Russel Wallace FRS. *Notes Rec R Soc Lond*, (2005) 59(2), 125-136.
- [9] Kutschera, U., Niklas, K. J. The modern theory of biological evolution: an expanded synthesis. *Naturwissenschaften*, (2004) 91(6), 255-276.
- [10] Farre, M., Micheletti, D., Ruiz-Herrera, A. Recombination rates and genomic shuffling in human and chimpanzee - a new twist in the chromosomal speciation theory. *Mol Biol Evol*, (2012).
- [11] Yunis, J. J., Prakash, O. The origin of man: a chromosomal pictorial legacy. *Science*, (1982) 215(4539), 1525-1530.
- [12] Ventura, M., Catacchio, C. R., Alkan, C. et al. Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Res*, (2011) 21(10), 1640-1649.
- [13] Ventura, M., Catacchio, C. R., Sajjadian, S., Vives, L., Sudmant, P. H., Marques-Bonet, T., Graves, T. A., Wilson, R. K., Eichler, E. E. The evolution of African great ape subtelomeric heterochromatin and the fusion of human chromosome 2. *Genome Res*, (2012) 22(6), 1036-1049.
- [14] Andrews, P., Kelley, J. Middle Miocene dispersals of apes. *Folia Primatol (Basel)*, (2007) 78(5-6), 328-343.
- [15] Locke, D. P., Hillier, L. W., Warren, W. C. et al. Comparative and demographic analysis of orang-utan genomes. *Nature*, (2011) 469(7331), 529-533.
- [16] Alba, D. M. Fossil apes from the valles-penedes basin. *Evol Anthropol*, (2012) 21(6), 254-269.
- [17] Casanovas-Vilar, I., Alba, D. M., Garces, M., Robles, J. M., Moya-Sola, S. Updated chronology for the Miocene hominoid radiation in Western Eurasia. *Proc Natl Acad Sci U S A*, (2011) 108(14), 5554-5559.
- [18] Crompton, R. H., Vereecke, E. E., Thorpe, S. K. Locomotion and posture from the common hominoid ancestor to fully modern hominins, with special

- reference to the last common panin/hominin ancestor. *J Anat*, (2008) 212(4), 501-543.
- [19] Lovejoy, C. O., McCollum, M. A. Spinopelvic pathways to bipedality: why no hominids ever relied on a bent-hip-bent-knee gait. *Philos Trans R Soc Lond B Biol Sci*, (2010) 365(1556), 3289-3299.
- [20] White, T. D., Asfaw, B., Beyene, Y., Haile-Selassie, Y., Lovejoy, C. O., Suwa, G., WoldeGabriel, G. *Ardipithecus ramidus* and the paleobiology of early hominids. *Science*, (2009) 326(5949), 75-86.
- [21] Ryan, T. M., Silcox, M. T., Walker, A. et al. Evolution of locomotion in Anthropoidea: the semicircular canal evidence. *Proc Biol Sci*, (2012) 279(1742), 3467-3475.
- [22] Sutou, S. Hairless mutation: a driving force of humanization from a human-ape common ancestor by enforcing upright walking while holding a baby with both hands. *Genes Cells*, (2012) 17(4), 264-272.
- [23] Ebersberger, I., Galgoczy, P., Taudien, S., Taenzer, S., Platzer, M., von Haeseler, A. Mapping human genetic ancestry. *Mol Biol Evol*, (2007) 24(10), 2266-2276.
- [24] Morimoto, N., Zollikofer, C. P., Ponce de Leon, M. S. Shared human-chimpanzee pattern of perinatal femoral shaft morphology and its implications for the evolution of hominin locomotor adaptations. *PLoS One*, (2012) 7(7), e41980.
- [25] Silva, J. C., Egan, A., Friedman, R., Munro, J. B., Carlton, J. M., Hughes, A. L. Genome sequences reveal divergence times of malaria parasite lineages. *Parasitology*, (2011) 138(13), 1737-1749.
- [26] Samson, D. R., Muehlenbein, M. P., Hunt, K. D. Do chimpanzees (*Pan troglodytes schweinfurthii*) exhibit sleep related behaviors that minimize exposure to parasitic arthropods? A preliminary report on the possible anti-vector function of chimpanzee sleeping platforms. *Primates*, (2013) 54(1), 73-80.
- [27] Zhang, J. Evolution of the human ASPM gene, a major determinant of brain size. *Genetics*, (2003) 165(4), 2063-2070.
- [28] Xu, S., Chen, Y., Cheng, Y., Yang, D., Zhou, X., Xu, J., Zhou, K., Yang, G. Positive selection at the ASPM gene coincides with brain size enlargements in cetaceans. *Proc Biol Sci*, (2012) 279(1746), 4433-4440.
- [29] Ponting, C., Jackson, A. P. Evolution of primary microcephaly genes and the enlargement of primate brains. *Curr Opin Genet Dev*, (2005) 15(3), 241-248.
- [30] Evans, P. D., Anderson, J. R., Vallender, E. J., Choi, S. S., Lahn, B. T. Reconstructing the evolutionary history of microcephalin, a gene controlling human brain size. *Hum Mol Genet*, (2004) 13(11), 1139-1145.
- [31] Bartek, J. Microcephalin guards against small brains, genetic instability, and cancer. *Cancer Cell*, (2006) 10(2), 91-93.
- [32] Gruber, R., Zhou, Z., Sukchev, M., Joerss, T., Frappart, P. O., Wang, Z. Q. MCPH1 regulates the neuroprogenitor division mode by coupling the centrosomal cycle with mitotic entry through the Chk1-Cdc25 pathway. *Nat Cell Biol*, (2011) 13(11), 1325-1334.
- [33] Lin, S. Y., Rai, R., Li, K., Xu, Z. X., Elledge, S. J. BRIT1/MCPH1 is a DNA damage responsive protein that regulates the Brca1-Chk1 pathway, implicating checkpoint dysfunction in microcephaly. *Proc Natl Acad Sci U S A*, (2005) 102(42), 15105-15109.
- [34] Chaplet, M., Rai, R., Jackson-Bernitsas, D., Li, K., Lin, S. Y. BRIT1/MCPH1: a guardian of genome and an enemy of tumors. *Cell Cycle*, (2006) 5(22), 2579-2583.
- [35] Liang, Y., Gao, H., Lin, S. Y. et al. BRIT1/MCPH1 is essential for mitotic and meiotic recombination DNA repair and maintaining genomic stability in mice. *PLoS Genet*, (2010) 6(1), e1000826.
- [36] Giallongo, C., Tibullo, D., La Cava, P. et al. BRIT1/MCPH1 expression in chronic myeloid leukemia and its regulation of the G2/M checkpoint. *Acta Haematol*, (2011) 126(4), 205-210.
- [37] Fish, J. L., Dehay, C., Kennedy, H., Huttner, W. B. Making bigger brains-the evolution of neural-progenitor-cell division. *J Cell Sci*, (2008) 121(Pt 17), 2783-2793.
- [38] Bond, J., Roberts, E., Mochida, G. H. et al. ASPM is a major determinant of cerebral cortical size. *Nat Genet*, (2002) 32(2), 316-320.
- [39] Szamalek, J. M., Goidts, V., Cooper, D. N., Hameister, H., Kehrer-Sawatzki, H. Characterization of the human lineage-specific pericentric inversion that distinguishes human chromosome 1 from the homologous chromosomes of the great apes. *Hum Genet*, (2006) 120(1), 126-138.
- [40] Kouprina, N., Pavlicek, A., Mochida, G. H. et al. Accelerated evolution of the ASPM gene controlling brain size begins prior to human brain expansion. *PLoS Biol*, (2004) 2(5), E126.
- [41] Rightmire, G. P. Out of Africa: modern human origins special feature: middle and later Pleistocene hominins in Africa and Southwest Asia. *Proc Natl Acad Sci U S A*, (2009) 106(38), 16046-16050.