

Open Symposium

Human evolution: Gradual or Punctuated? Accelerated integration of mtDNA pseudogenes into the nuclear genome coincides with speciation of the human genus. A Hypothesis.

Short: High mutation rates and Homo speciation

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Poster

The question: human evolution - gradual process or a rapid discontinuous change? Whether human origin was a gradual process or was a result of rapid change has been a focus of intense debate. Of particular interest is the climate change ~2.9-2.5 Ma, thought to have precipitated the separation of the genus *Homo* (~2.8Ma). The debate mostly concerned continuity/punctuality of the fossil record, but the rate of the underlying genetic change is of ultimate interest/importance. Did hominid lineage experience a mutation burst when a large number of hominins emerged and gave rise to the split between *Australopithecus/Paranthropus* and *Homo*?

The obstacle: vague timing of conventional mutations. The difficulty in answering the above question lies in the way past mutations are timed. Conventional point mutations are assigned to specific branches of the phylogenetic tree. The problem is that mutations can be positioned within segments from branching point to branching point, but the exact position within the segment is principally unknown. Because the hominid phylogenetic tree is rather sparsely populated with species/branches, the precision of mutation timing is low, e.g., human-specific mutations can be positioned within ~6 My segment (from human/chimp split to present time).

The solution: NUMTs – mutations with an internal clock. NUMTs are insertions of mtDNA sequences into the nuclear genome. Unlike point mutation, each NUMTs actually represents a branch on the mtDNA phylogenic tree and thus its time of insertion can be determined as precise as their branching point can be positioned on the tree. In a sense, NUMTs are “mutations with an internal clock”, which is synchronized with the well-established mtDNA evolution clock. By determining the NUMTs’ insertion time points, one can ask whether NUMTs were inserted uniformly over time or preferentially during certain periods of evolution, as implied by the “punctuated” model.

Results: Hundreds of pseudogenes of the human genome have been inserted over the last ~60 My of which we considered the last 6 My. Various quality filters resulted in the selection of 18 NUMTs most suitable for phylogenetic analysis. Insertion times of these 18 NUMTs cluster around 2.8Ma. While timing of insertion of NUMTs is imprecise, the observation such a cluster is highly statistically significant.

Discussion: It is tempting to hypothesize that accelerated insertion of NUMTs is somehow linked to the speciation process. NUMTs could be either "riders", i.e., their insertion could be facilitated by the overall higher genome rearrangement activity during the speciation period, or "drivers", i.e. they may more readily get fixed in the population during speciation due to increased selective pressures. If correct, the hypothesis of accelerated pseudogenization would support the idea that evolution of our genus might have been discontinuous.

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