

Explore articles making the most
impact from GBE

▶ Find out more



Genome Biology and Evolution

gbe.oxfordjournals.org

Genome Biol. Evol. (2016) doi: 10.1093/gbe/evw132
First published online: June 11, 2016**Emergence and evolution of Hominidae-specific coding and noncoding genomic sequences**Morteza Mahmoudi Saber^{1,2}, Isaac Adeyemi Babarinde^{3,2}, Nilmini Hettiarachchi^{3,2} and Naruya Saitou^{2,1,3,*}

+ Author Affiliations

*Corresponding Author: Naruya Saitou, Division of Population Genetics, National Institute of Genetics, Yata 1111, Mishima, 411-8540, Japan, TEL/FAX +81-55-981-6790/6789, Email: saitounr@nig.ac.jpReceived October 2, 2015.
Revision received February 10, 2016.
Revision received May 10, 2016.
Accepted May 31, 2016.

Abstract

Family Hominidae, which includes humans and great apes, is recognized for unique complex social behavior and intellectual abilities. Despite the increasing genome data, however, the genomic origin of its phenotypic uniqueness has remained elusive. Clade-specific genes and highly conserved noncoding sequences (HCNSs) are among the high-potential evolutionary candidates involved in driving clade-specific characters and phenotypes. On this premise, we analyzed whole genome sequences along with gene orthology data retrieved from major DNA databases to find Hominidae-specific genes and HCNSs. We discovered that Down syndrome critical region 4 (DSCR4) is the only experimentally verified gene uniquely present in Hominidae. DSCR4 has no structural homology to any known protein and was inferred to have emerged in several steps through LTR/ERV1, LTR/ERVL retrotransposition, and transversion. Using the genomic distance as neutral evolution threshold, we identified 1,658 Hominidae-specific HCNSs. Polymorphism coverage and derived allele frequency analysis of Hominidae-specific HCNSs showed that these HCNSs are under purifying selection, indicating that they may harbor important functions. They are overrepresented in promoters/untranslated regions, in close proximity of genes involved in sensory perception of sound and developmental process, and also showed a significantly lower nucleosome occupancy probability. Interestingly, many ancestral sequences of the Hominidae-specific HCNSs showed very high evolutionary rates. This suggests that new functions emerged through some kind of positive selection, and then purifying selection started to operate to keep these functions.

Key words Hominidae conserved noncoding sequences lineage-specific genes
DSCR4 accelerated evolution

© The Author(s) 2016. Published by Oxford University Press on behalf of the Society for Molecular Biology and Evolution.

This is an Open Access article distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/4.0/>), which permits unrestricted reuse, distribution, and reproduction in any medium, provided the original work is properly cited.

Disclaimer: Please note that abstracts for content published before 1996 were created through digital scanning and may therefore not exactly replicate the text of the original print issues. All efforts have been made to ensure accuracy, but the Publisher will not be held responsible for any remaining inaccuracies. If you require any further clarification, please contact our [Customer Services Department](#).

Online ISSN 1759-6653

Copyright © 2016 Society for Molecular Biology and Evolution

[Site Map](#) [Privacy Policy](#) [Cookie Policy](#) [Legal Notices](#) [Frequently Asked Questions](#)

Other Oxford University Press sites:

