

Insights into the structural evolution of amniote genomes

A. Ouangraoua^{1,2}, A. McPherson¹, C. Chauve¹ and E. Tannier³

¹ Simon Fraser University, Burnaby (BC), Canada
 aida_ouangraoua@sfu.ca, andrew_mcpherson@sfu.ca, cedric.chauve@sfu.ca
² Université du Québec à Montréal, Montréal (QC), Canada.
³ INRIA Rhône-Alpes, LBBE, University of Lyon, France I
 eric.tannier@inria.fr

Abstract

We investigate the problem of inferring contiguous ancestral regions (CARs) of the genome of the last common ancestor of all extant amniotes. We use the complete genome sequences and assemblies of 14 vertebrate species: 11 amniote genomes as ingroups and 3 teleost fish genomes as outgroups. We infer large regions or syntenies of three ancestral genomes: the amniote, therian and boreoeutherian ones. We then infer patterns of genome structural evolution in amniotes: types of rearrangements, long and short branches of the underlying phylogenetic tree, convergent and divergent karyotypic evolution.

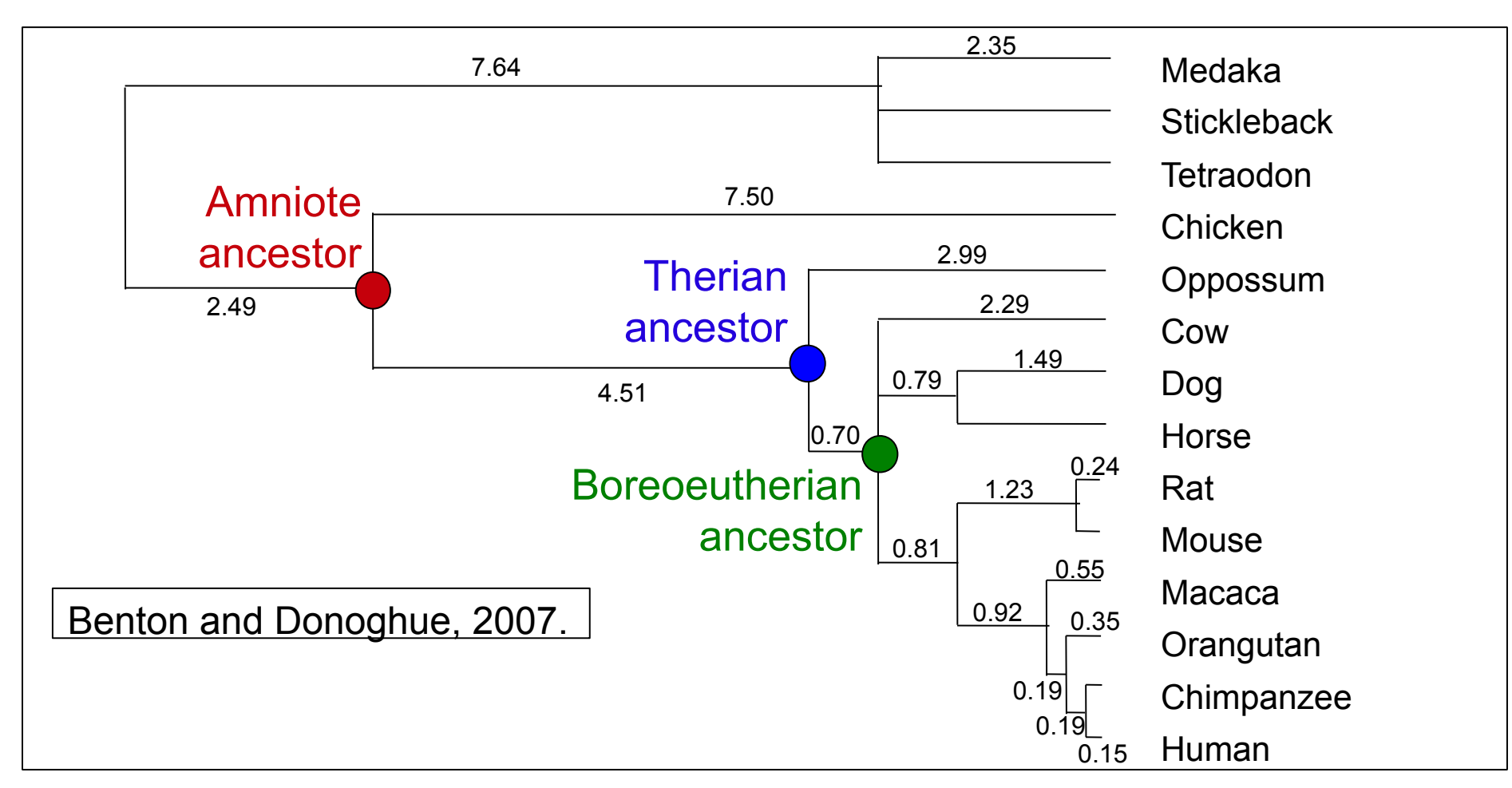
We encounter and explore several methodological issues:

- the construction of good conserved orthology blocks among all amniotes;
- the detection of conserved synteny signals between amniotes and teleost fishes based on the principle of Doubly Conserved Synteny (DCS) used in (Jaillon et al. 2004) and taking into account the whole genome duplication in the teleost lineage;
- the detection of conserved contiguity and synteny signals between amniotes and the construction of large Contiguous Ancestral Regions (CARs) of ancestral genomes (as in Chauve and Tannier, PLoS Comput Biol 2008) and the linkage of CARs according to the DCS signal;
- the detection of reliable ancestral genome rearrangements (as in Zaho and Bourque, Genome Res 2009).

The ancestral boreoeutherian genome we infer is in almost complete agreement with previous cytogenetics and computational studies. Therian and amniote ancestral genomes still miss good references, as two previous studies (Nakatani et al. Genome Res 2007, Kohn et al. Trends in Genetics 2004) gave divergent results. Still the amniote ancestral genome is found relatively close to the chicken genome in all studies, including this one. We analyse every karyotypic change in the chicken and therian branches.

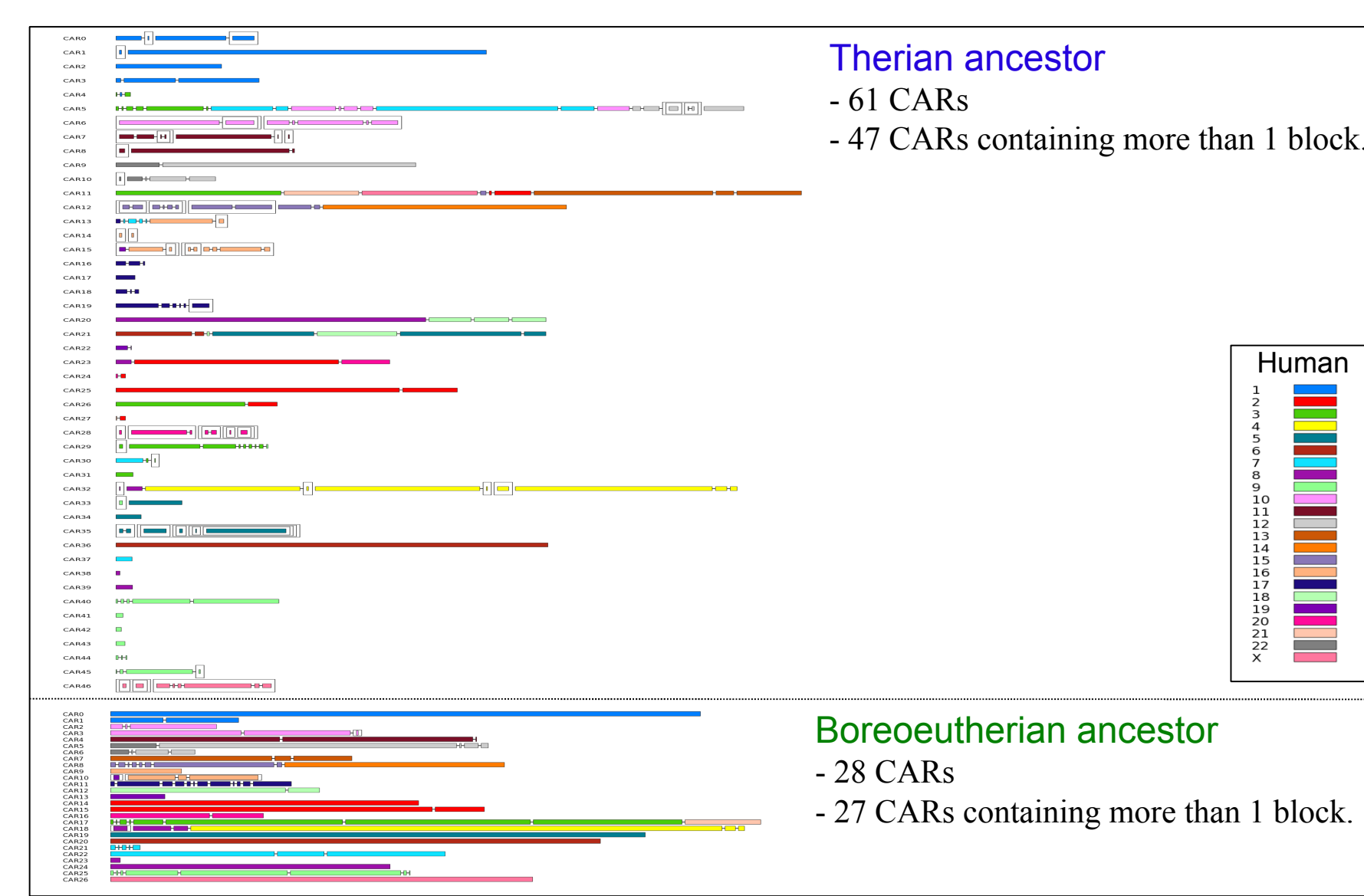
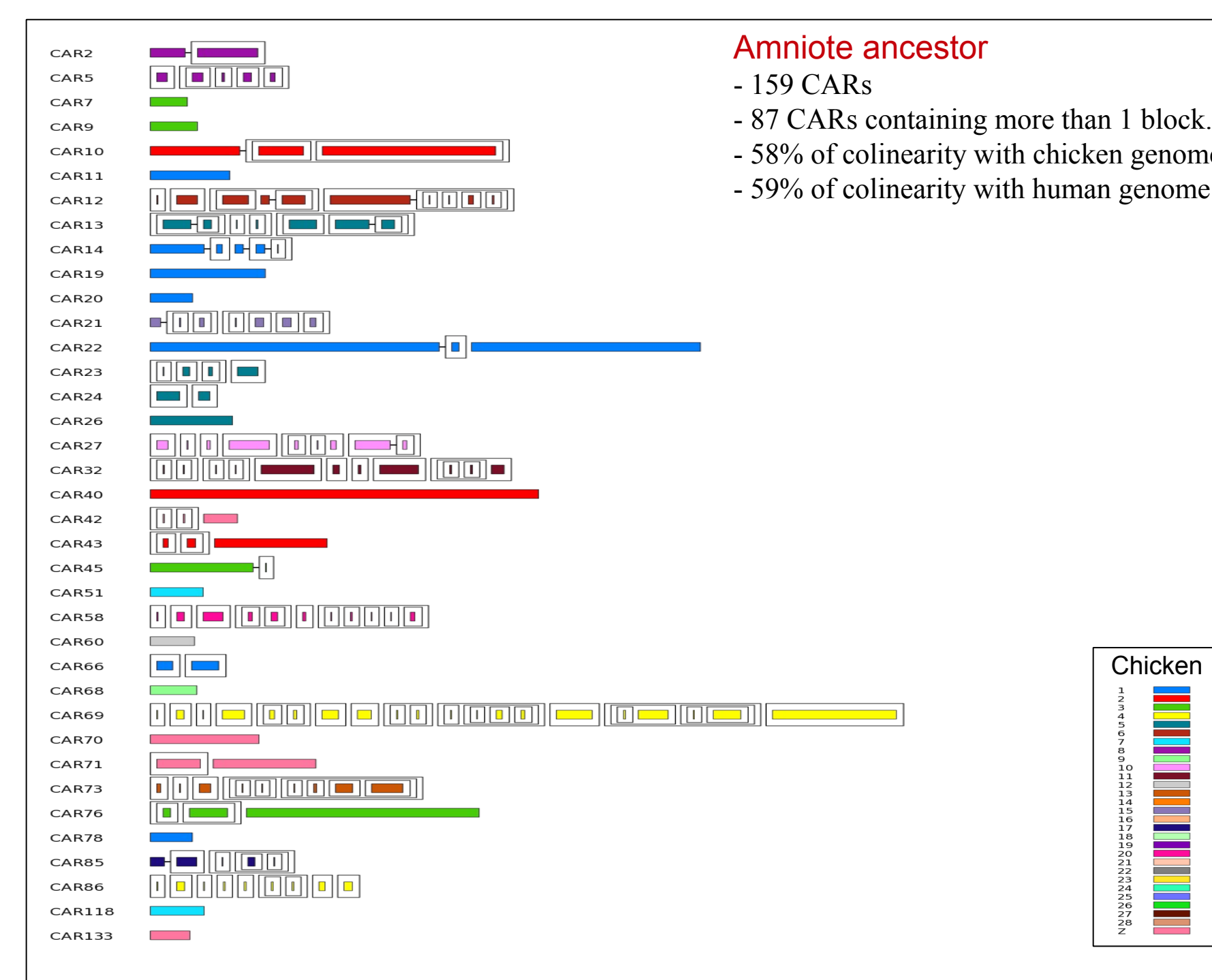
Input data (species tree and gene trees)

- -Pecan 12-amniotes-vertebrates multiple alignments from Ensembl-Compara 54,
- -Gene trees from Ensembl-Compara 54.



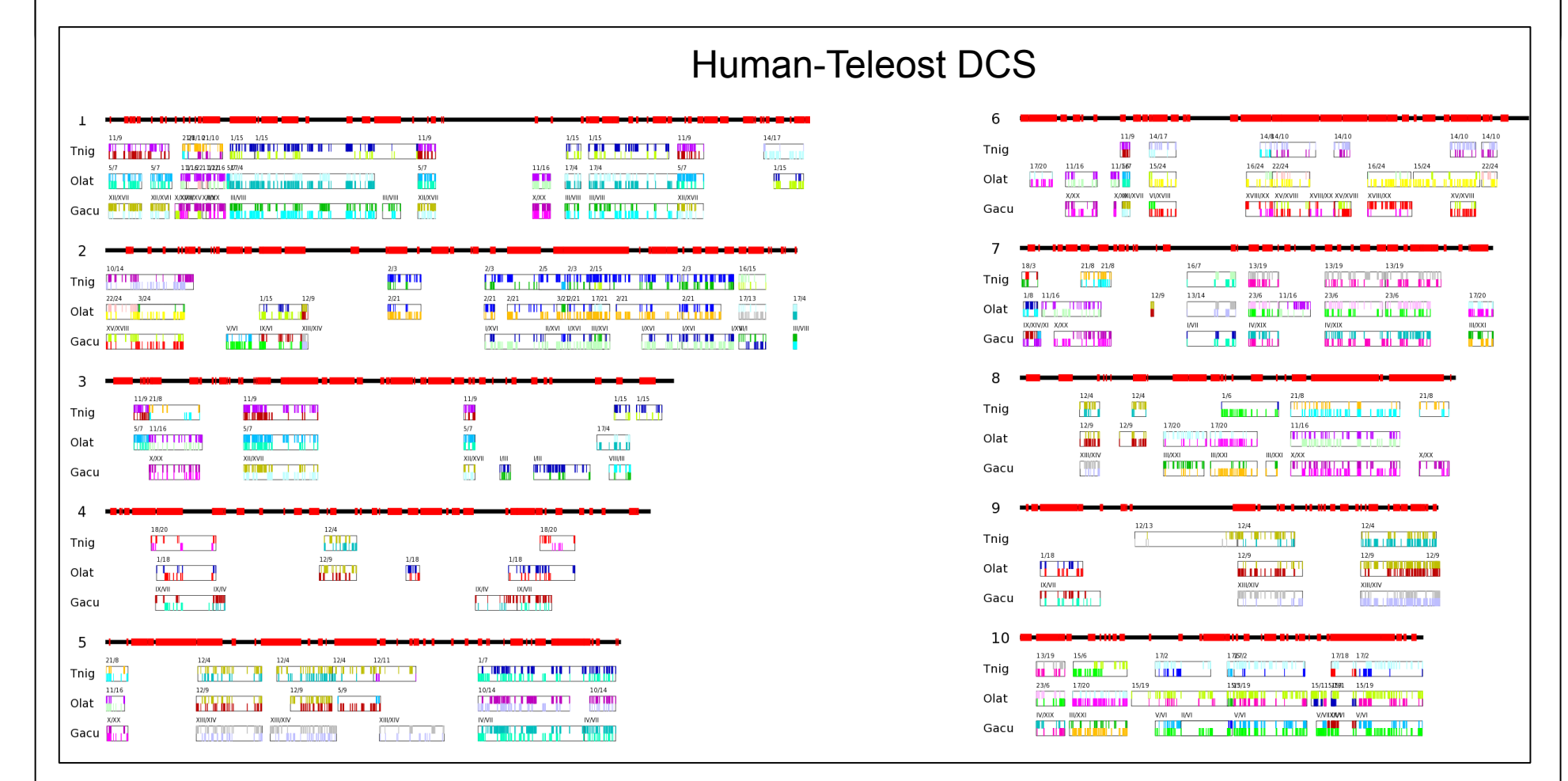
Computing Contiguous Ancestral Regions

- -Weighting adjacencies and ancestral synteny according to their conservation in species.
- -Removing false positive contiguity and synteny signals and ordering orthology blocks.



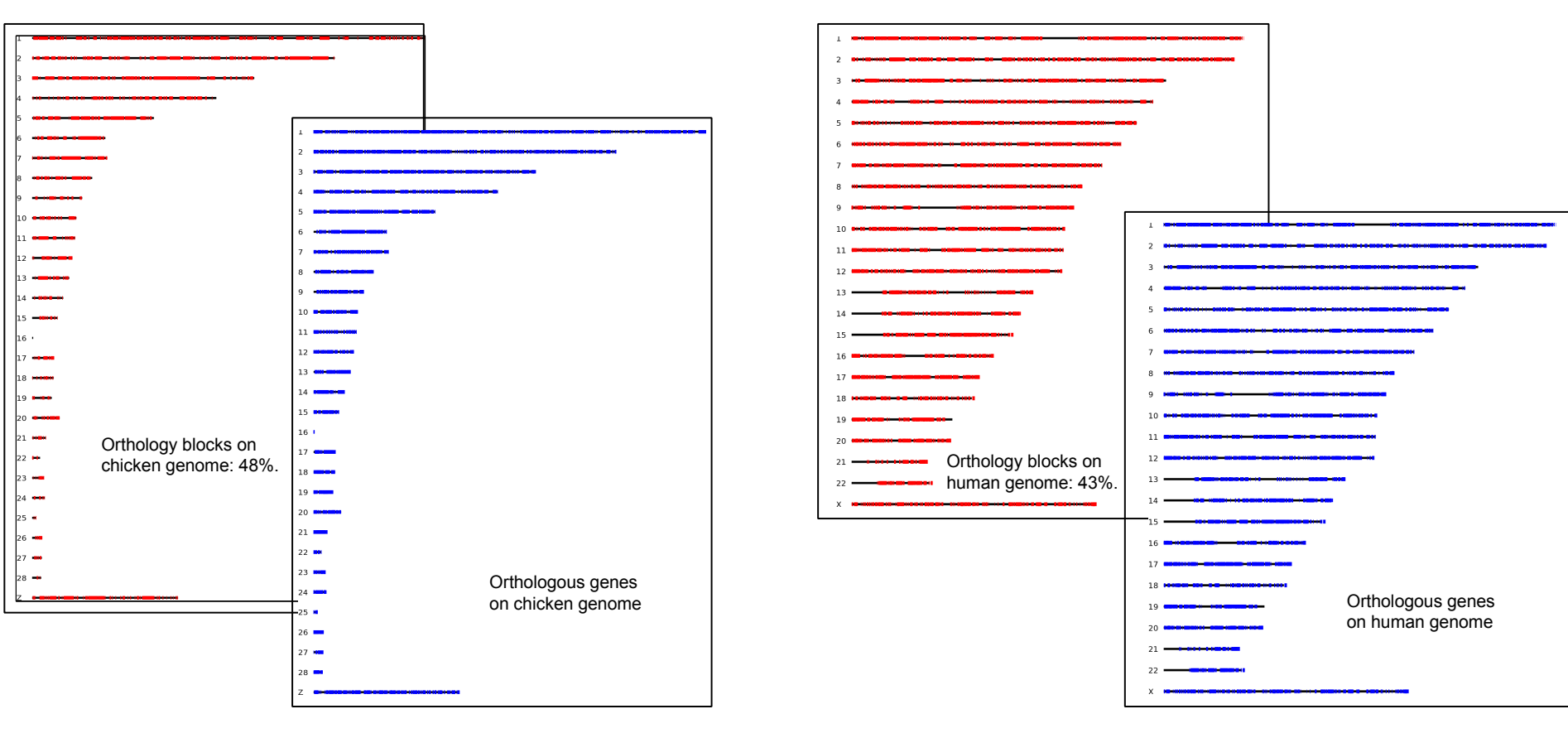
Detecting synteny signals (DCS) between amniotes and fishes

- -Double Conserved Synteny (DCS): sets of orthology blocks contained in an amniote genome segments whose genes have orthologs in two segments of a fish genome.



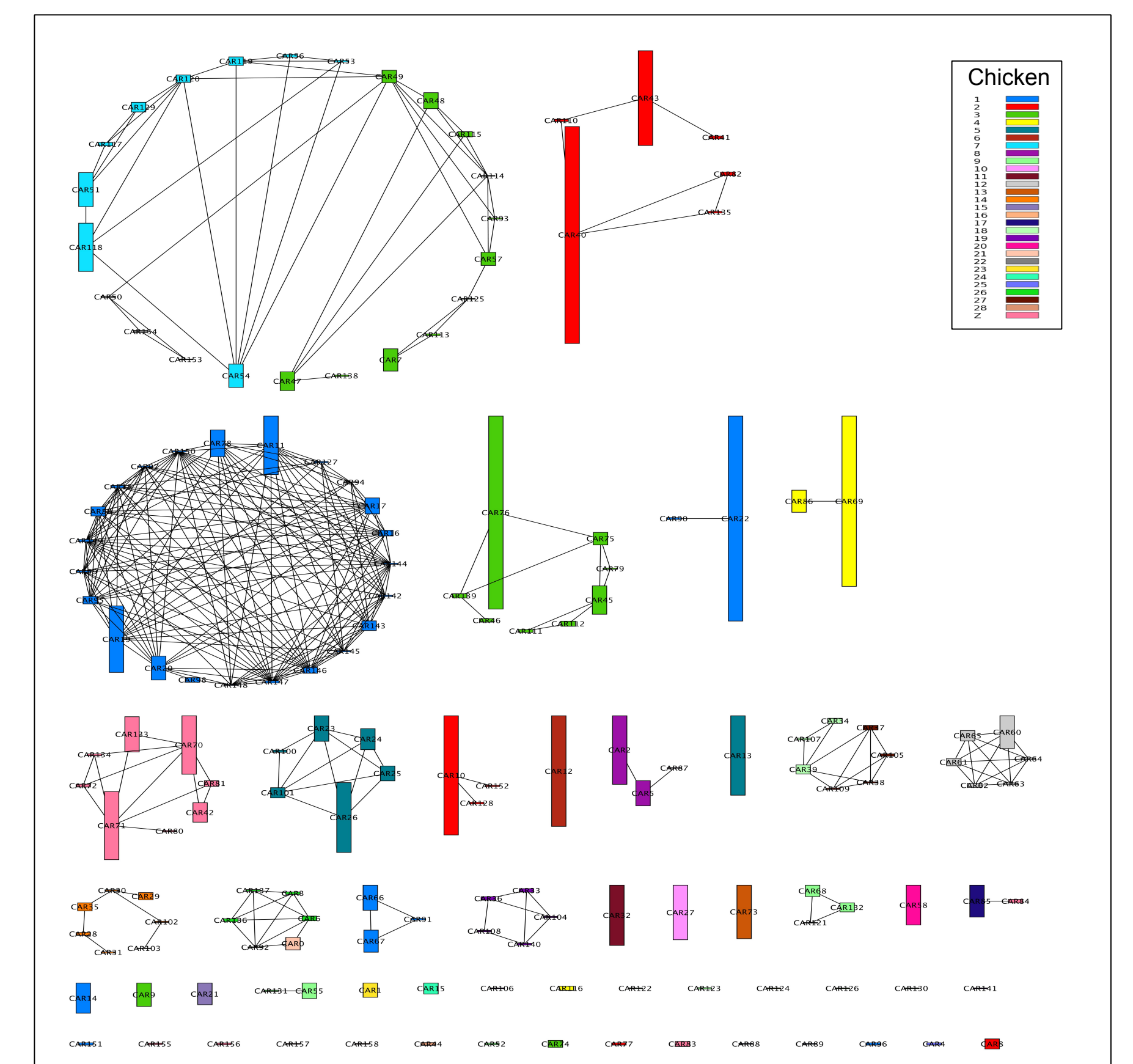
Orthology blocks and orthologous genes

- -690 non-overlapping orthology blocks on amniotes spanning 48% of human genome and 43% of chicken genome.
- - 572356 gene orthologies between amniotes and teleost fishes.



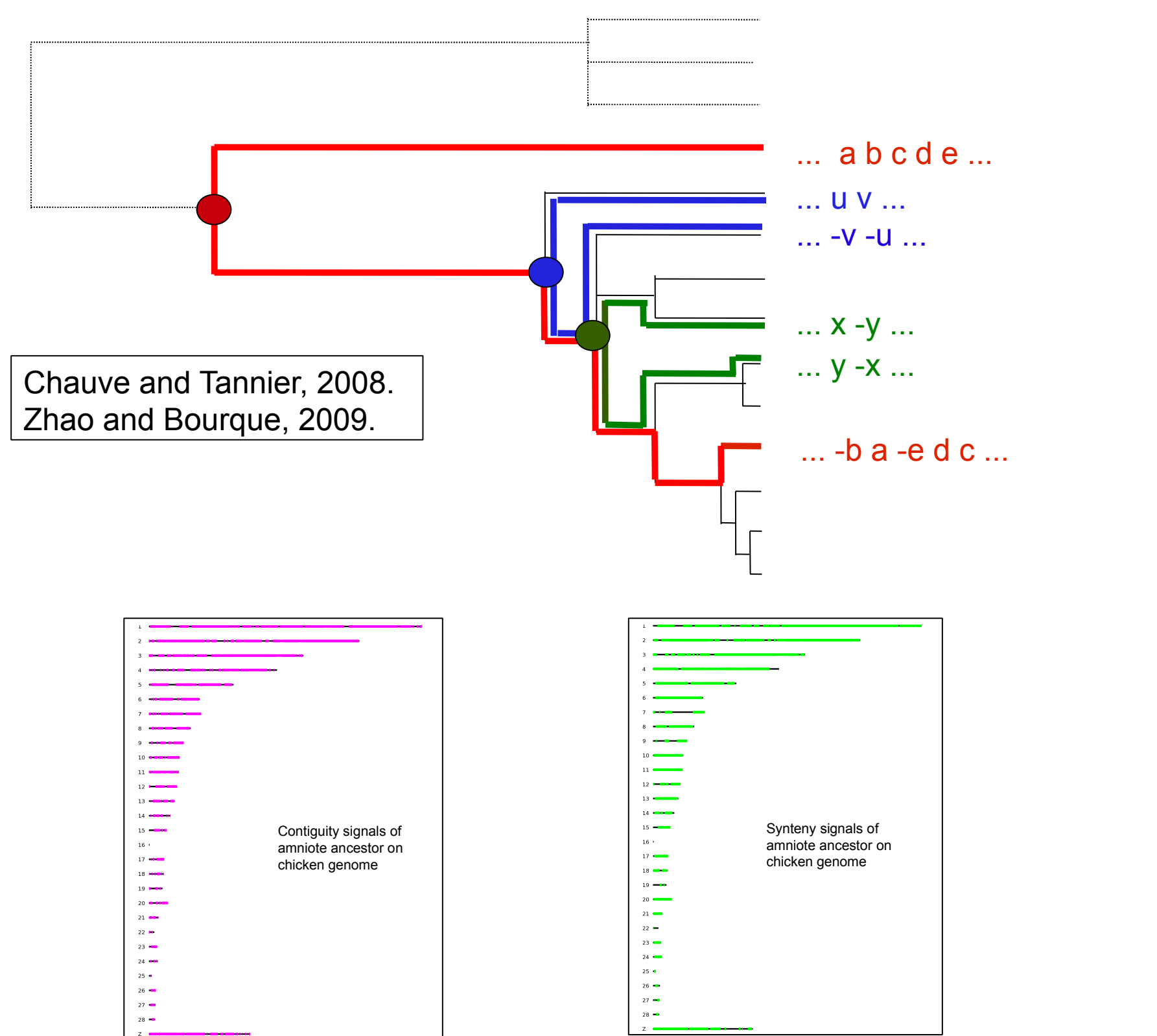
Linking CARs according to the DCS signal

- -Linking couples of orthology blocks that belong to two different CARs but to the same DCS.



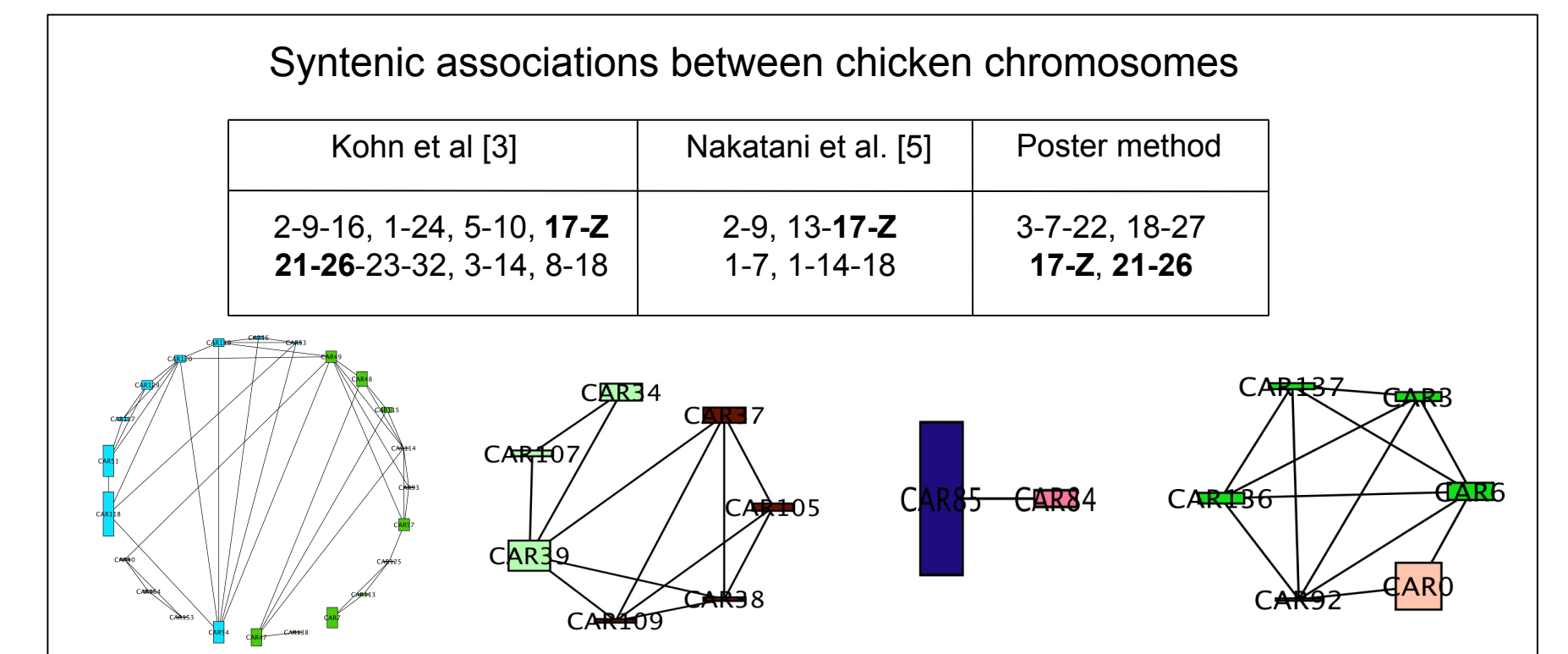
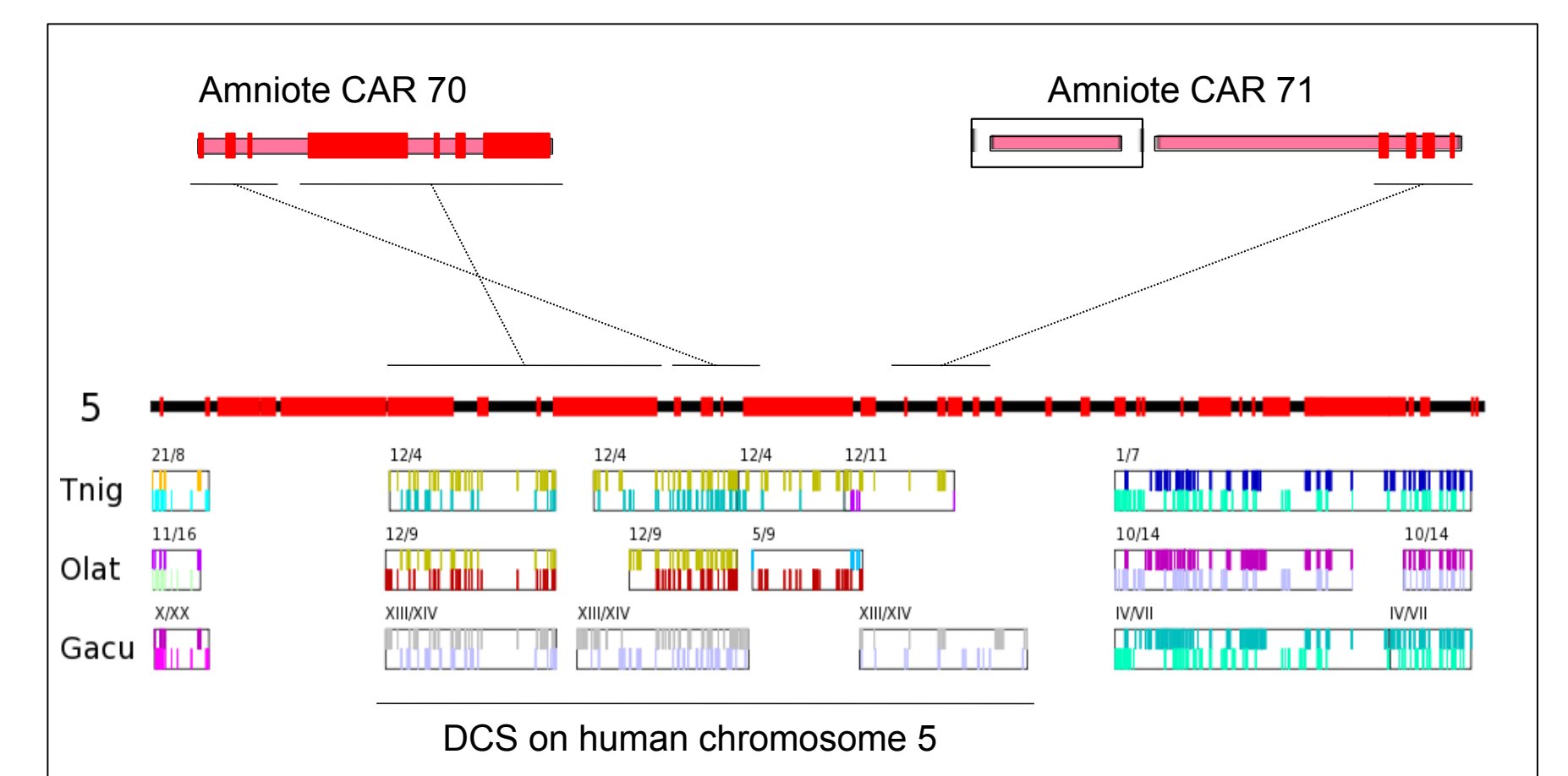
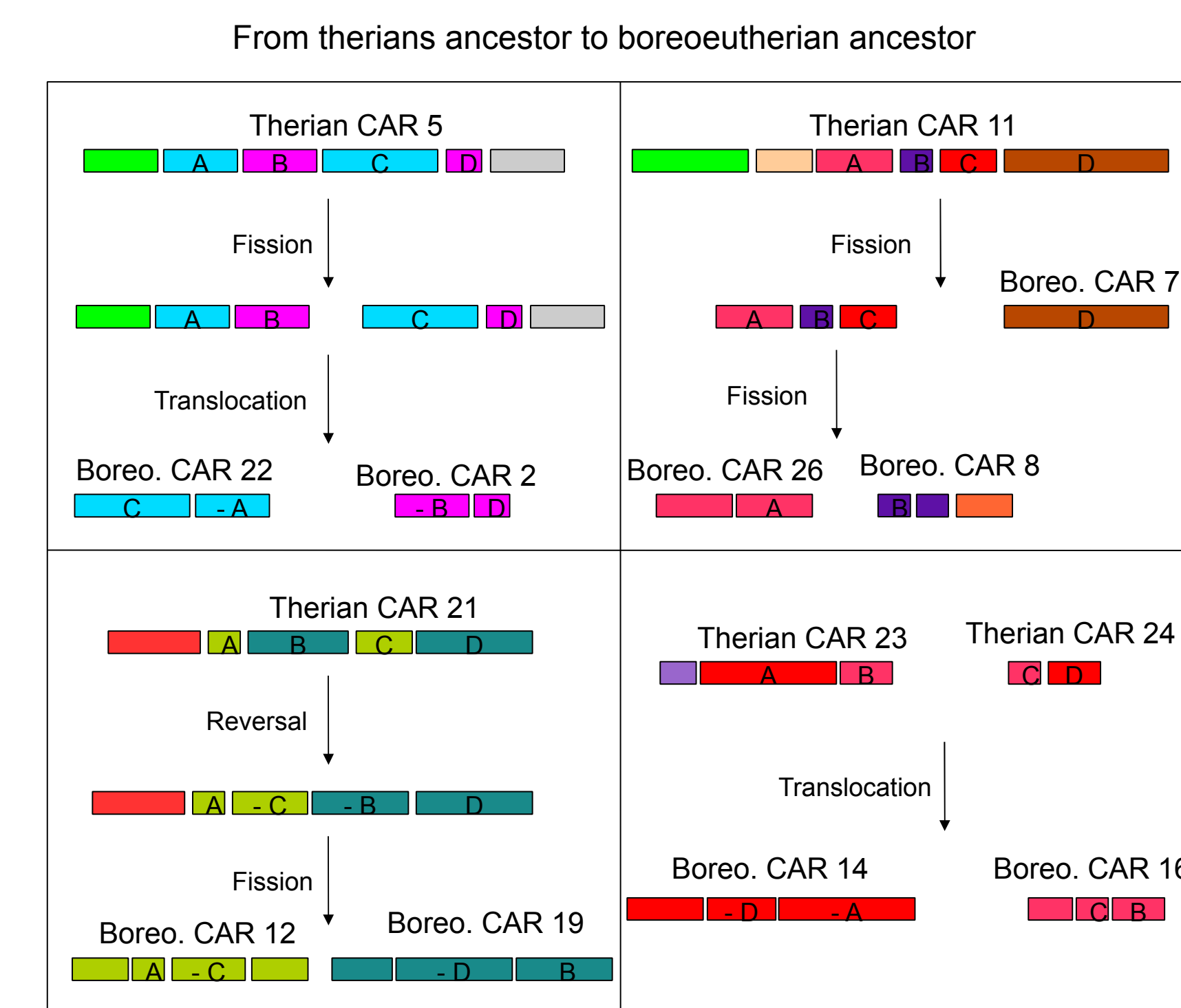
Detecting contiguity and synteny signals between amniotes

- -Contiguity signals: conserved adjacencies between extant amniotes species.
- -Synteny signals: maximum common intervals between extant amniote species.



Detecting signals of ancestral rearrangement events

- -Recovering adjacencies and telomeres in two ancestral genomes.
- -Rearrangement signals: Double-Cut-and-Join operations that correspond to reversals, translocation, fusion, fission or transpositions.



Acknowledgement

Eric Tannier is funded by the ANR (ANR-08-GENO-003-01 and NT05- 3 45205) and by the CNRS. Cedric Chauve is funded by NSERC and SFU. Aida Ouangraoua is funded by SFU and UQAM.

References

1. M. J. Benton and P. C. J. Donoghue. Paleontological Evidence to Date the Tree of Life. Molecular Biology and Evolution 24(1):26-53,2007.
2. C. Chauve and E. Tannier. A methodological framework for the reconstruction of contiguous regions of ancestral genomes and its application to mammalian genome. PLoS Comput. Biol. 4:e1000234, 2008.
3. M. Kohn et al. Reconstruction of a 450-my-old ancestral vertebrate protokaryotype. Trends Genet., 22:203-210, 2006.
4. J. Ma et al. D. Haussler, and W. Miller. Reconstructing contiguous regions of an ancestral genome. Genome Research, 16:1557-1565, 2006.
5. Y. Nakatani, H. Takeda, and S. Morishita. Reconstruction of the vertebrate ancestral genome reveals dynamic genome reorganization in early vertebrates. Genome Res., 17:1254-1265, 2007.
6. H. Zhao et G. Bourque. Recovering Genome Rearrangements in the Mammalian Phylogeny. Genome Research 19(5):934-42, 2009.