

Reviewer Report

Title: Chromosome-scale assemblies reveal the structural evolution of African cichlid genomes

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Reviewer name: Alexander Nater

Reviewer Comments to Author:

This paper by Conte and colleagues describes two new chromosome-scale genomic assemblies of African cichlids. The authors used multiple genetic maps to anchor contigs from high-coverage PacBio sequencing and correct misassemblies. Based on these two high-quality genomes and the genetic maps, the authors performed comprehensive comparative analyses of recombination landscapes, large-scale chromosomal rearrangements, and transposable element insertions. The paper presents extensive genomic resources, which will be valuable for future studies in the field. However, the manuscript in its current form is highly descriptive and many parts of the paper are repetitive and very tedious to read. I'm convinced that the appeal of the paper for a broader readership could be improved considerably by shortening the main text and putting a focus on the biologically interesting aspects. The purely descriptive details could be presented more effectively in tables and figures or additional supporting materials. For instance, instead of the lengthy description of rearrangements, regions of unusual recombination, and putative sex determination loci, I would like to see a carefully designed summary figure, which provides the reader with a good overview of these events in the two genomes. In its current form, many interesting aspects are buried in large amounts of text that provide information of little biological relevance. Most importantly, the discussion part should be written much more concisely, as it still largely descriptive and repeats most of the information that was already provided in the results section. Here, the authors should refrain from discussing every single aspect of their results and rather focus on the biological interpretation of the most interesting findings of the study.

Minor comments:

Page 4, line 15: Define "indel" here.

Page 4, line 52: Provide reference for PacBio sequencing.

Page 10, Table 1: Improve the labelling of the table. It is not immediately clear that the numbers represent base pairs.

Page 11, line 42: "relatively complete" compared to what?

Page 11, line 35: The description here seems to imply that the final p-contigs are not phased. The p-contigs are phased within the borders of their associated haplotigs. Please provide a more detailed explanation of p-contigs and haplotigs here.

Page 11, lines 42-55: This part is unclear and should be rewritten. What are "theoretical sizes of heterozygous regions"? Do you mean the theoretical expectation of the distribution of distances between heterozygous sites? The associated Additional File A is completely unclear and needs a much more detailed explanation and legend, e.g. what is the relationship between the two x-axes (length in base pairs and recombination rate) or the two y-axes (frequency and $E(r^2)$)? How do you derive information about the completeness of haplotigs from this graph?

Page 12, line 55: Provide full genus name for *A. koningsi*.

Page 13, Table 3: Why does the total length differ for the four different maps, given that it includes both anchored and unanchored contigs?

Page 15, lines 34-42: Given that the anchoring is based on a combination of four different maps, is it possible that certain contigs are represented multiple times in the final assembly?

Page 16, lines 4-47: Given that all genetic maps are from inter-species crosses, what are the expectations for inter-chromosomal rearrangements that are only present in one of the two species? It seems unlikely, that the given approach would have power to detect rearrangements in such cases.

Page 16 line 34: "... at most 1% of these Lake Malawi genomes is affected by inter-chromosomal rearrangements ..."

Page 23, lines 46-48: This sentence doesn't make sense without a distance qualifier, i.e. significant linkage disequilibrium over extended physical distances.

Page 26, line 59: "Only one contig longer than 1Mbp was not anchored ..."

Page 27, line 7: "Contigs in the M_zebra_UMD2 assembly were primarily anchored with" or "The M_zebra_UMD2 anchoring was primarily performed with"

Page 27, lines 46-48: The suggested link between TEs and chromosomal rearrangements seems a bit far-fetched. It appears more likely to me that low recombination is facilitating the enrichment of both TEs and rearrangements due to reduced N_e and therefore reduced efficacy of selection against slightly deleterious events in these regions.

Page 27, line 56: Not clear what is meant by "orthogonal mapping technologies" here. Alternative mapping technologies?

Page 28, line 51: Genetic differentiation between what?

Page 30, line 33: Linkage group information is missing for the sex determination locus.

Page 30, lines 45-49: Rather the alleles of the sex determination system segregate in three crosses.

Page 32, lines 36-44: The connection between lack of evidence for a chromosome fusion event on LG3 and the accumulation of repetitive elements is not clear.

Page 34, line 29: What are "centromere-containing repeats" and does this refer to the ONSATA and TZSAT satellite sequences in the next sentence? Please rephrase this part.

Page 37, line 38: "will be able to purge"

Page 40, line 9: Incomplete sentence

Page 40, line 29: Omit "that"

Page 40, line 44: Check reference. Reference to PLINK software doesn't make sense in this context.

Methods

Are the methods appropriate to the aims of the study, are they well described, and are necessary controls included? Choose an item.

Conclusions

Are the conclusions adequately supported by the data shown? Choose an item.

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