Chromosomal copy number variation reveals extensive levels of genomic plasticity among and within Trypanosoma cruzi DTUs

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The taxon T. cruzi is divided into six discrete typing units (DTUs), named TcI-TcVI. CL Brener, the reference strain of T. cruzi genome project belongs to the hybrid DTU TcVI, presenting 41 putative chromosomes. Chromosomal Copy Number Variation (CCNV) is a mechanism of gene expansion possibly related to rapid adaptation to new environments, and is already documented in yeast and several Leishmania species. Although studies point toward karyotype variability in T. cruzi strains, the extent of diversity in CCNV among and within DTUs based on read depth coverage (RDC) analysis has not been determined. To identify CCNV among T. cruzi DTUs, we sequenced genomes of strains from TcI, TcII and TcIII DTUs and estimated the ploidy based on RDC of single copy genes in each chromosome. TcI strains had few aneuploidies, while strains from TcII and TcIII DTUs presented a high degree of chromosomal expansions, which is in agreement with the average DNA mass per cell and genome plasticity in these DTUs. Chromosome 31, the only supernumerary chromosome in all T. cruzi samples evaluated, is enriched with genes related to glycosylation pathways, such as the enzyme UDP-GlcNAc-dependent glycosyltransferase, involved in the initial steps of mucin glycosylation. As the strains from the TcII DTU presented a divergent pattern of chromosomal expansions, we sequenced the genome of 7 T. cruzi TcII field isolates from Minas Gerais state, Brazil. These samples presented a complex pattern of chromosomal duplication/loss, which is not in agreement with the phylogeny based on single copy genes. Finally, we sequenced three clones of the TcII Y strain, which presented the same CCNVs as the non-cloned population, suggesting stability in the chromosomal expansions/loss pattern in the population of Y strain. Increased gene copy number due to chromosome amplification may contribute to alterations in gene expression, representing a crucial strategy for parasites that mainly depend on post-transcriptional mechanisms to control gene expression.