Tb035-SNP typing reveals similarity in Mycobacterium tuberculosis genetic diversity in Portugal and Northeast Brazil

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Introduction: Human tuberculosis is an infectious disease primarily caused by Mycobacterium tuberculosis (Mtb). At the moment, the gold standard of epidemiological genotyping of Mtb is Spoligtyping and MIRU-VNTR. These markers, however, have limited use for phylogenetics and strain identification since their fast substitution rate leads to convergent evolution. Recent studies have introduced the use of single nucleotide polymorphisms (SNP) data for identification of Mtb strain groups. In this work we use SNP typing techniques to characterize and compare Mtb populations from Portugal and Brazil. We complement this analysis by discussing the benefits and caveats of this technique. Material and Methods: We defined as set of 79 previously described SNPs to characterize Mtb samples. Using this set we analyze 1915 samples collected in Portugal. From these, 111 were further characterized using Spoligotypes. The set of SNPs was also used to analyze 141 samples from Brazil. All of these were further characterized using Spoligo types. Results: Themost frequent bacterial lineage in both countries is lineage 4 corresponding to around 95% of the analyzed samples. Lineage 1 is also presentin both populations equally, butin considerably low frequencies (around 1%). Contrastingly, Mtb bacteria of lineage 2 are only presentin the Portuguese population with a frequency of almost 3%. With in line age 4, the most frequent strain group sinboth Portugal and Brazil are LAM, followed by Haarlem and then X. Contrarily to these groups, strain group T shows a very different prevalence between Portugal and Brazil, with a frequency of 7% and less than 1.5%, respectively. A comparison between strain identification using Spolygotypes and SNPs show that the former marker misidentify more than 11% of the samples and are unable to identify almost 1%. Furthermore, SNP typing, as opposed to Spoligotypes can provide phylogenetic relationships between the strain group. By close examination of the genotypes of our samples we seem to observe signs of often disregarded recombination events. Main Conclusions: Overall, the use of SNP typing reveals striking similarities between Mtb populations from Portugal and Brazil. We also show that this technique can be useful to reveal signs of recombination events in Mtb. Email:j.sollari.lopes@gmail.com