Comparative assessment of genotyping-by-sequencing and whole-exome sequencing for estimating genetic diversity and geographic structure in natural jaguar populations

Gustavo Lorenzana¹, Henrique Figueiró², Luiz Coutinho³, Priscilla Villela³, and Eduardo Eizirik¹

$^{1}\mathrm{PUCRS}$

²George Mason University ³Escola Superior de Agricultura Luiz de Queiroz, Universidade de São Paulo

November 30, 2022

Abstract

Biologists currently have an assortment of high-throughput sequencing techniques allowing the study of population dynamics in increasing detail. The utility of genetic estimates depends on their ability to recover meaningful approximations while filtering out noise produced by artifacts. We empirically compared the congruence of two reduced representation approaches (genotypingby-sequencing, GBS, and whole-exome sequencing, WES) in estimating genetic diversity and population structure using SNP markers typed in small samples of five jaguar (Panthera onca) demes. Given their intrinsic properties as a targeted capture, WES allowed for a more straightforward reconstruction of loci compared to GBS, which in turn facilitated the identification of true polymorphisms across individuals. In contrast, GBS data showed a recurrent miscalling of heterozygous sites. We therefore used WES-derived metrics as a benchmark against which GBS-derived indicators were compared, varying the values of parameters for locus assembly, genotype calling and SNP filtering in the latter technique. Changes in parameterization induced measurable differences in summary statistics, both between approaches and among distinct batches of GBS data. The application of post-processing genotype filters based on mean depth of reads had major effects on the consistency between approaches. Overall, we observed that the direct empirical comparison of GBS and WES for estimating population genetic attributes from the same set of individuals provided an interesting opportunity to assess the consistency of these approaches, revealing relevant aspects that should be considered in such analyses. Our results highlight the importance of thorough data filtering in genomic approaches to obtain robust genetic diversity and differentiation estimates.

Hosted file

Lorenzana_et_al_jaguar_RRS_MER_final_draft.docx available at https://authorea.com/users/ 400999/articles/608790-comparative-assessment-of-genotyping-by-sequencing-and-wholeexome-sequencing-for-estimating-genetic-diversity-and-geographic-structure-in-naturaljaguar-populations