Towards an interoperating ecosystem of tools and resources for population genetics in R

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Motivation

The broad and inexpensive availability of modern next-generation sequencing and genotyping technologies has led to a wealth of data and analytical methods for population genetics research. There are now dozens of packages available for analyzing and visualizing population genetics data in the popular statistical and mathematical computing platform R. However, this organically grown wealth of methods and packages, combined with the exponential growth of datasets, has also created challenges for researchers to take full advantage of these resources. It can be difficult to know which R packages are best used, and many packages do not interoperate well. A common base class that provides efficient storage of genetic data and promotes interoperability remains lacking even though the need was identified years ago. Algorithm implementations often do not scale well to the kind of large volume datasets that are increasingly common. Creating complex analysis workflows that need to pass data, metadata, and other state information from one package to another can be challenging.

To address these gaps, the Population Genetics in R Hackathon was sponsored by and held at the National Evolutionary Synthesis Center (NESCent) in March 2015. The event targeted interoperability, scalability, workflow gaps, and gaps in end-user documentation. Its goal was ultimately to help foster an interoperating ecosystem of tools and resources for both users and researcher-developers. For more details, see https://github.com/NESCent/r-popgen-hackathon

Result I: Community resource of population genetics analysis vignettes

Motivation: Identifying which R packages can be recruited to answer a particular biological question in population genetics is difficult.

Results:

- Website generated from Rmarkdown + Knitr
- Source and website hosted on Github
- Pull requests auto-tested by Circle CI, master branch changes auto-rebuild the website
- Eight vignettes (5 biological) currently

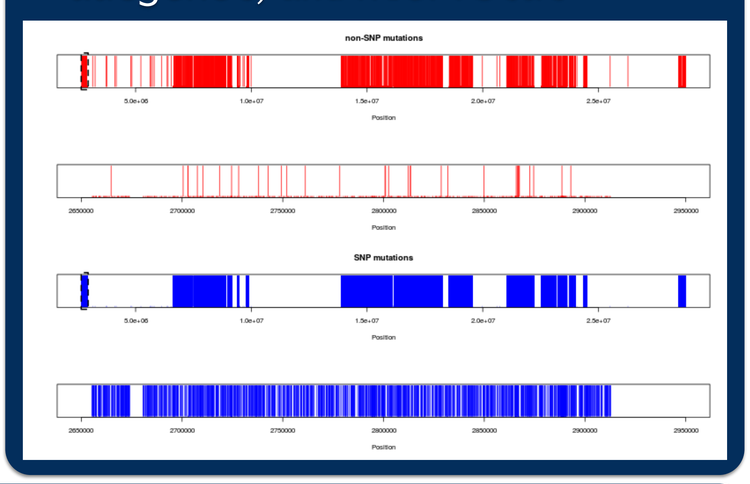
28 Participants from the France, Ireland, New Zealand, Switzerland, UK, US;
Wide variety of career stages, types of employer

Result II: Streamlined, scalable, interoperable handling of VCF data

Motivation: Reading Variant Calling Format (VCF) does not scale well to large-volume data, and interoperability between packages is poor.

Results:

- *apex*: new R package extending ape for multiple genes. On Github and CRAN.
- *adegenet* 2.0 release on CRAN with fast scanning and reading of VCF files
- Genetics data object (*genind*) now interoperable between *pegas*, *adegenet*, and *hierfstat*

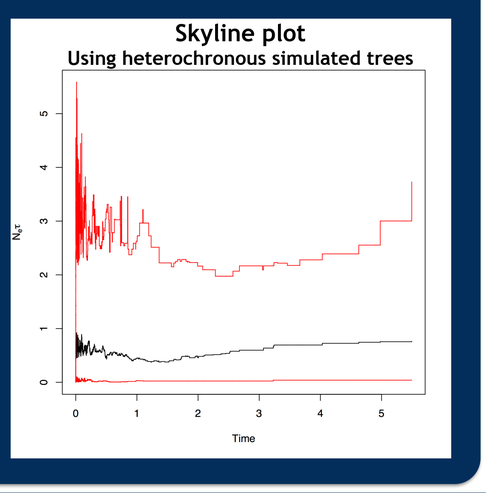


Result V: R package for calculating effective population size in multiple ways

Motivation: Some algorithms for calculating N_e are in different packages, others are not available in R.

Results:

- New multiNe package
- Hosted on Github



Result IV: Identifying and visualizing outliers in multi-variate summary statistics space

Motivation: Multivariate statistics are a common product in population genetic analysis. R tools to summarize and visualize these and detected outliers interactively are lacking.

Results:

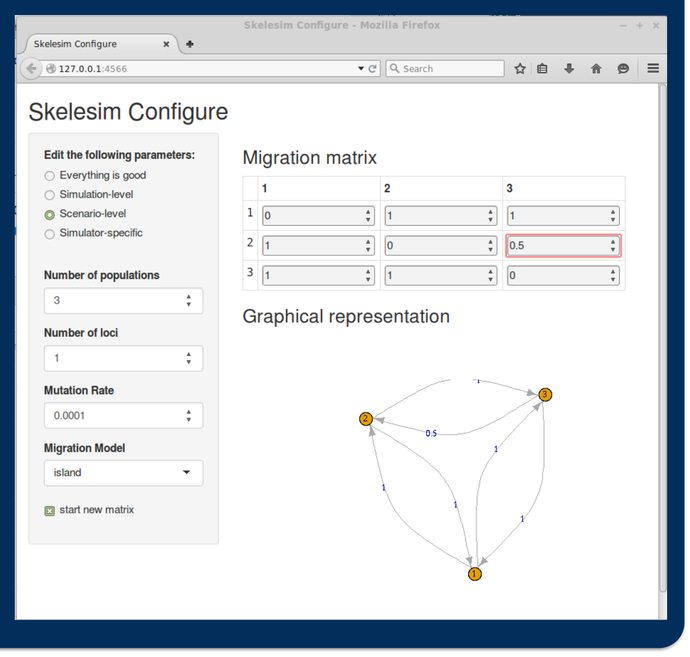
- New MINOTAUR package
- Hosted on Github
- Uses a shiny-powered web user-interface

Result III: Use-cases and power-testing for simulation

Motivation: There are simulator type, parameters, summary metrics, etc to choose when implementing simulations for population genetic questions. There is little guidance on how to decide.

Results:

- New sleleSim package
- Hosted on Github



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