Phylofriend User Guide

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August 17, 2014

Contents

1	Intr	roduction	3
2	2 Installation		4
3	Cor	nmand Line Options	5
4	Examples		
	4.1	Create a Phylogenetic Tree	6
	4.2	Pimp Your Tree with Nicer Labels	6
	4.3	Use a Specific Set of Mutation Rates	7
	4.4	Calibrate Your Data	7
	4.5	Count Mutations	8
	4.6	Extract Data from a Spreadsheet	8
5	Technicalities 10		
	5.1	Source Code Documentation	10
	5.2	Mutation Model	10
	5.3	Mutation Rates	10
	5.4	CSV Input Format	12
	5.5	Text Format	12
	5.6	PHYLIP Format	13
$\mathbf{R}_{\mathbf{c}}$	References		

1 Introduction

Phylofriend's main purpose is to calculate genetic distances from Y-DNA data. The results can be used as input for the PHYLIP[3] program to create phylogenetic trees.

When I started creating phylogenetic trees I often found myself in a difficult position. As a Linux user I was missing some of the tools available under Windows. So I started to write this program to fill in the gaps and make myself comfortable again.

This does not mean that you can not use Phylofriend when working under Windows or the Mac. But currently there is no binary distribution available and you will probably face a hard time installing Phylofriend and the associated programs. So I only recommend this if you are an experienced user.

Phylofriend has some nice features. It can be used

- to create phylogenetic trees using the PHYLIP[3] program. Y-STR values from Family Tree DNA projects can easily be imported.
- as a programming library. Phylofriend is written in Google's Go programming language. This language is not only suited to solve Google's large scale programming problems. It is also an excellent tool for part time programmers who have to concentrate on their projects (often students).
- to extract Y-DNA data from Family Tree DNA projects and convert it into simpler text files that are better suited for further processing.
- to automate phylogenetic tree creation. Phylofriend is a command line tool and this scares many people away. But if you have to repeat the same tasks over and over again you will eventually start to write some scripts and this is where command line tools come in handy.

I hope this program will be useful. Have a good time!

Dirk

2 Installation

This guide is mainly targeted towards persons who use Linux Mint or other Linux versions of the Debian family. Some familiarity with the use of Linux commands is assumed.

Currently there are no binary distributions available for Windows or the Mac. Users of these operating systems can use Phylofriend as well, but they will experience some laborious installation work. The best way is to follow the instructions provided on the Go home page and the PHYLIP home page.

The following list applies to Linux users only:

- Make sure that the Go programming language is installed. If not it can be installed by typing sudo apt-get install golang
- 2. Read the Go Getting Started guide. Make sure to set your GOPATH variable and include it in your PATH so that Go programs can be found.
- 3. For the creation of phylogenetic trees install the PHYLIP program package by typing sudo apt-get install phylip
- 4. Fetch the Phylofriend program with go get code.google.com/p/phylofriend
- 5. Install the program with go install code.google.com/p/phylofriend

3 Command Line Options

Command line options may be given in arbitrary order.

- -help Prints available program options.
- -personsin Filename of the file containing the persons' Y-STR values. This may be in CSV (comma separated values) or text format.
- **-namescol** Number of the column that contains the names when reading CSV files.
- **-mrin** Filename of the mutation rates to use.
- -anonymize If this is true persons' names are replaced by numbers.
- **-modal** Creates modal haplotype.
- **-phylipout** Filename for the distance matrix that can be fed into the PHYLIP[3] program.
- **-mrout** Filename for the output of the currently used mutation rates.
- **-txtout** Filename for text output of persons and Y-STR values.
- **-nvalues** Number of Y-STR values to write to the text output file.
- **-gendist** Generation distance.
- -cal Calibration factor.

4 Examples

4.1 Create a Phylogenetic Tree

- 1. Copy persons' data from a Family Tree DNA project website into a spreadsheet. If the Y-STR values do not appear properly try inserting them into the spreadsheet as unformatted text.
- 2. Save the spreadsheet in CSV (comma separated values) format, for example *persons.csv*.
- 3. Start a terminal or command line interpreter and go to the directory where you stored *persons.csv*.
- 4. Create a matrix of genetic distances by typing phylofriend -personsin persons.csv -phylipout infile
- 5. Use the PHYLIP program to create a tree in Newick format with /usr/lib/phylip/bin/kitsch

You will need to answer some questions. Usually the default values are good enough. The results will be two text files, one named *outtree* which contains the tree in Newick format and another one named *outfile* which contains a more human readable description.

6. Create an image of the tree by typing

/usr/lib/phylip/bin/drawgram

Use *outtree* as the input file name. The resulting tree will be stored in a file named *plotfile*.

A nice alternative to visualize the tree is the use of the Trex[6] webserver. You can copy the contents of the file *outtree* into the Trex window.

If you do not specify a file containing mutation rates Phylofriend will use average 37 marker values as default. They can be found in [5].

4.2 Pimp Your Tree with Nicer Labels

By default Phylofriend assumes that your persons input file's first column contains a list of IDs. This is usually a Family Tree DNA Kit number. The resulting tree is hard to read. Many projects keep names in another column. You can access this column with the *namescol* option. Suppose your second column contains names. You can create a distance matrix with names instead of IDs by typing

phylofriend -personsin persons.csv -namescol 2
-phylipout infile

Due to compatibility issues with other programs the names should be 10 characters long and may not contain Unicode characters. Phylofriend will apply a name transformation but the result is sometimes a bit strange.

You can also use the *namescol* option to create trees that contain the origins of people or the haplogroups. Although I strongly recommend to build trees only from people who belong to the same haplogroup this is sometimes useful if you want to know if different haplogroups are close on their Y-STR values.

If you want to publish your tree you will often need to protect the privacy of the members. This is what the *anonymize* option is for. By typing phylofriend -personsin persons.csv -phylipout infile -anonymize you will get a distance matrix where the names are replaced by numbers.

4.3 Use a Specific Set of Mutation Rates

The first example uses Phylofriend's build in mutation rates which are average values for the standard 37 marker test. Phylofriend supports the use of arbitrary mutation rates by the *mrfile* option. The *phylofriend/mutationrates* directory contains some files with mutation rates. The average mutation rates where taken from [5]. If you like to compare on 67 markers you can use phylofriend -personsin persons.csv -phylipout infile -mrin code.google.com/p/phylofriend/mutationrates/67-average.txt

4.4 Calibrate Your Data

Mutation rates depend on the method applied to calculate genetic distances and the sample populations used. Mutations themselves occur by coincidence. Average mutation rates often yield acceptable results but in most cases you will have to calibrate your data especially if you want to calculate in years.

Phylofriend provides two options for data calibration: *gendist*, the generation distance in years and *cal* an additional calibration factor. Internally they are just multiplied together but using two separate factors seems more convenient for typical use cases.

The default value for the generation distance is 25 years. For time spans over the last few hundred years a generation distance of 30 years often yields better results. This can be done by

phylofriend -personsin persons.csv -phylipout infile
-gendist 30

It is often difficult to calibrate data because you need a reliable paper trail or a well defined historic event. If you are lacking both you can try to apply Klyosov's statistical method[4]. For large enough sample sizes this will effectively reduce the statistical error but you will still be left with an unknown systematical one.

4.5 Count Mutations

The *phylofriend/mutationrates* directory contains sets of mutation rates where all markers are set to 1. This makes mutation counting easy but you will need an additional little trick. Internally Phylofriend uses average values. So it is possible to compare persons who have tested on different sets of markers.

If you want to count mutations for example on a 37 marker scale, you must multiply Phylofriend's internal results with 37. The easiest way to archive this is by misusing the *gendist* option.

```
phylofriend -personsin persons.csv
-mrin code.google.com/p/phylofriend/mutationrates/37-1.txt
-phylipout distancecount.txt -gendist 37
```

4.6 Extract Data from a Spreadsheet

Spreadsheet data is often uncomfortable to handle, especially if you want to write your own program and need to parse it. For this purpose Phylofriend supplies the *txtout* option. It writes data to a text file in simplified form.

The easiest way to use it is

```
phylofriend -personsin persons.csv -txtout persons.txt
```

This extracts the data from *persons.csv* and writes it to *persons.txt*. The first column of *persons.txt* contains the first column found in *persons.csv*, usually a set of IDs. The following columns contain the Y-STR values. All columns are separated by tabs.

If you want to use another column as the first column you can use the *namescol* option:

```
phylofriend -personsin persons.csv -txtout persons.txt
-namescol 2
```

This extracts the second column from *persons.csv* and writes it to the first column of *persons.txt*.

When using the *txtout* option you will need to specify how many Y-STR values are written to the text file. This is done by *nvalues*. Phylofriend will write the same number of Y-STR values to each line. Missing values are written as 0, larger sets of Y-STR values are truncated. This is how to write a full set of 111 markers:

phylofriend -personsin persons.csv -txtout persons.txt
-nvalues 111

5 Technicalities

5.1 Source Code Documentation

To access the source code documentation point your web browser to:

• http://godoc.org/code.google.com/p/phylofriend

If you want to modify the source code it is best to use *godoc* locally on your computer.

- 1. If *godoc* is not yet installed install it by typing sudo apt-get install golang-go.tools (Linux Mint)
- 2. Start the documentation web server with godoc -http=:6060
- 3. Point your web browser to localhost:6060.
- 4. Click on *Packages* and search for *phylofriend*.

This will give you a nice overview of the internal program documentation. You can also click on function names to browse the source code.

5.2 Mutation Model

The two basic mutation models are the infinite alleles model and the stepwise mutation model as explained by Bruce Walsh[8]. Phylofriend uses a hybrid mutation model. Most markers are calculated using the stepwise model, palindromic markers are calculated as described in [2].

As the method for calculating the genetic distance is likely to change with time please look at the internal program documentation if you need more details.

5.3 Mutation Rates

The directory *phylofriend/mutationrates* contains files with predefined mutation rates.

All 1 Mutation Rates

- 12-1.txt
- 37-1.txt
- 67-1.txt
- 111-1.txt

In these files all mutation rates are set to 1 for the corresponding set of markers. They are useful for simple mutation counting.

Average Mutation Rates

- 12-average.txt
- 37-average.txt
- 67-average.txt
- 111-average.txt

In these files average mutation rates are used for the corresponding set of markers. The mutation rates were taken from [5]. The distance matrix contains years but these years should not be interpreted as TMRCA (Time to Most Recent Common Ancestor). It is more like a *How long do I have to wait on average before such a genetic distance occurs?*

These mutation rates are useful for genealogical purposes. During genealogical time frames (about 500 years) long time stable markers rarely change. If such an event happens a son may be immediately 1000 years away from his father in terms of genetic distance. This is a correct result because mutations are governed by coincidence and such events do happen from time to time but it does not make much sense to place a son far away from his family. So very stable markers are purposely underweighted by using average values.

12 Marker Single Mutation Rates

- 12.txt
- 37-12.txt
- 67-12.txt

• 111-12.txt

These files are basically the same as the average mutation rates files but for the first 12 markers the mutation rates are set per marker. This puts on the appropriate weight on very stable markers. The mutation rates were taken from Wikipedia[7]. Although there may be some doubt if data from Wikipedia can be trusted the first 12 markers are well known and they are in use for a long time. So I just adopted them. The values should be good enough for most purposes.

For each file the 12 marker mutation rates were multiplied by a calibration factor so that their average value matches that from the rest of the file.

These files are useful for deeper history or if you observe changes on long time stable markers. If several persons share the same value on a very stable marker they probably belong together. So try these mutation rates to see if the results make more sense than those obtained by using average markers.

5.4 CSV Input Format

Example:

```
id1, "Dirk Struve", Germany, R1b-CTS4528, 13, 24, 14, 11, 11-14, 12, ... id2, "Pyl. O. Friend", Germany, R1b-CTS4528, 13, 24, 14, 11, 11-14, ...
```

When importing a file in comma separated values format the first column must contain IDs. An arbitrary number of columns containing custom information may follow. The last columns must contain at least 12 Y-STR values in Family Tree DNA order. Rows containing comments are allowed.

Phylofriend will always try to parse the file as best as it can.

5.5 Text Format

Example:

```
Dirk_Struv 13 24 14 11 11 14 12 12 12 12 14 28 Pyl._O._Fr 13 24 14 11 11 14 12 12 12 12 14 28
```

The text format is a simplified format intended for easy parsing and to work well with other programs. For compatibility reasons the first column is exactly 10 characters long and contains only non Unicode characters. Spaces are transformed into underscores. The following columns contain Y-STR values separated by tabs.

5.6 PHYLIP Format

Example:

```
2
Dirk_Struv 0 0
Pyl._O._Fr 0 0
```

The first line contains the number of entries. An entry line contains an ID that is 10 characters long and contains only non Unicode characters. Spaces are transformed into underscores. The columns containing genetic distances are separated by tabs. For readability reasons Phylofriend writes only integers. If you need more precision you can scale the distance by using the *cal* option.

References

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- [4] Anatole A. Klyosov, DNA Genealogy, Mutation Rates, and Some Historical Evidence Written in Y-Chromosome, Part I: Basic Principles and the Method. Journal of Genetic Genealogy, 5(2):186-216, 2009.
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- [7] Wikipedia, *List of Y-STR markers*. Date accessed: 2014-08-15.
- [8] Bruce Walsh, *Details on the various assumptions used in computing TMRCA*. Family Tree DNA, 2002, Date accessed: 2014-08-07.