

MendelGeneticCounseling ASHG Pre Workshop Installation and Checks

last update: September 12 2019, 3:13pm

As of the time of the last update to this tutorial, Open Mendel supports Julia versions 1.0, 1.1 and 1.2, but it is currently an unregistered package.

Download files

Download the datasets BRCA, Cholesterol and SmallSample from https://github.com/OpenMendel/GeneticCounseling_ASHG2019 (https://github.com/OpenMendel/GeneticCounseling_ASHG2019)

How to execute commands in the Jupyter notebook:

To execute a notebook command, hold down `Shift` `Enter` within the box. This tutorial and corresponding modules have been checked with Julia versions 1.0, 1.1 and 1.2.

For convenience, have this notebook (OpenMendel_Instructions.ipynb) in the directory SmallSample.

NOTE: When Finished with this notebook. Go to the File tab and first select save and checkpoint to save your results. Then under the file tab, select close and halt to prevent copies of Jupyter notebook from running indefinitely in the background.

To install, press `]` to invoke the package manager mode and install these packages by typing:

```
add https://github.com/OpenMendel/SnpArrays.jl
add https://github.com/OpenMendel/MendelSearch.jl
add https://github.com/OpenMendel/MendelBase.jl
add https://github.com/OpenMendel/MendelGeneticCounseling.jl#ASHG2019
build SpecialFunctions
```

Check Julia version:

For reproducibility, check the machine information below. Please report any issues running this notebook or the module to Janet Sinsheimer PhD. (jsinshei@g.ucla.edu).

```
In [1]: versioninfo()

Julia Version 1.2.0
Commit c6da87ff4b (2019-08-20 00:03 UTC)
Platform Info:
  OS: macOS (x86_64-apple-darwin18.6.0)
  CPU: Intel(R) Core(TM) i7 CPU           860  @ 2.80GHz
  WORD_SIZE: 64
  LIBM: libopenlibm
  LLVM: libLLVM-6.0.1 (ORCJIT, nehalem)
```

It is a good idea update all packages before running any analyses

In [2]:] update

```

Updating registry at ~/.julia/registries/General`
Updating git-repo `https://github.com/JuliaRegistries/General.git`
Updating git-repo `https://github.com/OpenMendel/MendelBase.jl`
Updating git-repo `https://github.com/OpenMendel/MendelSearch.jl`
Updating git-repo `https://github.com/OpenMendel/MendelGeneticCounseling.jl`
Updating git-repo `https://github.com/OpenMendel/SnpArrays.jl`
Resolving package versions...
Updating ~/.julia/environments/v1.2/Project.toml`
[no changes]
Updating ~/.julia/environments/v1.2/Manifest.toml`
[no changes]

```

Run a simple analysis to determine if MendelGeneticCounseling has been correctly installed.

We will now show how to set up the specific files and the options available as well as how to interpret the results during the workshop so we won't go over that information here.

Instead we will run a classic example of determining the risk of a fully penetrant disease for an individual just to make sure you will be able to do . Make certain necessary files are available. On a mac the appropriate command is "ls", which should also work if you are using a PC PowerShell. For a dos command window use "dir"

In [3]: ; ls

```

BRCA
BRCAExampleOut.txt
BreastCancerExamplePedigree.jpg
CholHeterozygousRisk.txt
Cholestrol
ControlBRCAExample.txt
ControlParametricPenetranceExample.txt
ControlSmallParametric.txt
HeterozygousRisk.txt
LICENSE
LocusBRCAExample.txt
LocusChol.txt
LocusSmall.txt
MendelGeneticCounselingTutorial.ipynb
MendelGeneticCounselingTutorial.pdf
OpenMendel_Instructions.ipynb
PedBRCAExample.csv
PedChol.csv
PedSmall.csv
PenBRCAExample.csv
PhenoBRCAExample.txt
PhenoChol.txt
PhenoSmall.txt
README.md
SmallSample

```

The files required for our test are PedSmall.csv, PhenoSmall.txt, LocusSmall.txt, and ControlSmallParametric.txt. If they aren't in your current directory, either move to the directory where the files are located or copy your files into the current working directory. Once you have the files in place, invoke the command:

In [4]: **using** MendelGeneticCounseling

```
In [5]: GeneticCounseling("ControlSmallParametric.txt")
```

```
Welcome to OpenMendel's  
Genetic Counseling Analysis Option
```

```
Reading the data.
```

```
The current working directory is "/Users/janets/Documents/janet/short_courses_20  
19/ASHGproposal/GeneticCounseling_ASHG2019-master".
```

```
Keywords modified by the user:
```

```
control_file = ControlSmallParametric.txt  
glm_link = IdentityLink  
glm_mean = 0.999999  
glm_response = BinomialDist  
glm_trait = Trait  
glm_trials = 1  
locus_file = LocusSmall.txt  
output_file = HeterozygousRisk.txt  
pedigree_file = PedSmall.csv  
phenotype_file = PhenoSmall.txt
```

```
no penetrance file
```

```
Analyzing the data.
```

```
The risk = 0.03892.
```

```
Mendel's analysis is finished.
```

Results

You should get a file with the results as well as seeing results displayed above. The correct answer is 0.03892. If you fail to get this answer, don't get any output, or get error messages, please contact Janet Sinsheimer at jsinshei@g.ucla.edu.

NOTE: When Finished with this notebook. Go to the File tab and first select save and checkpoint to save your results. Then under the file tab, select close and halt to prevent copies of Jupyter notebook from running indefinitely in the background