Guide for trident v0.28.0

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1 Poseidon package repositories

Trident generally requires Poseidon datasets to work with. Most trident subcommands therefore have a central parameter, called --baseDir or simply -d to specify one or more base directories to look for Poseidon packages. For example, if all Poseidon packages live inside a repository at /path/to/poseidon/packages you would simply say trident <subcommand> -d /path/to/poseidon/dirs/ and trident would automatically search all subdirectories inside of the repository for valid poseidon packages (as identified by valid POSEIDON.yml files).

You can arrange a poseidon repository in a hierarchical way. For example:

You can use this structure to select only the level of packages you're interested in, and you can make use of the fact that -d can be given multiple times.

Let's use the list command to list all packages in the modern and Reference_Genomes:

```
trident list -d /path/to/poseidon/packages/modern \
  -d /path/to/poseidon/packages/ReferenceGenomes --packages
```

2 Analysing your own dataset outside of the main repository

Being able to specify one or multiple repositories is often not enough, as you may have your own data to co-analyse with the main repository. This is easy to do, as you simply need to provide your own genotype data as yet another poseidon package to be added to your trident list command. For example, let's say you have genotype data in EIGENSTRAT format (trident supports EIGENSTRAT and PLINK as formats.):

```
~/my_project/my_project.geno
~/my_project/my_project.snp
~/my_project/my_project.ind
```

then you can make that to a skeleton Poseidon package with the **init** command. You can also do it manually by simply adding a POSEIDON.yml file, with for example the following content:

```
poseidonVersion: 2.5.0
title: My_awesome_project
description: Unpublished genetic data from my awesome project
contributor:
    - name: Stephan Schiffels
        email: schiffels@institute.org
packageVersion: 0.1.0
lastModified: 2020-10-07
genotypeData:
    format: EIGENSTRAT
    genoFile: my_project.geno
    snpFile: my_project.snp
    indFile: my_project.ind
jannoFile: my_project.janno
bibFile: sources.bib
```

Two remarks: 1) all file paths are considered *relative* to the directory in which POSEIDON.yml resides. Here I assume that you put this file into the same directory as the three genotype files. 2) Besides the genotype data files there are two (technically optional) files referenced by this example POSEIDON.yml file: sources.bib and my_project.janno. Of course you can add them manually - init automatically creates empty dummy versions.

Once you have set up your own "Poseidon" package (which is really only a skeleton so far), you can add it to your trident analysis, by simply adding your project directory to the command using -d:

```
trident list -d /path/to/poseidon/packages/modern \
  -d /path/to/poseidon/packages/ReferenceGenomes
  -d ~/my_project --packages
```

3 Package creation and manipulation commands

3.1 Init command

init creates a new, valid poseidon package from genotype data files. It adds a valid POSEIDON.yml file, a dummy janno file for context information and an empty .bib file for literature references.

Click here for command line details

```
Usage: trident init (-r|--inFormat ARG) (-g|--genoFile ARG) (-s|--snpFile ARG) (-i|--indFile ARG) [--snpSet ARG] (-o|--outPackagePath ARG)
```

```
Available options:
```

-h,--help Show this help text -r,--inFormat ARG the format of the input genotype data: EIGENSTRAT or PLINK the input geno file path -g,--genoFile ARG -s,--snpFile ARG the input snp file path -i,--indFile ARG the input ind file path --snpSet ARG the snpSet of the new package: 1240K, HumanOrigins or Other. Default: Other -o,--outPackagePath ARG the output package directory path the output package name - this is optional: If no -n,--outPackageName ARG name is provided, then the package name defaults to the basename of the (mandatory) --outPackagePath argument

--minimal
The command

```
trident init \
  -r EIGENSTRAT/PLINK \
  -g path/to/geno_file \
  -s path/to/snp_file \
  -i path/to/ind_file \
  --snpSet 1240K|HumanOrigins|Other \
  -o path/to/new_package_name
```

requires the format -r (--inFormat) of your input data (either EIGENSTRAT or PLINK), the paths to the respective files in -g (--genoFile), -s (--snpFile), and -i (--indFile), and optionally the "shape" of these files (--snpSet), so if they cover the 1240K, the HumanOrigins or an Other SNP set.

should only a minimal output package be created?

	EIGENSTRAT	PLINK
genoFile	.geno	.bed
	.ind	$_{ m .fam}$

The output package of init is created as a new directory -o, which should not already exist, and gets the package title corresponding to the basename of -o. You can also set the title explicitly with -n. The --minimal flag causes init to create a minimal package with a very basic POSEIDON.yml and no .bib and .janno files.

3.2 Fetch command

fetch allows to download poseidon packages from a remote poseidon server.

Click here for command line details

```
Usage: trident fetch (-d|--baseDir DIR) [-f|--fetchString ARG] [--fetchFile ARG] [--remoteURL ARG] [-u|--upgrade] [--downloadAll] Download data from a remote Poseidon repository
```

Available options:

-h,--help Show this help text
-d,--baseDir DIR a base directory to search for Poseidon Packages
(could be a Poseidon repository)
-f,--fetchString ARG List of packages to be downloaded from the remote server. Package names should be wrapped in asterisks:

```
*package_title*. You can combine multiple values with
                           comma, so for example: "*package 1*, *package 2*,
                           *package_3*". fetchString uses the same parser as
                           forgeString, but does not allow excludes. If groups
                           or individuals are specified, then packages which
                           include these groups or individuals are included in
                           the download.
  --fetchFile ARG
                           A file with a list of packages. Works just as -f, but
                           multiple values can also be separated by newline, not
                           just by comma. -f and --fetchFile can be combined.
  --remoteURL ARG
                           URL of the remote Poseidon
                           server (default: "https://c107-224.cloud.gwdg.de")
  -u,--upgrade
                           overwrite outdated local package versions
  --downloadAll
                           download all packages the server is offering
It works with
trident fetch -d ... -d ... \
  -f "*package_title_1*,*package_title_2*,*package_title_3*,group_name,<Individual1>" \
  --fetchFile path/to/forgeFile
```

and the entities you want to download must be listed either in a simple string with comma-separated values (-f/--fetchString) or in a text file (--fetchFile). Entities are specified using a special syntax: Package titles are wrapped in asterisks: $package_title$ (see also the documentation of forge below), group names are spelled as is, and individual names are wrapped in angular brackets, liks <Individual1>. Fetch will figure out which packages need to be downloaded to include all specified entities. --downloadAll causes fetch to ignore -f and download all packages from the server. The downloaded packages are added in the first (!) -d directory, but downloads are only performed if the respective packages are not already present in an up-to-date version in any of the -d dirs.

Note that trident fetch makes most sense in combination with trident list --remote: First one can inspect what is available on the server, then one can create a custom fetch command.

fetch also has the optional arguments --remote https:://..." do name an alternative poseidon server. The default points to the DAG server.

To overwrite outdated package versions with fetch, the -u/--upgrade flag has to be set. Note that many file systems do not offer a way to recover overwritten files. So be careful with this switch.

3.3 Forge command

-r,--inFormat ARG

forge creates new poseidon packages by extracting and merging packages, populations and individuals from your poseidon repositories.

Click here for command line details

the format of the input genotype data: EIGENSTRAT or

PLINK

-g,--genoFile ARG -s,--snpFile ARG -i,--indFile ARG --snpSet ARG

the input geno file path the input snp file path the input ind file path

the snpSet of the new package: 1240K, HumanOrigins or

--forgeFile ARG

Other. Default: Other A file with a list of packages, groups or individual

-f,--forgeString ARG

samples. Works just as -f, but multiple values can also be separated by newline, not just by comma. Empty lines are ignored and comments start with "#", so everything after "#" is ignored in one line.

List of packages, groups or individual samples to be combined in the output package. Packages follow the syntax *package_title*, populations/groups are simply group_id and individuals <individual_id>. You can combine multiple values with comma, so for example: "*package_1*, <individual_1>, <individual_2>,

group_1". Duplicates are treated as one entry.

Negative selection is possible by prepending "-" to the entity you want to exclude (e.g. "*package_1*, -<individual 1>, -group 1"). forge will apply excludes and includes in order. If the first entity is negative, then forge will assume you want to merge all individuals in the packages found in the baseDirs (except the ones explicitly excluded) before the exclude entities are applied. An empty forgeString will therefore merge all available individuals.

To extract specific SNPs during this forge operation, provide a Snp file. Can be either Eigenstrat (file ending must be '.snp') or Plink (file ending must be '.bim'). When this option is set, the output package will have exactly the SNPs listed in this file. Any SNP not listed in the file will be excluded. If option '--intersect' is also set, only the SNPs overlapping between the SNP file and the forged

packages are output.

--intersect

Whether to output the intersection of the genotype files to be forged. The default (if this option is not set) is to output the union of all SNPs, with genotypes defined as missing in those packages which do not have a SNP that is present in another package. With this option set, the forged dataset will typically have fewer SNPs, but less missingness. the format of the output genotype data: EIGENSTRAT or PLINK. Default: PLINK

should only a minimal output package be created? should only the resulting genotype data be returned? This means the output will not be a Poseidon package

the output package directory path

the output package name - this is optional: If no name is provided, then the package name defaults to the basename of the (mandatory) --outPackagePath argument

-w,--warnings --no-extract

Show all warnings for merging genotype data Skip the selection step in forge. This will result in outputting all individuals in the relevant packages, and hence a superset of the requested

5

--selectSnps ARG

--outFormat ARG

--minimal --onlyGeno

-o,--outPackagePath ARG -n,--outPackageName ARG individuals/groups. It may result in better performance in cases where one wants to forge entire packages or almost entire packages. Note that this will also ignore any ordering in the output groups/individuals. With this option active, individuals from the relevant packages will just be written in the order that they appear in the original packages.

forge can be used with

```
trident forge -d ... -d ... \
  -f "*package_name*, group_id, <individual_id>" \
  --forgeFile path/to/forgeFile \
  -o path/to/new_package_name
```

where the entities (packages, groups/populations, individuals/samples) you want in the output package can be denoted either as as simple string with comma-separated values (-f/--forgeString) or in a text file (--forgeFile).

Including one or multiple Poseidon packages with -d is not the only way to include data for a forge operation. It is also possible to include unpackaged genotype data directly with -r + -g + -s + -i (+ --snpSet). This makes the following example possible, where we merge data from one Poseidon package and two genotype datasets.

```
trident forge \
  -d 2017_GonzalesFortesCurrentBiology \
  -r PLINK \
  -g 2017_HaberAJHG/2017_HaberAJHG.bed \
  -s 2017_HaberAJHG/2017_HaberAJHG.bim \
  -i 2017_HaberAJHG/2017_HaberAJHG.fam \
  -r PLINK \
  -g 2018_VeeramahPNAS/2018_VeeramahPNAS.bed \
  -i 2018_VeeramahPNAS/2018_VeeramahPNAS.fam \
  -s 2018_VeeramahPNAS/2018_VeeramahPNAS.bim \
  -f "<STR241.SG>,<ERS1790729.SG>,Iberia_HG.SG" \
  -o testpackage \
  --onlyGeno
```

3.3.1 The forge selection language

Entities in the --forgeString or the --forgeFile have to be marked in a certain way:

- Each package is surrounded by *, so if you want all individuals of 2019_Jeong_InnerEurasia in the output package you would add *2019_Jeong_InnerEurasia* to the list.
- Groups/populations are not specially marked. So to get all individuals of the group Swiss_Roman_period, you would simply add Swiss_Roman_period.
- Individuals/samples are surrounded by < and >, so ALA026 becomes <ALA026>.

Do not forget to wrap the forgeString in quotes.

You can either use -f/--forgeString or --forgeFile. In the file each line is treated as a separate forgeString, empty lines are ignored and #s start comments. So this is a valid forgeFile:

```
# Packages
*package1*, *package2*

# Groups and individuals from other packages beyond package1 and package2
group1, <individual1>, group2, <individual2>, <individual3>

# group2 has two outlier individuals that should be ignored
```

```
-<bad_individual1> # This one has very low coverage
-<bad individual2> # This one is from a different time period
```

By prepending - to the bad individuals, we can exclude them from the forged package. forge figures out the final list of samples to include by executing all forge-entities in order. So an entity list *PackageA*,-<Individual1>,GroupA may result in a different outcome than *PackageA*,GroupA,-<Individual1>, depending on whether <Individual1> belongs to GroupA or not. If the forge entity list starts with a negative entity, or if the entity list is empty, forge will implicitly assume you want to include all individuals in all packages found in the baseDirs (except the ones explicitly excluded, of course). An empty forgeString will therefore merge all available individuals.

3.3.2 Other options

Just as for init the output package of forge is created as a new directory -o. The title can also be explicitly defined with -n.

--minimal allows for the creation of a minimal output package without .bib and .janno. This might be especially useful for data analysis pipelines, where only the genotype data is required. Even more basic output comes with --onlyGeno, which means that only the genotype data is returned without any Poseidon package.

forge has a an optional flag --intersect, that defines, if the genotype data from different packages should be merged with an **union** or an **intersect** operation. The default (if this option is not set) is to output the union of all SNPs, with genotypes defined as missing in samples from packages which do not have a SNP that is present in another package. With this option set, on the other hand, the forged dataset will typically have fewer SNPs, but less missingness.

--intersect also influences the automatic determination of the snpSet field in the POSEIDON.yml file for the resulting package. If the snpSets of all input packages are identical, then the resulting package will just inherit this configuration. Otherwise forge applies the following pairwise merging logic:

Input snpSet A	Input snpSet B	intersect	Ouput snpSet
Other	*	*	Other
1240K	HumanOrigins	True	HumanOrigins
1240K	HumanOrigins	False	1240K

--selectSnps allows to provide forge with a SNP file in EIGENSTRAT (.snp) or PLINK (.bim) format to create a package with a specific selection. When this option is set, the output package will have exactly the SNPs listed in this file. Any SNP not listed in the file will be excluded. If --intersect is also set, only the SNPs overlapping between the SNP file and the forged packages are output.

Merging genotype data across different data sources and file formats is tricky. forge is more verbose about potential issues, if the -w/--warnings flag is set.

3.4 Genoconvert command

genoconvert converts the genotype data in a Poseidon package to a different file format. The respective entries in the POSEIDON.yml file are changed accordingly.

Click here for command line details

```
Usage: trident genoconvert [-d|--baseDir DIR]

[(-r|--inFormat ARG) (-g|--genoFile ARG)

(-s|--snpFile ARG) (-i|--indFile ARG)

[--snpSet ARG]] --outFormat ARG [--onlyGeno]

[--removeOld]
```

Convert the genotype data in a Poseidon package to a different file format

```
Available options:
```

```
-h,--help Show this help text
```

```
a base directory to search for Poseidon Packages
-d,--baseDir DIR
                         (could be a Poseidon repository)
-r,--inFormat ARG
                         the format of the input genotype data: EIGENSTRAT or
                         PLINK
-g,--genoFile ARG
                         the input geno file path
-s,--snpFile ARG
                         the input snp file path
-i,--indFile ARG
                         the input ind file path
--snpSet ARG
                         the snpSet of the new package: 1240K, HumanOrigins or
                         Other. Default: Other
--outFormat ARG
                         the format of the output genotype data: EIGENSTRAT or
                         PLINK.
                         should only the resulting genotype data be returned?
--onlyGeno
                         This means the output will not be a Poseidon package
--removeOld
                         Remove the old genotype files when creating the new
                         ones
```

With the default setting

```
trident genoconvert -d ... -d ... --outFormat EIGENSTRAT|PLINK
```

all packages in -d will be converted to the desired --outFormat (either EIGENSTRAT or PLINK), if the data is not already in this format. This includes updating the respective POSEIDON.yml files.

Instead of -d to change Poseidon packages, the combination -r + -g + -s + -i (+ --snpSet) allows to directly convert genotype data that is not wrapped in a Poseidon package. See this example:

```
trident genoconvert \
  -r PLINK \
  -g 2018_Mittnik_Baltic/Mittnik_Baltic.bed \
  -s 2018_Mittnik_Baltic/Mittnik_Baltic.bim \
  -i 2018_Mittnik_Baltic/Mittnik_Baltic.fam \
  --outFormat EIGENSTRAT
```

The "old" data is not deleted, but kept around. That means conversion will result in a package with both PLINK and EIGENSTRAT data, but only one is linked in the POSEIDON.yml file, and that is what will be used by trident. To delete the old data in the conversion you can add the --removeOld flag.

Remember that the POSEIDON.yml file can also be edited by hand if you want to replace the genotype data in a package.

3.5 Update command

update automatically updates POSEIDON.yml files of one or multiple packages if the packages were changed.

Click here for command line details

Available options:

```
-h,--help Show this help text
-d,--baseDir DIR a base directory to search for Poseidon Packages
(could be a Poseidon repository)

--poseidonVersion ARG Poseidon version the packages should be updated to:
e.g. "2.5.3" (default: Nothing)

--ignorePoseidonVersion Read packages even if their poseidonVersion is not
compatible with the trident version. The assumption
is, that the package is already structurally adjusted
```

```
to the trident version and only the version number is
                         lagging behind.
--versionComponent ARG
                         Part of the package version number in the
                         POSEIDON.yml file that should be updated: Major,
                         Minor or Patch (see
                         https://semver.org) (default: Patch)
--noChecksumUpdate
                         Should update of checksums in the POSEIDON.yml file
                         be skipped
--ignoreGeno
                         ignore SNP and GenoFile
--newContributors ARG
                         Contributors to add to the POSEIDON.yml file in the
                         form "[Firstname Lastname](Email address);..."
                         Log text for this version jump in the CHANGELOG
--logText ARG
                         file (default: "not specified")
--force
                         Normally the POSEIDON.yml files are only changed if
                         the poseidonVersion is adjusted or any of the
                         checksums change. With --force a package version
                         update can be triggered even if this is not the case.
```

It can be called with a lot of optional arguments

```
trident update -d ... -d ... \
   --poseidonVersion "X.X.X" \
   --versionComponent Major/Minor/Patch \
   --noChecksumUpdate
   --ignoreGeno
   --newContributors "[Firstname Lastname](Email address);..."
   --logText "short description of the update"
   --force
```

By default update will not edit a package's POSEIDON.yml file, even when arguments like --versionComponent, --newContributors or --logText are explicitly set. This default exists to run the function on a large set of packages where only few of them were edited and need an active update. A package will only be modified by update if either

- any of the files with checksums (e.g. the genotype data) in it were modified,
- $\bullet \ \, \text{the $-$-poseidonVersion} \ \, \text{argument differs from the poseidonVersion in the package's POSEIDON.yml} \ \, \text{file} \\$
- or the --force flag was set in update.

If any of these applies to a package in the search directory (--baseDir/-d), it will be updated. This includes the following steps:

- If --poseidonVersion is different from the poseidonVersion field in the package, then that will be updated.
- The packageVersion will be incremented. If --versionComponent is not set, then it falls back to Patch, so a change in the last position of the three digit version number. Minor increments the middle, and Major the first position (see semantic versioning).
- The lastModified field will be updated to the current day (based on your computer's system time).
- The contributors in --newContributors will be added to the contributor field if they're not there already.
- If any checksums changed, then they will be updated. If certain checksums are not set yet, then they will be added. The checksum update can be skipped with --noChecksumUpdate or partially skipped for the genotype data with --ignoreGeno.
- The CHANGELOG.md file will be updated with a new row for the new version and the text in --logText (default: "not specified"), which will be appended as the first line of the file. If no CHANGELOG.md file exists, then it will be created and referenced in the POSEIDON.yml file.

:heavy_exclamation_mark: As update reads and rewrites POSEIDON.yml files, it may change their inner order, layout or even content (e.g. if they have fields which are not in the Poseidon package definition). Create a backup of the POSEIDON.yml file before running update if you are uncertain.

4 Inspection commands

4.1 List command

list lists packages, groups and individuals of the datasets you use, or of the packages available on the server.

Click here for command line details

Available options:

-h,--help Show this help text -d,--baseDir DIR a base directory to search for Poseidon Packages (could be a Poseidon repository) --remote list packages from a remote server instead the local file system URL of the remote Poseidon --remoteURL ARG server (default: "https://c107-224.cloud.gwdg.de") list all packages --packages --groups list all groups, ignoring any group names after the first as specified in the Janno-file --individuals list individuals -j,--jannoColumn JANNO_HEADER list additional fields from the janno files, using the Janno column heading name, such as Country, Site, Date_C14_Uncal_BP, Endogenous, ...

output table as tsv without header. Useful for piping

into grep or awk

--ignoreGeno ignore SNP and GenoFile

To list packages from your local repositories, as seen above you can run

```
trident list -d ... -d ... --packages
```

This will yield a table like this

--raw

Title	Date	Nr Individuals
2015_1000Genomes_1240K_haploid_pulldown 2016_Mallick_SGDP1240K_diploid_pulldown 2018_BostonDatashare_modern_published 	2020-08-10	280

so a nicely formatted table of all packages, their last update and the number of individuals in it.

To view packages on the remote server, instead of using directories to specify the locations of repositories on your system, you can use --remote to show packages on the remote server. For example

```
trident list --packages --remote
```

will result in a view of all published packages in our public online repository.

You can also list groups, as defined in the third column of EIGENSTRAT .ind files (or the first column of a PLINK .fam file), and individuals:

```
trident list -d ... -d ... --groups trident list -d ... -d ... --individuals
```

The --individuals flag also provides a way to immediately access information from the .janno files on the command line. This works with the -j/--jannoColumn option. For example adding --jannoColum Country --jannoColumn Date_C14_Uncal_BP to the commands above will add the Country and the Date_C14_Uncal_BP columns to the respective output tables.

Note that if you want a less fancy table, for example because you want to load this into Excel, or pipe into another command that cannot deal with the neat table layout, you can use the --raw option to output that table as a simple tab-delimited stream.

4.2 Summarise command

summarise prints some general summary statistics for a given poseidon dataset taken from the .janno files.

Click here for command line details

Usage: trident summarise (-d|--baseDir DIR) [--raw]

Get an overview over the content of one or multiple Poseidon packages

Available options:

-h,--help Show this help text

-d,--baseDir DIR a base directory to search for Poseidon Packages

(could be a Poseidon repository)

--raw output table as tsv without header. Useful for piping

into grep or awk

You can run it with

```
trident summarise -d ... -d ...
```

which will show you context information like – among others – the number of individuals in the dataset, their sex distribution, the mean age of the samples (for ancient data) or the mean coverage on the 1240K SNP array in a table. summarise depends on complete .janno files and will silently ignore missing information for some statistics.

You can use the --raw option to output the summary table in a simple, tab-delimited layout.

4.3 Survey command

survey tries to indicate package completeness (mostly focused on .janno files) for poseidon datasets.

Click here for command line details

```
Usage: trident survey (-d|--baseDir DIR) [--raw]
Survey the degree of context information completeness for Poseidon packages
```

Available options:

-h,--help Show this help text

-d,--baseDir DIR a base directory to search for Poseidon Packages

(could be a Poseidon repository)

--raw output table as tsv without header. Useful for piping

into grep or awk

Running

```
trident survey -d ... -d ...
```

will yield a table with one row for each package. See trident survey -h for a legend which cell of this table means what.

Again you can use the --raw option to output the survey table in a tab-delimited format.

4.4 Validate command

validate checks poseidon datasets for structural correctness.

Click here for command line details

Usage: trident validate (-d|--baseDir DIR) [--verbose]
Check one or multiple Poseidon packages for structural correctness

Available options:

-h,--help Show this help text

-d,--baseDir DIR a base directory to search for Poseidon Packages

(could be a Poseidon repository)

--verbose print more output to the command line

--ignoreGeno ignore SNP and GenoFile

--noExitCode do not produce an explicit exit code

You can run it with

```
trident validate -d ... -d ...
```

and it will either report a success (Validation passed) or failure with specific error messages to simplify fixing the issues.

validate tries to ensure that each package in the dataset adheres to the schema definition. Here is a list of what is checked:

- Presence of the necessary files
- Full structural correctness of .bib and .janno file
- Superficial correctness of genotype data files. A full check would be too computationally expensive
- Correspondence of BibTeX keys in .bib and .janno
- Correspondence of individual and group IDs in .janno and genotype data files

In fact much of this validation already runs as part of the general package reading pipeline invoked for many trident subcommands (e.g. forge). validate is meant to be more thorough, though, and will explicitly fail if even a single package is broken.

5 Analysis commands

All analysis commands (e.g. trident fstats) have been moved from trident to the analysis tool xerxes.