Guide for trident v1.5.4.0

2

3 Contents

4	1	Inst	allatio	n	2
5	2	The	triden	nt CLI	2
6		2.1	Genera	al notes	4
7			2.1.1	Logging and command line output	4
8			2.1.2	Package duplicates and versions	4
9			2.1.3	Individual/Sample duplicates	5
10			2.1.4	Group names in .fam files	5
11			2.1.5	Whitespaces in the .janno file	5
12	3	Pac	kage cı	reation and manipulation commands	5
13		3.1	Init co	mmand	5
14		3.2	Fetch o	command	7
15		3.3	Forge of	command	8
16			3.3.1	The forge selection language	11
17			3.3.2	Ordered output	13
18			3.3.3	Treatment of the genotype data while merging	13
19			3.3.4	Treatment of the .janno file while merging	14
20			3.3.5	Treatment of the .ssf file while merging	15
21			3.3.6	Treatment of the .bib file while merging	15
22			3.3.7	Output modes	15
23			3.3.8	Other options	16
24		3.4	Genoc	onvert command	17
25		3.5	Janno	coalesce command	18
26		3.6	Rectify	y command	19
27	4	Insp	pection	commands	21
28		4.1	List co	mmand	21
29		4.2	Summa	arise command	22
30		4.3	Survey	command	23
31		4.4	Validat	te command	23

$_{32}$ 1 Installation

- See the Poseidon website (https://www.poseidon-adna.org/#/trident) or the GitHub repository (https:
- //github.com/poseidon-framework/poseidon-hs) for up-to-date installation instructions.

$_{ iny 5}$ 2 The trident CLI

- Trident is a command line software tool structured in multiple subcommands. If you installed it properly you
- 37 can call it on the command line by typing trident. This will show an overview of the general options and all
- 38 subcommands, which are explained in detail below.

Usage: trident [--version] [--logMode MODE | --debug] [--errLength INT]

[--inPlinkPopName MODE] (COMMAND | COMMAND)

trident is a management and analysis tool for Poseidon packages. Report issues here: https://github.com/poseidon-framework/poseidon-hs/issues

Available options:

-h,--help-versionShow this help textShow version number

--logMode MODE How information should be reported: NoLog, SimpleLog,

DefaultLog, ServerLog or VerboseLog.

(default: DefaultLog)

--debug Short for --logMode VerboseLog.

--errLength INT After how many characters should a potential genotype

data parsing error message be truncated. "Inf" for no

truncation. (default: CharCount 1500)

--inPlinkPopName MODE Where to read the population/group name from the FAM

file in Plink-format. Three options are possible:

asFamily (default) | asPhenotype | asBoth.

Package creation and manipulation commands:

init Create a new Poseidon package from genotype data

fetch Download data from a remote Poseidon repository

forge Select packages, groups or individuals and create a

new Poseidon package from them

genoconvert Convert the genotype data in a Poseidon package to a

different file format

jannocoalesce Coalesce information from one or multiple janno files

to another one

rectify Adjust POSEIDON.yml files automatically to package

changes

Inspection commands:

list List packages, groups or individuals from local or

remote Poseidon repositories

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

survey Survey the degree of context information completeness
for Poseidon packages

validate Check Poseidon packages or package components for
structural correctness
```

trident allows to work directly with genotype data (see -p below), but it is optimized for the interaction with Poseidon packages, which wrap and contextualize the data. Most trident subcommands therefore have a central parameter, called --baseDir or simply -d to specify one or more base directories to look for 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).

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

This structure then allows to select only the level of packages you are interested in, even individual ones. -d
can be given multiple times, which is particularly useful as you may have your own data to co-analyse with
external reference data. In this case you simply need to provide your own genotype data as yet another Poseidon
package to be added to your trident command. For example, you may 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 transform that into 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: 2.7.1
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 in this POSEIDON.yml file are considered relative to the directory in which POSEIDON.yml resides. For this example we assume that this file is added into the same directory as the three genotype files. 2) Besides the genotype data files there are two (technically optional) files referenced in this 56 example: sources.bib and my_project.janno. Of course you can add them manually - init automatically 57 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, for example:

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

General notes 2.1

Logging and command line output

- For all subcommands the general argument --logMode defines how trident reports messages (to stderr) on the command line:
 - NoLog: Hides all messages.

65

67

70

71

74

75

76

77

78

79

80

81

- SimpleLog: Plain and simple output.
- DefaultLog: Adds the severity indicators (log levels) Info, Warning and Error before each message. This is the default setting. 68
 - ServerLog: Additionally adds timestamps before each message.
 - VerboseLog: Shows not just messages on the log levels Info, Warning and Error like the other modes, but also on the more verbose level Debug. Use this mostly relevant for debugging.
- --debug is short for --logMode VerboseLog to activate this important log level more easily.

2.1.2 Package duplicates and versions 73

- For trident multiple packages in a set of base directories can share the same title, if they have different packageVersion numbers. If the version numbers are also identical or missing, then trident stops with an exception.
- The trident subcommands genoconvert, list, rectify, survey and validate by default consider all versions of each Poseidon package in the given base directories. The --onlyLatest flag causes them to instead only consider the latest versions.
- fetch and forge generally consider all package versions. Their selection language (see below) allows for detailed version handling.

• summarize and jannocoalesce consider always only the latest package versions.

Individual/Sample duplicates 83

- Poseidon_IDs (so individual/sample names) within one package have to be unique, or trident will stop.
- We also discourage sample duplicates across packages in package repositories, but trident will generally continue with them. validate will fail though, if the --ignoreDuplicates flag is not set.
- forge offers a special mechanism to resolve sample duplicates within its selection language.

2.1.4Group names in .fam files

85

The .fam file of PLINK-formatted genotype data is used inconsistently across different popular aDNA software 89 tools to store group/population name information. The (global) trident option --inPlinkPopName with the arguments as Family (default), as Phenotype and as Both allows to control the reading of the population name 91 from PLINK .fam files. The subcommands that write genotype data (forge, genoconvert) have a corresponding 92 option --outPlinkPopName to specify this for the output.

Whitespaces in the .janno file 2.1.5

While reading the .janno file trident trims all leading and trailing whitespaces around individual cells. Also all instances of the No-Break Space unicode character will be removed. This means these whitespaces will not be preserved when a package is forged.

Package creation and manipulation commands

3.1 Init command

init creates a new Poseidon package from genotype data files. It adds a POSEIDON.yml file, a dummy .janno file for context information and an empty .bib file for literature references. 101

Command line details

```
Usage: trident init ((-p|--genoOne FILE) | --inFormat FORMAT --genoFile FILE
                      --snpFile FILE --indFile FILE) [--snpSet SET]
                    (-o|--outPackagePath DIR) [-n|--outPackageName STRING]
                    [--minimal]
```

Create a new Poseidon package from genotype data

۲A

vailable options:					
-h,help	Show this help text				
-p,genoOne FILE	One of the input genotype data files. Expects .bed,				
	.bim or .fam for PLINK and .geno, .snp or .ind for				
	EIGENSTRAT. The other files must be in the same				
	directory and must have the same base name.				
inFormat FORMAT	The format of the input genotype data: EIGENSTRAT or				
	PLINK. Only necessary for data input withgenoFile				
	+snpFile +indFile.				
genoFile FILE	Path to the input geno file.				

```
--snpFile FILE
                         Path to the input snp file.
--indFile FILE
                         Path to the input ind file.
                         The snpSet of the package: 1240K, HumanOrigins or
--snpSet SET
                         Other. Only relevant for data input with -p|--genoOne
                         or --genoFile + --snpFile + --indFile, because the
                         packages in a -d|--baseDir already have this
                         information in their respective POSEIDON.yml files.
                         (default: Other)
-o,--outPackagePath DIR
                         Path to the output package directory.
-n, -- outPackageName STRING
                         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. (default: Nothing)
                         Should the output data be reduced to a necessary
--minimal
                         minimum and omit empty scaffolding?
```

103 The command

```
trident init \
    --inFormat EIGENSTRAT/PLINK \
    --genoFile path/to/genoFile \
    --snpFile path/to/snpFile \
    --indFile path/to/indFile \
    --snpSet 1240K|HumanOrigins|Other \
    -o path/to/new_package_name
```

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

A simpler interface is available with -p (+ --snpSet), which only requires a path to one of the genotype data files and automatically discovers the others if they share the same base name:

```
trident init \
  -p path/to/genoFile \
  --snpSet 1240K|HumanOrigins|Other \
  -o path/to/new_package_name
```

The following file extensions are expected:

	EIGENSTRAT	PLINK
genoFile	.geno	.bed
${\rm snpFile}$.snp	.bim
indFile	.ind	.fam

The output package created by init is located in a new directory -o, which should not already exist when init

is called, and gets the package title corresponding to the basename of -o. You can also set the title explicitly with -n. 112

The --minimal flag causes init to create a minimal package with a very basic POSEIDON.yml and no .bib and .janno files. 114

Fetch command 3.2115

fetch allows to download Poseidon packages from a remote Poseidon server via a Web API. This server provides all packages in the Poseidon public archives. 117

Command line details

```
Usage: trident fetch (-d|--baseDir DIR)
                     (--downloadAll |
                        (--fetchFile FILE | (-f|--fetchString DSL)))
                     [--remoteURL URL] [--archive STRING]
```

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. --downloadAll Download all packages the server is offering. --fetchFile FILE 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. -f,--fetchString DSL 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. --remoteURL URL URL of the remote Poseidon server. (default: "https://server.poseidon-adna.org") --archive STRING The name of the Poseidon package archive that should be queried. If not given, then the query falls back

to the default archive of the server selected with --remoteURL. See the archive documentation at https://www.poseidon-adna.org/#/archive_overview for a list of archives currently available from the official Poseidon Web API. (default: Nothing)

It works with

```
trident fetch -d ... -d ... \
  -f "*package_title_1*,*package_title_2-1.0.1*,group_name,<individual1>"
```

and the entities you want to download must be listed either in a simple string of comma-separated values, which can be passed via -f/--fetchString, or in a text file (--fetchFile). Entities are then combined from these sources.

Entities are specified using a special syntax (see also the documentation of forge below): packages are wrapped in asterisks, with or without a version number appended after a dash (e.g. *package_title* or *package_title-1.2.3), group names are spelled as is, and individual names are wrapped in angular brackets (e.g. <individual1>). Fetch will figure out which packages need to be downloaded to include all specified entities.

--downloadAll, which can be given instead of -f and --fetchFile, causes fetch to download all packages from
the server. The downloaded packages are added in the first (!) -d directory (which gets created if it doesn't
exist), but downloads are only performed if the respective packages are not already present in the latest version
in any of the -d directories.

Note that trident fetch is usually used in a workflow with trident list --remote: First one inspects what is available on the server with list, to then compile a custom, targeted fetch command.

fetch has the optional arguments --remote https:://..." to name an alternative Poseidon server and --archive to select a specific Poseidon archive on the server.

135 3.3 Forge command

forge creates new Poseidon packages by extracting and merging packages, populations and individuals/samples from Poseidon repositories.

138 Command line details

Available options:

-h,--help Show this help text
-d,--baseDir DIR A base directory to search for Poseidon packages.
-p,--genoOne FILE One of the input genotype data files. Expects .bed,
.bim or .fam for PLINK and .geno, .snp or .ind for
EIGENSTRAT. The other files must be in the same
directory and must have the same base name.

--inFormat FORMAT The format of the input genotype data: EIGENSTRAT or PLINK. Only necessary for data input with --genoFile + --snpFile + --indFile. --genoFile FILE Path to the input geno file. --snpFile FILE Path to the input snp file. --indFile FILE Path to the input ind file. --snpSet SET The snpSet of the package: 1240K, HumanOrigins or Other. Only relevant for data input with -p|--genoOne or --genoFile + --snpFile + --indFile, because the packages in a -d|--baseDir already have this information in their respective POSEIDON.yml files. (default: Other) A file with a list of packages, groups or individual --forgeFile FILE 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. Multiple instances of -f and --forgeFile can be given. They will be evaluated according to their input order on the command line. -f,--forgeString DSL 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 (and no --forgeFile) will therefore merge all available individuals. If there are individuals in your input packages with equal individual id, but different main group or source package, they can be specified with the special syntax "<package:group:individual>". --selectSnps FILE 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. (default: Nothing) Whether to output the intersection of the genotype --intersect 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 --outFormat FORMAT PLINK. (default: PLINK) --onlyGeno Should only the resulting genotype data be returned? This means the output will not be a Poseidon package. --minimal Should the output Poseidon package be reduced to a necessary minimum? Should the output Poseidon package mimic the input --preservePyml package? With this option some fields of the source package's POSEIDON.yml file, its README file and its CHANGELOG file (if available) are copied to the output package. Only works for a singular source package. -o,--outPackagePath DIR Path to the output package directory. -n, -- outPackageName STRING 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. (default: Nothing) Skip the within-package selection step in forge. This --packagewise will result in outputting all individuals in the relevant packages, and hence a superset of the requested individuals/groups. It may result in better performance in cases where one wants to forge entire packages or almost entire packages. Details: Forge conceptually performs two types of selection: First, it identifies which packages in the supplied base directories are relevant to the requested forge, i.e. whether they are either explicitly listed using

still performs the first.

PackageName, or because they contain selected individuals or groups. Second, within each relevant package, individuals which are not requested are removed. This option skips only the second step, but

```
--outPlinkPopName MODE Where to write the population/group name into the FAM file in Plink-format. Three options are possible:

asFamily (default) | asPhenotype | asBoth. See also

--inPlinkPopName.

--ordered With this option, the output of forge is ordered according to the entities given.
```

139 forge can be used with

```
trident forge -d ... -d ... \
  -f "*package_name*, group_id, <individual_id>" \
  -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 a string on the command line (-f/--forgeString), or in an input text file (--forgeFile).
See the section below for the syntax of this selection language. Do not forget to wrap the --forgeString query
in quotes.

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 consider unpackaged genotype data directly with -p (+ --snpSet) or --inFormat +

--genoFile + --snpFile + --indFile (+ --snpSet). This makes the following example possible, where we

merge data from one Poseidon package and two unpackaged genotype datasets to get a new EIGENSTRAT

dataset.

```
trident forge \
  -d 2017_GonzalesFortesCurrentBiology \
  -p 2018_VeeramahPNAS/2018_VeeramahPNAS.fam \
  --inFormat PLINK \
  --genoFile 2017_HaberAJHG/2017_HaberAJHG.bed \
  --snpFile 2017_HaberAJHG/2017_HaberAJHG.bim \
  --indFile 2017_HaberAJHG/2017_HaberAJHG.fam \
  -f "<STR241.SG>,<ERS1790729.SG>,Iberia_HG.SG" \
  -o testpackage \
  --outFormat EIGENSTRAT \
  --onlyGeno
```

3.3.1 The forge selection language

156

157

158

The text in --forgeString and --forgeFile (and with a reduced syntax also in --fetchString and --fetchFile) are parsed as a domain specific query language that describes precisely which entities should be compiled in the output package of a given forge operation. The language has multiple syntactic elements and a specific evaluation logic.

In general a --forgeString query consists of multiple entities, separated by ,. The main entities are Poseidon packages, groups/populations and individuals/samples:

Each package title is surrounded by *: *package*. That means if you want all individuals of the Poseidon package 2019_Jeong_InnerEurasia in the output package you would add *2019_Jeong_InnerEurasia* to the query.

- Groups/populations are not specially marked: group. So to get all individuals of the group Swiss_Roman_Period, you would simply add Swiss_Roman_Period.
- Individuals/samples are surrounded by < and >: <individual>. ALA026 therefore becomes <ALA026>. A second way to denote individuals is with the more verbose and specific syntax <package:group:individual>. Such defined individuals take precedence over differently defined ones (so directly with <individual> or as a subset of *package* or group). This allows to resolve duplication issues precisely at least in cases where the duplicated individuals differ in source package or primary group.
- Package versions can be appended to package names, such as *package-1.2.3*.
- This also works with the verbose individual syntax: <package-1.2.3:group:individual>.

In the --forgeFile each line is treated as a separate forgeString, empty lines are ignored and # symbols start comments. So this is a valid example of a forgeFile:

```
# Packages
*pac1*, *pac2-1.2.3*

# Groups and individuals from other packages beyond pac1 and pac2
group1, <ind1>, group2, <ind2>, <pac3:group3:ind3>

# pac2 has two outlier individuals that should be ignored
-<ind4>  # This one has very low coverage
-<pac2:group4:ind5> # This one is from a different time period
```

By prepending - to entities, we can exclude them from the forged package (this feature is not available for fetch). forge figures out the final list of samples to include by interpreting all forge-entities in order. So an entity list *pac1*,-<ind1>,group1 may result in a different outcome than *pac1*,group1,-<ind1>, depending on whether <ind1> belongs to group1 or not.

174 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 **latest** versions of packages found in the base directories (except the ones explicitly excluded, of course).

177 The specific semantics of the various ways to include or exclude entities are as follows:

3.3.1.1 Inclusion queries

159

160

161

162

163

164

165

166

167

179

180

181

183

184

185

186

187

188

189

190

- *pac1*: Select all individuals in the latest version of package "pac1"
- *pac1-1.0.1*: Select all individuals in package "pac1" with version "1.0.1"
- group1: Select all individuals associated with "group1" in all latest versions of all packages
- <ind1>: Select the individual named "ind1", searching in all latest packages.
- <pac1:group1:ind1>: Select the individual named "ind1" associated with "group1" in the latest version of package "pac1"
- <pac1-1.0.1:group1:ind1>: Select the individual named "ind1" associated with "group1" in the package "pac1" with version "1.0.1"

3.3.1.2 Exclusion queries

- -*pac1*: Remove all individuals in all versions of package "pac1"
- -*pac1-1.0.1*: Remove only individuals in package "pac1" with version "1.0.1" (but leave other versions in)

- -group1: Remove all individuals associated with "group1" in all versions of all packages (not just the latest)
 - -<ind1>: Remove all individuals named "ind1" in all versions of all packages (not just the latest)
 - -<pac1:group1:ind1>: Remove the individual named "ind1" associated with "group1", searching in all versions of package "pac1"
- -<pac1-1.0.1:group1:ind1>: Remove the individual named "ind1" associated with "group1", but only if they are in "pac1" with version "1.0.1"
- 198 If a query results in multiple individuals with the same name, forge will throw an error.

199 3.3.2 Ordered output

193

194

195

214

215

216

217

218

- By default the order of samples in a Poseidon package created with forge depends on the order in which the relevant source packages are discovered by trident (e.g. when it crawls for packages in the -d base directories) and then the sample order within these packages.
- The option --ordered gives more control over the output order. It causes trident to output the resulting package with samples ordered according to the selection in -f or --forgeFile. This works through an alternative, slower sample selection algorithm that loops through the list of entities and checks for each entity which samples it adds or removes respectively to and from the final selection.
- For simple, positive selection, packages, groups and samples are added as expected. Negative selection removes samples from the list again. If an entity is selected twice via positive selection, then its first occurrence is considered for the ordering.
- 210 **3.3.2.1 Reordering samples in a package** One particular application of --ordered is the reordering of samples in an existing Poseidon package, here for example MyPac. We suggest the following workflow for this application:
 - 1. Generate a --forgeFile with the desired order of the samples in MyPac. This can be done manually or with any suitable tool. Here is an example, where we employ qjanno to generate a forge selection so that the samples are ordered alphabetically by their Poseidon_ID:

```
qjanno "SELECT '<'||Poseidon_ID||'>' FROM d(MyPac) ORDER BY Poseidon_ID" \
    --raw --noOutHeader > myOrder.txt
```

Use trident forge with --ordered and --preservePyml (see below) to create the package with the specified order:

```
trident forge -d MyPac --forgeFile myOrder.txt -o MyPac2 --ordered --preservePyml
```

3. Apply trident rectify to increment the package version number and document the reordering:

```
trident rectify -d MyPac2 --packageVersion Minor \
    --logText "reordered the samples alphabetically by Poseidon_ID"
```

MyPac2 then acts as a stand-in replacement for MyPac that only differs in the order of samples (and maybe the order of variables/fields in the POSEIDON.yml, .janno, .ssf or .bib files).

3.3.3 Treatment of the genotype data while merging

222 Forge performs a series of steps to merge the genotype data of multiple source files:

- 1. Genotype data from each package is streamed in parallel. Because our packages may have different SNP locations (specified by chromosome-position pairs) listed in their .bim/.snp file, we first perform a zipping-operation, whose behaviour depends on whether --intersect is set or not. Without --intersect, any SNP position listed in any package will be forwarded to the output, with missing values being filled in in all packages that do not list that particular SNP. With --intersect, only SNP positions that are present in all packages are considered. Note that relevant for this step is only whether a given SNP position is part of the genotype data, not whether the actual genotypes are missing or not.
- 2. At each SNP, the consensus alleles are selected, by collecting all reference and alternative alleles from all sources. If more than two non-dummy alleles (alleles different from N) are present in that collection, an error is thrown. If exactly two non-dummy alleles are present (which should be the case for binary SNPs), the two alleles are declared "reference" and "alternative" alleles for the output. If only one non-dummy allele is present, it is set to be the reference allele, and "N" is set to be the alternative.
- 3. All source genotype data is then read and recoded in terms of the two chosen consensus alleles. This will make sure that source data with flipped reference and alternative allele gets correctly merged in.
- 4. SNP IDs, as part of PLINK .bim files are checked across the source files. If all SNP IDs for a given SNP are missing, then the result will also be missing. If there is only one SNP ID present in some or all source packages, that ID gets forwarded to the output. In the (unusual) case that there are multiple different non-missing SNP IDs (of the form "rs" followed by a number), then a debug warning is output (which gets printed to the screen when --debug is selected), and simply the first value is chosen to be output into the forged .bim file. We decided not to throw an error in that case, because we consider the physical position of the SNP (specified by Chromosome and position) to be definitive, and the SNP ID to be of secondary importance.
- 5. Genetic positions, as part of PLINK .bim files are checked in a similar manner, with "0.0" being interpreted as missing.

3.3.4 Treatment of the . janno file while merging

forge merges and subsets .janno files along with the genotype data. If a package lacks a .janno file, then a
basic one will be created internally an on-the-fly based on the information in the genotype data, and used for
the output. Missing columns across packages will be filled with n/a.

For merging two .janno files $\bf A$ and $\bf B$ the following rules apply regarding undefined, arbitrary additional columns:

- If A has an additional column which is not in B then empty cells in the rows imported from B are filled with n/a.
- If **A** and **B** share additional columns with identical column name, then they are treated as semantically identical units and merged accordingly.
- In the resulting .janno file, all additional columns from both A and B are sorted alphabetically and appended after the normal, specified variables.

259 The following example illustrates the described behaviour:

A.janno

Poseidon_ID	Group_Name	Genetic_Sex	AdditionalColumn1	AdditionalColumn2
XXX011	POP1	M	A	D
XXX012	POP2	F	В	E

Poseidon_ID	Group_Name	Genetic_Sex	AdditionalColumn1	AdditionalColumn2
XXX013	POP1	M	С	F

261 B.janno

Poseidon_ID	Group_Name	Genetic_Sex	AdditionalColumn3	AdditionalColumn2
YYY022	POP5	F	G	J
YYY023	POP5	F	H	K
YYY024	POP5	\mathbf{M}	I	L

$_{262}$ A.janno + B.janno

Poseidon_ID	Group_Name	e Genetic_Sex	AdditionalColumn1	AdditionalColumn2	AdditionalColumn3
XXX011	POP1	M	A	D	n/a
XXX012	POP2	F	В	\mathbf{E}	n/a
XXX013	POP1	M	C	\mathbf{F}	n/a
YYY022	POP5	F	n/a	J	G
YYY023	POP5	F	n/a	K	Н
YYY024	POP5	M	n/a	L	I

3.3.5 Treatment of the .ssf file while merging

The Sequencing Source File (short .ssf file) is forged in exactly the same way as the .janno file. .ssf files
that are present are included in the forge product, following selection of those entities which are listed in the
poseidon_IDs columns. Columns that are only present in some packages, including those not defined in the
Poseidon package specification, are also included in the forged product in the same way as described for .janno
files above.

3.3.6 Treatment of the .bib file while merging

In the forge process all relevant samples for the output package are determined. This includes their .janno entries and therefore the information on the publication keys documented for them in the .janno Publication column. The output .bib file compiles only the relevant references for the samples in the output package. It includes the references exactly once and is sorted alphabetically by key.

274 3.3.7 Output modes

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

forge by default returns a new output package with a generic POSEIDON.yml file, the genotype data as created from the input and the selection, and a .janno file. If the input includes .bib or .ssf files, the output will as well.

Other output formats can be selected with these mutually exclusive flags:

280 --onlyGeno:

Only the genotype data is returned without any Poseidon package wrapping around it. This is especially useful for data analysis pipelines, where only the genotype data is required.

283 --minimal:

A minimal output package without .janno, .bib and .ssf. This wraps the genotype data in a very basic Poseidon package.

286 --preservePyml:

- A full Poseidon package just as the default, but with various settings copied from the source package. This only works in case of a single source package.
- For the specific task of sub-setting or reordering (see above) a singular, existing Poseidon package it can be useful to preserve some fields of the POSEIDON.yml file of this input package, as well as supplementary information in the README.md and the CHANGELOG.md file. These are typically discarded by forge, but can be copied over to the output package with the new --preservePyml output mode.
- 293 --preservePyml specifically preserves the following POSEIDON.yml fields:
 - description

294

- contributor
- packageVersion
- lastModified
- readmeFile
- changelogFile
- This does not include the package title, which can be easily set to be identical to the source with -n or -o
 if it is desired. The poseidonVersion field is also not copied, because trident can only ever produce output
 packages with the latest Poseidon schema version.

3.3.8 Other options

- forge has a an optional flag --intersect, that defines, if the genotype data from different packages should be merged with a union or an intersect operation. See *Treatment of the genotype data while merging* above.
- --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
$1240 \mathrm{K}$	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.

With --packagewise the within-package selection step in forge can be skipped. This will result in outputting all individuals in the relevant packages, and hence a superset of the requested individuals/groups. It may result in better performance in cases where one wants to forge entire packages.

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.

Command line details

```
Usage: trident genoconvert ((-d|--baseDir DIR) |
                             ((-p|--genoOne FILE) | --inFormat FORMAT
                               --genoFile FILE --snpFile FILE --indFile FILE)
                             [--snpSet SET]) --outFormat FORMAT [--onlyGeno]
                           [-o|--outPackagePath DIR] [--removeOld]
                           [--outPlinkPopName MODE] [--onlyLatest]
  Convert the genotype data in a Poseidon package to a different file format
Available options:
  -h,--help
                           Show this help text
  -d,--baseDir DIR
                           A base directory to search for Poseidon packages.
  -p,--genoOne FILE
                           One of the input genotype data files. Expects .bed,
                           .bim or .fam for PLINK and .geno, .snp or .ind for
                           EIGENSTRAT. The other files must be in the same
                           directory and must have the same base name.
  --inFormat FORMAT
                           The format of the input genotype data: EIGENSTRAT or
                           PLINK. Only necessary for data input with --genoFile
                           + --snpFile + --indFile.
  --genoFile FILE
                           Path to the input geno file.
  --snpFile FILE
                           Path to the input snp file.
  --indFile FILE
                           Path to the input ind file.
  --snpSet SET
                           The snpSet of the package: 1240K, HumanOrigins or
                           Other. Only relevant for data input with -p|--genoOne
                           or --genoFile + --snpFile + --indFile, because the
                           packages in a -d|--baseDir already have this
                           information in their respective POSEIDON.yml files.
                           (default: Other)
  --outFormat FORMAT
                           the format of the output genotype data: EIGENSTRAT or
                           PLINK.
  --onlyGeno
                           Should only the resulting genotype data be returned?
                           This means the output will not be a Poseidon package.
  -o,--outPackagePath DIR Path to the output package directory. This is
                           optional: If no path is provided, then the output is
                           written to the directories where the input genotype
```

data file (.bed/.geno) is stored. (default: Nothing)

```
--removeOld Remove the old genotype files when creating the new ones.

--outPlinkPopName MODE Where to write the population/group name into the FAM file in Plink-format. Three options are possible: asFamily (default) | asPhenotype | asBoth. See also --inPlinkPopName.

--onlyLatest Consider only the latest versions of packages, or the groups and individuals within the latest versions of packages, respectively.
```

320 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.

The "old" data is not deleted, but kept around. That means conversion can 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.

-p (+ --snpSet) or --inFormat + --genoFile + --snpFile + --indFile (+ --snpSet) allow to directly convert genotype data that is not wrapped in a Poseidon package and store it to a directory given in -o. See this example:

```
trident genoconvert \
  -p 2018_Mittnik_Baltic/Mittnik_Baltic.bed \
  --outFormat EIGENSTRAT \
  -o my_directory
```

3.5 Jannocoalesce command

-t,--targetFile FILE

-o,--outFile FILE

jannocoalesce merges information from one or multiple source .janno files into a target .janno file.

Command line details

An optional file to write the results to. If not

The target .janno file to fill.

```
specified, change the target file in place.
                         (default: Nothing)
                         A comma-separated list of .janno column names to
--includeColumns ARG
                         coalesce. If not specified, all columns that can be
                         found in the source and target will get filled.
--excludeColumns ARG
                         A comma-separated list of .janno column names NOT to
                         coalesce. All columns that can be found in the source
                         and target will get filled, except the ones listed
                         here.
-f,--force
                         With this option, potential non-missing content in
                         target columns gets overridden with non-missing
                         content in source columns. By default, only missing
                         data gets filled-in.
--sourceKey ARG
                         The .janno column to use as the source key.
                         (default: "Poseidon_ID")
                         The .janno column to use as the target key.
--targetKey ARG
                         (default: "Poseidon ID")
--stripIdRegex ARG
                         An optional regular expression to identify parts of
                         the IDs to strip before matching between source and
                         target. Uses POSIX Extended regular expressions.
```

 $^{332}\,$ A most basic run may just include two arguments:

```
trident jannocoalesce \
   --sourceFile path/to/source.janno \
   --targetFile path/to/target.janno
```

jannocoalesce generally works by reading a source .janno file with -s|--sourceFile (or all .janno files in a -d|--baseDir) and a target .janno file with -t|--targetFile.

It then merges these files by a key column, which can be selected with --sourceKey and --targetKey. The
default for both of these key columns is the Poseidon_ID. In case the entries in the key columns slightly and
systematically differ, e.g. because the Poseidon_IDs in either have a special suffix (for example _SG), then the
--stripIdRegex option allows to strip these with a regular expression to thus match the keys.

jannocoalesce generally attempts to fill all empty cells in the target .janno file with information from the source. --includeColumns and --excludeColumns allow to select specific columns for which this should be done. In some cases it may be desirable to not just fill empty fields in the target, but overwrite the information already there with the -f|--force option. If the target file should be preserved, then the output can be directed to a new output .janno file with -o|--outFile.

3.6 Rectify command

rectify automatically harmonizes POSEIDON.yml files of one or multiple packages. This is not an automatic update from one Poseidon version to the next, but rather a clean-up wizard after manual modifications.

347 Command line details

```
Usage: trident rectify (-d|--baseDir DIR) [--ignorePoseidonVersion]
                       [--poseidonVersion ?.?.?]
                       [--packageVersion VPART [--logText STRING]]
                       [--checksumAll | [--checksumGeno] [--checksumJanno]
                         [--checksumSSF] [--checksumBib]]
                       [--newContributors DSL] [--onlyLatest]
  Adjust POSEIDON.yml files automatically to package changes
Available options:
  -h,--help
                           Show this help text
  -d,--baseDir DIR
                           A base directory to search for Poseidon packages.
  --ignorePoseidonVersion
                           Read packages even if their poseidonVersion is not
                           compatible with trident.
  --poseidonVersion ?.?.? Poseidon version the packages should be updated to:
                           e.g. "2.5.3".
  --packageVersion VPART
                           Part of the package version number in the
                           POSEIDON.yml file that should be updated: Major,
                           Minor or Patch (see https://semver.org).
  --logText STRING
                           Log text for this version in the CHANGELOG file.
  --checksumAll
                           Update all checksums.
  --checksumGeno
                           Update genotype data checksums.
  --checksumJanno
                           Update .janno file checksum.
  --checksumSSF
                           Update .ssf file checksum
  --checksumBib
                           Update .bib file checksum.
  --newContributors DSL
                           Contributors to add to the POSEIDON.yml file in the
                           form "[Firstname Lastname] (Email address);...".
  --onlyLatest
                           Consider only the latest versions of packages, or the
                           groups and individuals within the latest versions of
                           packages, respectively.
```

It can be called with a lot of optional arguments. Note that rectify by default does **not** apply any changes if none of these arguments are set. Each change requires explicit opt-in.

```
trident rectify -d ... -d ... \
    --poseidonVersion "X.X.X" \
    --packageVersion Major|Minor|Patch \
    --logText "short description of the update" \
    --checksumAll \
    --newContributors "[Firstname Lastname](Email address);..."
```

The following arguments determine which fields of the POSEIDON.yml file should be modified:

351

352

353

354

- --poseidonVersion allows a simple change of the poseidonVersion field in the POSEIDON.yml file.
- --packageVersion increments the package version number in the first, the second or the third position. It can optionally be called with --logText, which appends an entry to the CHANGELOG file for the respective package version update. --logText also creates a new CHANGELOG.md file if it does not exist

355 yet.

356

357

- --checksumGeno, --checksumJanno, --checksumSSF and --checksumBib add or modify the respective checksum fields in the POSEIDON.yml file. --checksumAll is a wrapper to call all of them at once.
- --newContributors adds new contributors.

As rectify 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.yml specification). Create a backup of the POSEIDON.yml file before running rectify if you are uncertain if this might affect you negatively.

₂ 4 Inspection commands

363 4.1 List command

list lists packages, groups and individuals of local datasets, or of packages available in the archives on the web

366 Command line details

```
Usage: trident list ((-d|--baseDir DIR) | --remote [--remoteURL URL]
                      [--archive STRING])
                    (--packages | --groups | --individuals
                      [--fullJanno | [-j|--jannoColumn COLNAME]]) [--raw]
                     [--onlyLatest]
  List packages, groups or individuals from local or remote Poseidon
  repositories
Available options:
  -h,--help
                           Show this help text
  -d,--baseDir DIR
                           A base directory to search for Poseidon packages.
  --remote
                           List packages from a remote server instead the local
                           file system.
  --remoteURL URL
                           URL of the remote Poseidon server.
                            (default: "https://server.poseidon-adna.org")
  --archive STRING
                           The name of the Poseidon package archive that should
                           be queried. If not given, then the query falls back
                           to the default archive of the server selected with
                            --remoteURL. See the archive documentation at
                           https://www.poseidon-adna.org/#/archive_overview for
                           a list of archives currently available from the
                           official Poseidon Web API. (default: Nothing)
  --packages
                           List all packages.
  --groups
                           List all groups, ignoring any group names after the
                           first as specified in the .janno-file.
  --individuals
                           List all individuals/samples.
  --fullJanno
                           output all Janno Columns
  -j,--jannoColumn COLNAME List additional fields from the janno files, using
```

the .janno column heading name, such as "Country",

"Site", "Date_C14_Uncal_BP", etc..

--raw Return the output table as tab-separated values

without header. This is useful for piping into grep

or awk.

--onlyLatest Consider only the latest versions of packages, or the

groups and individuals within the latest versions of

packages, respectively.

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

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

- This will yield a nicely formatted table of all packages, their version and the number of individuals in them.
- You can use --remote to show packages on the remote server. For example

```
trident list --packages --remote --archive "community-archive"
```

- will result in a view of all packages available in one of the public Poseidon archives. Just as for fetch, the
 --archive flag allows to choose which public archive to query.
- 372 Independent of whether you query a local or an online archive, you can not just list packages, but also groups,
- $_{\rm 373}$ $\,$ as defined in the third column of EIGENSTRAT .ind files (or the first/last column of a PLINK .fam file), and
- individuals with the flags --groups and --individuals (instead of --packages).
- The --individuals flag additionally provides a way to immediately access information from .janno files
- on the command line. This works with the -j|--jannoColumn option. For example adding -j Country -j
- Date_C14_Uncal_BP to the commands above will add the Country and the Date_C14_Uncal_BP columns to the
- respective output tables. --fullJanno outputs all available columns.
- Note that if you want a less ornate table, for example because you want to load this into Excel, or pipe into
- another command that cannot deal with the table layout, you can use the --raw option to output that table as
- 381 a simple tab-delimited stream.

382 4.2 Summarise command

- summarise prints some general summary statistics for a given Poseidon dataset taken from the .janno files.
- 384 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.

--raw Return the output table as tab-separated values

without header. This is 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 or the mean coverage on the 1240K SNP array in a table. summarise
- depends on complete . janno files and will silently ignore missing information.
- 389 You can use the --raw option to output the summary table in a simple, tab-delimited layout.

390 4.3 Survey command

- 391 survey tries to indicate package completeness (mostly focused on .janno files) for Poseidon datasets.
- 392 Command line details

```
Usage: trident survey (-d|--baseDir DIR) [--raw] [--onlyLatest]

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.
--raw Return the output table as tab-separated values without header. This is useful for piping into grep or awk.
--onlyLatest Consider only the latest versions of packages, or the groups and individuals within the latest versions of
```

393 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.

packages, respectively.

397 4.4 Validate command

- validate checks Poseidon packages and individual package components for structural correctness.
- 399 Command line details

Check Poseidon packages or package components for structural correctness

Available options:

-h,--help Show this help text

-d,--baseDir DIR A base directory to search for Poseidon packages.

--ignoreGeno Ignore snp and geno file.

--fullGeno Test parsing of all SNPs (by default only the first

100 SNPs are probed).

--ignoreDuplicates Do not stop on duplicated individual names in the

package collection.

-c,--ignoreChecksums Whether to ignore checksums. Useful for speedup in

debugging.

--ignorePoseidonVersion Read packages even if their poseidonVersion is not

compatible with trident.

--pyml FILE Path to a POSEIDON.yml file.

-p,--genoOne FILE One of the input genotype data files. Expects .bed,

.bim or .fam for PLINK and .geno, .snp or .ind for EIGENSTRAT. The other files must be in the same directory and must have the same base name.

--inFormat FORMAT The format of the input genotype data: EIGENSTRAT or

PLINK. Only necessary for data input with --genoFile

+ --snpFile + --indFile.

--genoFile FILE Path to the input geno file.
--snpFile FILE Path to the input snp file.
--indFile FILE Path to the input ind file.
--janno FILE Path to a .janno file.

--ssf FILE Path to a .ssf file.
--bib FILE Path to a .bib file.

--noExitCode Do not produce an explicit exit code.

--onlyLatest Consider only the latest versions of packages, or the

groups and individuals within the latest versions of

packages, respectively.

400 You can run it with

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

- to check packages and it will either report a success (Validation passed) or failure with specific error messages.
- 402 Instead of validating entire packages with -d you can also apply it to individual files and package com-
- 403 ponents: --pyml (POSEIDON.yml), -p | --inFormat + --genoFile + --snpFile + --indFile (genotype
- data), --janno (.janno file), --ssf (.ssf file) or --bib (.bib file). In this case validate attempts to read and
- parse the respective files individually and reports any issues it encounters. Note that this considers the files in
- isolation and does not include any cross-file consistency checks.
- When applied to packages, validate tries to ensure that each package adheres to the Poseidon package
- specification. Here is a list of what is checked:

• Structural correctness of the POSEIDON.yml file.

410

411

415

416

417

- Presence of all files references in the POSEIDON.yml file.
 - Full structural correctness of .janno, .ssf and .bib file.
- Superficial correctness of genotype data files by parsing the first 100 SNPs. A full check that parses all SNPs can be triggered with the --fullGeno option. --ignoreGeno, on the other hand, causes validate to ignore the genotype data entirely, which speeds up the validation significantly.
 - Correspondence of BibTeX keys in .bib and .janno
 - Correspondence of sample IDs in .janno and .ssf.
 - Correspondence of sample 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 other trident subcommands (e.g. forge). validate is meant to be more thorough and brittle, though, and will explicitly fail if even a single package is broken. For special cases more flexibility can be enabled with the options --ignoreDuplicates, --ignoreChecksums and --ignorePoseidonVersion.
- Remember to run validate with --debug to get more information in case the default output is not sufficient to analyse an issue.