Poseidon package specification v2.7.1

PDF version of the standard at https://github.com/poseidon-framework/poseidon-schema generated on 2024-02-26 based on the Git commit 66afd9a

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I The Poseidon Standard v2.7.1

Poseidon is a solution for archaeogenetic genotype data organisation. This standard defines the core components of the Poseidon package.

A .pdf version of the latest instance of this document can be downloaded here.

Further details on genotype data, the .janno file and the .ssf file are documented on the Poseidon website.

The website also features a changelog documenting the changes across different schema versions here.

The key words MUST, MUST NOT, REQUIRED, SHALL, SHALL NOT, SHOULD, SHOULD NOT, RECOMMENDED, MAY, and OPTIONAL in this document are to be interpreted as described in RFC 2119.

1 The Poseidon package structure

A Poseidon package stores genotype data with context information for DNA samples from (ancient) (human) individuals. Packages are defined by the POSEIDON.yml file, which holds relative paths to all other files in a package.

A package therefore MUST contain:

- A POSEIDON.yml file to formally define the package
- Genotype data in PLINK or EIGENSTRAT format

It SHOULD additionally contain:

- A . janno file to store context information on spatiotemporal origin or sample quality
- A .bib file for literature references

It MAY also contain:

- A README.md file for arbitrary, additional context information
- A CHANGELOG.md file to document changes to the package
- A .ssf file with information on the underlying raw sequencing data

Here is an example of a package Switzerland_LNBA_Roswita in one directory:

```
Switzerland_LNBA_Roswita/POSEIDON.yml
Switzerland_LNBA_Roswita/Switzerland_LNBA.bed
Switzerland_LNBA_Roswita/Switzerland_LNBA.bim
Switzerland_LNBA_Roswita/Switzerland_LNBA.fam
Switzerland_LNBA_Roswita/Switzerland_LNBA.janno
Switzerland_LNBA_Roswita/Switzerland_LNBA.ssf
Switzerland_LNBA_Roswita/Switzerland_LNBA.bib
Switzerland_LNBA_Roswita/README.md
Switzerland_LNBA_Roswita/CHANGELOG.md
```

All text files in the package MUST be UTF-8 encoded.

2 The POSEIDON.yml file

The POSEIDON.yml file defines Poseidon packages by listing metainformation and relative paths in a standardised, machine-readable format.

- It MUST be a valid YAML file.
- Its mandatory and optional fields are documented in the POSEIDON_yml_fields.tsv file.

Here is an example for a POSEIDON.yml file:

```
poseidonVersion: 2.7.1
title: Switzerland_LNBA_Roswita
description: LNBA Switzerland genetic data not yet published
contributor:
  - name: Roswita Malone
    email: roswita.malone@example.org
    orcid: 1234-1234-1234-1234
  - name: Paul Panther
    email: paul.panther@example.edu
packageVersion: 1.1.2
lastModified: 2021-01-28
genotypeData:
  format: PLINK
  genoFile: Switzerland_LNBA_Roswita.bed
  genoFileChkSum: 95b093eefacc1d6499afcfe89b15d56c
  snpFile: Switzerland_LNBA_Roswita.bim
  snpFileChkSum: 6771d7c873219039ba3d5bdd96031ce3
  indFile: Switzerland_LNBA_Roswita.fam
```

indFileChkSum: f77dc756666dbfef3bb35191ae15a167

snpSet: 1240K

jannoFile : Switzerland_LNBA_Roswita.janno

jannoFileChkSum: 555d7733135ebcabd032d581381c5d6f
sequencingSourceFile: Switzerland_LNBA_Roswita.ssf

sequencingSourceFileChkSum: 19db1906240ee2f076e1a9659567dca4

bibFile: Switzerland_LNBA_Roswita.bib

bibFileChkSum: 70cd3d5801cee8a93fc2eb40a99c63fa

readmeFile: README.md
changelogFile: CHANGELOG.md

When a package is modified in any way (including updates of the context information in the .janno file), then the packageVersion field SHOULD be incremented and the lastModified field updated to the current date.

2.1 Package versioning

The packageVersion field is a mandatory entry of the POSEIDON.yml file. It denotes the version of the individual package, using a three-component versioning system derived from semantic versioning.

Each version number is comprised of three numbers, separated by a .. For example: 0.1.0, 1.0.0 or 2.1.3. The first number gives the Major, the second the Minor and the third the Patch component of the version number. For a Poseidon package these components SHOULD be incremented when the following changes occur:

- Major (e.g. 1.4.2 -> 2.0.0)
 - When samples are added to a package.
 - When samples are removed from a package.
 - When the genotype data (i.e. the contents of the .bed/.bim/.fam or .geno/.snp/.ind files) for any number of samples is changed.
- Minor (e.g. 1.4.2 -> 1.5.0)
 - When larger pieces of meta- or context information are added or modified in any package file, except the genotype data. For example:
 - * An entire .janno, .bib or .ssf file is added or replaced.
 - * Entire columns in the .janno or .ssf file are added or replaced.
 - * Primary publications for samples in the .janno and .bib file are added or replaced.
- Patch (e.g. 1.4.2 -> 1.4.3)
 - When smaller pieces of meta- or context information are added or modified in any package file, except the genotype data. For example:
 - * Individual entries in the .janno or .ssf file are added or replaced.
 - * Secondary publications for samples in the .janno and .bib file are added or replaced.
 - * BibTeX entries in the .bib file are modified.
 - * The package description changes in the POSEIDON.yml file.
 - * The CHANGELOG.md file is modified with additional information on previous entries.

When the packageVersion is changed, then the lastModified date MUST be updated and an entry to the CHANGELOG.md file SHOULD be added summarising the changes made.

Packages SHOULD start at packageVersion 0.1.0.

3 Genotype data

Genotype data in Poseidon packages is stored either in (binary) PLINK or EIGENSTRAT format.

	PLINK (binary)	EIGENSTRAT
genotype file	.bed (binary biallelic genotype table)	.geno (genotype file)
SNP file	.bim (extended MAP file)	.snp (snp file)
individual file	$. \\ {\tt fam} \ ({\rm sample \ information})$.ind (indiv file)

In addition to these files (and optionally their checksums), the POSEIDON.yml file SHOULD also provide a snpSet entry which determines the shape of the genotype file.

4 The . janno file

The .janno file is a tab-separated text file with a header line. It holds context information (variables/columns) for each sample (objects/rows) in a package.

- A set of strictly defined core variables (defined by column name) and their possible content are documented here: janno columns.tsv
- A .janno file MAY have all of these core variables, or only a subset of them.
- Only three columns MUST be present to make the file valid: Poseidon_ID, Group_Name and Genetic_Sex
- Arbitrary columns not defined here MAY be added as long as their column names do not clash with the defined ones.
- The column order is irrelevant.
- If information is unknown or a variable does not apply for a certain sample, then the respective cell(s) MAY be filled with n/a or simply an empty string.
- The order of the samples (rows) in the .janno file MUST be equal to the order in the genetic data files (.ind, .fam) in the package.
- The values in the columns **Poseidon_ID**, **Group_Name** and **Genetic_Sex** MUST be equal to the terms used in the genetic data files (.ind, .fam).
- Multiple predefined columns of the .janno file are list columns that can hold multiple values (either strings or numerics) separated by ;.
- The decimal separator for all floating point numbers MUST be ...

5 The .bib file

A BibTeX file with all references listed in the .janno file. The entry keys MUST fit the ones used in the .janno file.

Example:

```
@article{CassidyPNAS2015,
    doi = {10.1073/pnas.1518445113},
    url = {https://doi.org/10.1073%2Fpnas.1518445113},
    year = 2015,
    month = {dec},
    publisher = {Proceedings of the National Academy of Sciences},
    volume = {113},
    number = {2},
    pages = {368--373},
```

```
author = {Lara M. Cassidy and Rui Martiniano and Eileen M. Murphy and Matthew D.
Teasdale and James Mallory and Barrie Hartwell and Daniel G. Bradley},
title = {Neolithic and Bronze Age migration to Ireland and establishment of the
insular Atlantic genome},
journal = {Proceedings of the National Academy of Sciences}
}
```

To connect a sample in the package to this particular literature reference, the .janno file column Publication would have to be filled with CassidyPNAS2015.

6 The README.md file

A simple markdown file with informal, arbitrarily structured information accompanying the package.

Example:

This package contains a rather interesting set of samples relevant for the peopling of the Territory of Christmas Island in the Indian Ocean. We consider this especially relevant, because ...

7 The CHANGELOG.md file

A markdown file to document changes in the history of a package.

Example:

```
V 1.1.1: Fixed a spelling mistake in one site name: "Hosenacker" -> "Rosenacker"
V 1.1.0: Added mtDNA contamination estimation to the .janno file
V 1.0.0: Added spatial coordinates and age information to the .janno file and finalized a first stable version of the package
V 0.2.0: Added previously restricted sample L1337
V 0.1.0: Creation of the package
```

The structure with - V X.X.X: at the beginning of each line is not mandatory, but SHOULD be followed for reasons of interoperability.

8 The .ssf file

The .ssf file is another tab-separated text file with a header line. It stores sequencing source data, so metainformation about the raw sequencing data behind the genotypes in a Poseidon package. The primary entities in this table are sequencing entities, typically corresponding to DNA libraries or even multiple runs/lanes of the same library.

- The predefined columns are specified here: ssf_columns.tsv
- All columns of this schema are optional, so a .ssf MAY have all of these core variables, only a subset of them, or even none. It SHOULD have a poseidon_IDs column, though, to link the sequencing entities to the Poseidon package.
- The link to the individuals listed in the .janno-file (and therefore to the entire Poseidon package) is made through a many-to-many foreign-key relationship between the .janno column Poseidon_ID and the .ssf column poseidon_IDs. That means each entry in the .janno file can be linked to many rows in the .ssf file and vice versa.
- As in the .janno file arbitrary columns not defined here MAY be added to the .ssf file as long as their column names do not clash with the defined ones.
- The order of columns and rows is irrelevant.

- If information is unknown or a variable does not apply, then the respective cell(s) MAY be filled with n/a or simply an empty string.
- Multiple predefined columns of the .ssf file are list columns that can hold multiple values (either strings or numerics) separated by ;.
- $\bullet\,$ The decimal separator for all floating point numbers MUST be \dots

II Appendix

The following tables specify individual fields/variables/columns in the POSEIDON.yml, the .janno and the .ssf file.

An asterisk * after the field name indicates a mandatory field that a given file MUST include to be valid.

1 POSEIDON.yml file fields

POSEIDON.yml file fields

Field	Description
poseidonVersion*	Poseidon package format version (e.g. 2.0.1) type: String format: X.Y.Z
${ m title}^*$	title of the package type: String
description	description of the package (one or multiple sentences) type: String
contributor	list of contributors to the package (not the publication author, but the Poseidon package creator) type: Array
name*	name of one contributor subfield of: contributor type: String
email*	email of one contributor <u>subfield of</u> : contributor <u>type</u> : String <u>format</u> : Email
orcid	orcid of one contributor <u>subfield of</u> : contributor <u>type</u> : String <u>format</u> : ORCID
$package Version^*$	package version (should be changed/incremented when the package is changed) type: String format: X.Y.Z
lastModified	date of last modification of the package (should be updated when the package is changed) type: Date format: YYYY-MM-DD
genotypeData*	genotype data section

POSEIDON.yml file fields (continued)

Field	Description
format*	genotype data file format subfield of: genotypeData type: String format: EIGENSTRAT;PLINK
genoFile*	relative path to the geno file subfield of: genotypeData type: String format: Path
genoFileChkSum	md5 checksum of the geno file subfield of: genotypeData type: String format: md5 hash
snpFile*	relative path to the snp file subfield of: genotypeData type: String format: Path
snpFileChkSum	md5 checksum of the snp file subfield of: genotypeData type: String format: md5 hash
$indFile^*$	relative path to the ind file subfield of: genotypeData type: String format: Path
indFileChkSum	md5 checksum of the ind file subfield of: genotypeData type: String format: md5 hash
snpSet	SNP set in the genotype data subfield of: genotypeData type: String format: 1240K;HumanOrigins;Other
jannoFile	relative path to the .janno file $\frac{\text{type}}{\text{format}}$: Path
jannoFileChkSum	md5 checksum of the .janno file type: String format: md5 hash

${\it POSEIDON.yml file fields} \ (continued)$

Field	Description
sequencingSourceFile	relative path to the .ssf file type : String format : Path
${\it sequencing Source File Chk Sum}$	md5 checksum of the .ssf file type: String format: md5 hash
bibFile	relative path to the .bib file type : String format : Path
bibFileChkSum	md5 checksum of the .bib file type: String format: md5 hash
readmeFile	relative path to the README file type: String format: Path
${\it changelogFile}$	relative path to the CHANGELOG file type: String format: Path

2 .janno file variables

.janno file variables

Variable	Description
Poseidon_ID*	sample identifier as defined by the genetics laboratory (e.g. I1234, BOT001), must fit to the values in the Poseidon package .fam/.ind file, must be unique within one package, if multiple datasets exist for the same individual different Poseidon_IDs are required type: String
Genetic_Sex*	genetic sex of the individual derived from this sample, only F, M or U because the EIGENSTRAT and PLINK formats only support these three, edge cases (e.g. XXY, XYY, X0) are undefined and should be grouped as F, M or U, with a Note added type: Char allowed values: F; M; U

Variable	Description
Group_Name*	meaningful population/group identifiers for the sample, should follow the geographic-temporal nomenclature proposed by Eisenmann et al. 2018 (https://doi.org/10.1038/s41598-018-31123-z), multiple entries separated by ;, the first value must be equal the group name in the .fam/.ind file list column type: String
Alternative_IDs	alternative identifiers for the same sampled individual, e.g. IDs in other databases or popular names like Ötzi/Iceman list column type: String
Relation_To	other samples (by Poseidon_ID) that are related/identical to this sample, multiple entries separated by ; <u>list column</u> <u>type</u> : String
Relation_Degree	relationship degree for relatives mentioned in Related_To, multiple values separated by; in the same order as Related_To in case of multiple relations list column type: String allowed values: identical; first; second; thirdToFifth; sixthToTenth; unrelated; other
Relation_Type	relationship type for relatives mentioned in Related_To (e.g. sister_of, child_of, nephew_of), multiple values separated by ; in the same order as Related_To in case of multiple relations list column type: String
Relation_Note	arbitrary comments about the genetic relationships of the sampled individual type: String
Collection_ID	alternative sample identifier shared by the provider/owner of the sample (e.g. grave 40 skeleton 2) type: String
Country	present-day political country of origin for the sample $\underline{\text{type}}$: String
Country_ISO	present-day political country expressed in ISO 3166-1 alpha-2 country codes type : String
Location	unspecified location information for the sample, e.g. administrative or topographic region or mountains/rivers/lakes/cities nearby type: String

Variable	Description
Site	name of the archaeological site where the sample was found type: String
Latitude	latitude where the sample was found with up to 5 places after the decimal point type: Float allowed range: -90 to 90
Longitude	longitude with up to 5 places after the decimal point type: Float allowed range: -180 to 180
Date_Type	type of dating information available for the sample, C14 if there is a set of radiocarbon dates in the columns Date_C14_Labnr, Date_C14_Uncal_BP and Date_C14_Uncal_BP_Err whose post-calibration probability distribution is a meaningful prior for the individual's year of death, contextual for any other age information only given in Date_BC_AD_Start, Date_BC_AD_Median and Date_BC_AD_Stop, "modern" for present-day individuals type: String allowed values: C14; contextual; modern
Date_C14_Labnr	lab numbers of C14 ages, multiple values separated by ; in case of multiple dates <u>list column</u> type: String
Date_C14_Uncal_BP	uncalibrated years BP (as in before 1950AD) for the C14 ages as reported by C14 labs, multiple values separated by; in the same order as Date_C14_Labnr in case of multiple dates, only relevant if Date_Type is C14 list column type: Integer allowed range: 0 to Inf
Date_C14_Uncal_BP_Err	standard deviation (1-sigma ±) for the uncalibrated C14 ages as reported by the C14 labs, multiple values separated by; in the same order as Date_C14_Labnr in case of multiple dates, only relevant if Date_Type is C14 list column type: Integer allowed range: 0 to Inf
Date_BC_AD_Start	lower (older) bound for the age of the sample in years BC/AD, negative numbers for BC, positive numbers for AD, in case of C14 dates 2-sigma post calibration interval, 2000 for modern samples type: Integer allowed range: -Inf to 2050

Variable	Description
Date_BC_AD_Median	median age of the sample in years BC/AD, for C14-dated samples median, for contextually dated samples simple mid-point of the archaeological intervals, 2000 for modern samples type: Integer allowed range: -Inf to 2050
Date_BC_AD_Stop	upper (more recent) bound for the age of the sample in years BC/AD, counter point to Date_BC_AD_Start <u>type</u> : Integer <u>allowed range</u> : -Inf to 2050
Date_Note	arbitrary comments about the dating information for the sample type: String
MT_Haplogroup	mitochondrial haplogroup derived for the sample as specified on phylotree.org and as reported by the Haplofind or Haplogrep software tools type: String
Y_Haplogroup	Y-chromosome haplogroup derived for the sample following a syntax with the main branch $+$ the most terminal derived Y-SNP (e.g. R1b-P312) $\underline{\text{type}}$: String
Source_Tissue	skeletal element, tissue or other material sampled, the specific bone should be reported after an underscore (e.g. bone_phalanx), multiple values separated by; list column type: String
Nr_Libraries	number of libraries produced for the sample type: Integer
Library_Names	identifiers of the libraries used to generate the genotype data for the sample, multiple values separated by ; <u>list column</u> <u>type</u> : String
Capture_Type	specifics of the data generation method (e.g. capture method) for the individual libraries generated for the sample, multiple values separated by; list column type: String allowed values: Shotgun; 1240K; ArborComplete; ArborPrimePlus; ArborAncestralPlus; TwistAncientDNA; OtherCapture; ReferenceGenome
UDG	udg treatment for the libraries, mixed in case multiple libraries with different UDG treatment were merged type: String allowed values: minus; half; plus; mixed

Variable	Description
Library_Built	strandedness of the libraries, "mixed" in case multiple libraries with different protocols were merged type : String allowed values : ds; ss; mixed
Genotype_Ploidy	ploidy of the genotypes for the sample type: String allowed values: diploid; haploid
${\bf Data_Preparation_Pipeline_URL}$	url pointing to a description of the computational pipeline used to generate the genotype data from the source data type: String
Endogenous	% endogenous DNA as estimated from SG libraries (before capture) as for example estimated by EAGER, not on target and no quality filter, in case of multiple libraries only the highest values should be reported type: Float allowed range: 0 to 100
Nr_SNPs	number of non-missing SNPs for the sample, counted on the SNP-set stored in the Poseidon package type : Integer
Coverage_on_Target_SNPs	average X-fold coverage across targeted SNP sites after quality filtering $\underline{\text{type}}$: Float
Damage	% damage on the 5' end for the main shotgun library used for sequencing and/or capture, in case of multiple libraries a value from the merged read alignment should be reported type : Float allowed range : 0 to 100
Contamination	(modern) contamination of the sample as measured by the method in Contamination_Meas, multiple values separated by; (for different methods, in case of multiple libraries report a value from the merged read alignment), the variables Contamination, Contamination_Err and Contamination_Meas must have the same number and order of (non-n/a) entries list.column type: String
Contamination_Err	(modern) contamination estimate error of the sample list column type : String
Contamination_Meas	method to measure contamination, should be a software tool (ANGSD, Schmutzi,) and the respective software versions, details should go to Contamination_Note list column type: String

Variable	Description
Contamination_Note	arbitrary comments about the contamination estimation type: String
Genetic_Source_Accession_IDs	ENA or SRA accession identifiers pointing to the source data used to generate the genotyping data for the sample, multiple values separated by ;, if multiple are given they should be arranged by descending specificity (e.g. project id > sample id > sequencing run id) list column type: String
Primary_Contact	project lead or first author who generated and published the data for the sample type: String
Publication	bibtex keys for the publications where a sample was published (e.g. "AuthorJournalYear") or "unpublished", multiple values separated by ;, all must be present with complete BibTeX entries in the Poseidon package's .bib file list column type: String
Note	arbitrary comments about the sample type: String
Keywords	arbitrary tags, multiple values separated by ; list column type: String

3 .ssf file variables

.ssf file variables

Variable	Description
poseidon_IDs	Poseidon_IDs (in the .janno file) the sequencing entity corresponds to, multiple entries separated by ; list column type: String
udg	udg treatment applied to the library for the sequencing entity type: String allowed values: minus; half; plus
library_built	library preparation method applied for the sequencing entity (single- or double-stranded) type: String allowed values: ds; ss

.ssf file variables (continued)

Variable	Description
sample_accession	sample accession code as used in the INSDC databases (including ENA and SRA) to identify the sequencing entity (e.g. SAMEA7050454) type: String
study_accession	study accession code as used in the INSDC databases (e.g. PRJEB39316) $\underline{\rm type} \colon \mathrm{String}$
run_accession	run accession code as used in the INSDC databases (e.g. ERR4331996), this should be a unique identifier in a Poseidon package type: String
sample_alias	sample alias defined by the submitter in the raw sequencing data repository type: String
secondary_sample_accession	a secondary sample accession, used in the ENA database for historical reasons (e.g. ERS4811084) type: String
first_public	date (YYYY-MM-DD) the sequencing entity was first made public in the raw sequencing data repository type: Date
last_updated	date (YYYY-MM-DD) the sequencing entity was last updated in the raw sequencing data repository $\underline{\rm type} \colon$ Date
instrument_model	name of the instrument used to process the sequencing entity (e.g. Illumina HiSeq 2500) type: String
library_layout	library layout of the sequencing entity (e.g. SINGLE) type: String
library_source	source of the DNA library (e.g. GENOMIC) type: String
$instrument_platform$	platform, brand or type of the sequencer (e.g. ILLUMINA) type: String
library_name	library identifier, so library name the submitter has entered to the raw sequencing data repository, data entries across which optical duplicates could exist should have matching library names type : String
library_strategy	strategy used to create the library for the sequencing entity (e.g. WGS) type: String

.ssf file variables (continued)

Variable	Description
fastq_ftp	ftp links to the FASTQ files for the sequencing entity in the raw sequencing data repository (e.g. ftp.sra.ebi.ac.uk/vol1/fastq/ERR433/009/ERR4332639/ERR4332639.fastq.gz), multiple entries separated by ; list column type: URL
fastq_aspera	aspera links to the FASTQ files for the sequencing entity in the raw sequencing data repository (e.g. fasp.sra.ebi.ac.uk:/vol1/fastq/ERR433/009/ERR4332639/ERR4332639.fastq.gz), multiple entries separated by; list column type: URL
fastq_bytes	number of bytes in the FASTQ files, multiple entries separated by ;, must be in the same order as the ftp and/or aspera links list column type: Integer allowed range: 0 to Inf
${\rm fastq_md5}$	md5 hashes of the FASTQ files, multiple entries separated by ;, must be in the same order as the ftp and/or aspera links list column type: String
read_count	number of reads in the sequencing entity <u>type</u> : Integer <u>allowed range</u> : 0 to Inf
$submitted_ftp$	urls to the originally submitted files before they got converted to FASTQ in the INSDC databases, multiple entries separated by ; $\frac{\text{list column}}{\text{type: String}}$