Guide for qjanno v1.0.0.0

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13	1	Background	
14	qja	anno started as a fork of the qhs software tool, which was, in turn, inspired by the CLI tool q. All of the	em
15	en	able SQL queries on delimiter-separated text files (e.gcsv or .tsv). For qjanno, we copied the source code	e of
16	qh	as $v0.3.3$ (MIT-License) and adjusted it to provide a smooth experience with a special kind of .tsv file: T	The
17	Po	oseidon .janno file.	
18	Ur	nlike trident or xerxes qjanno does not have a complete understanding of the .janno-file structure, a	ind
19	(m	ostly) treats it like a normal .tsv file. It does not validate the files upon reading and takes them at face val	lue.
20	St	ill .janno files are given special consideration: With a set of pseudo-functions in the FROM field of the So	QL

2 How does this work?

with .janno files (e.g. for JOIN operations).

On startup, qjanno creates an SQLite [1] database in memory. It then reads the requested, structured text files and attributes each column a type (either character or numeric). With this annotation the contents of the files can be written to tables in the in-memory database. qjanno finally sends the user-provided SQL query to said database, waits for the result, parses it and returns it on the command line.

qjanno still supports most features of qhs, so it can still read .csv and .tsv files independently or in conjunction

query they can be searched recursively and loaded together into one table.

29 The query gets pre-parsed to extract file names and then forwarded to an SQLite database server via the Haskell

- library sqlite-simple. That means qianno can parse and understand basic SQLite3 syntax, though not everything.
- PRAGMA functions, for example, are not available. The examples below show some of the available syntax, but
- they are not exhaustive. Trial and error is recommended to see what does and what does not work. Please report
- missing functionality in our issue board on GitHub.

34 3 Installation

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

37 4 The CLI interface

This is the CLI interface of qjanno:

```
Usage: qjanno [--version] [QUERY] [-q|--queryFile FILE] [-c|--showColumns]
              [-t|--tabSep] [--sep DELIM] [--noHeader] [--raw] [--noOutHeader]
  Command line tool to allow SQL queries on .janno (and arbitrary .csv and .tsv)
  files.
Available options:
  -h,--help
                           Show this help text
  --version
                           Show qjanno version
  QUERY
                           SQLite syntax query with paths to files for table
                           names. See the online documentation for examples. The
                           special table name syntax 'd(path1,path2,...)' treats
                           the paths (path1, path2, ...) as base directories
                           where .janno files are searched recursively. All
                           detected .janno files are merged into one table and
                           can thus be subjected to arbitrary queries.
  -q,--queryFile FILE
                           Read query from the provided file.
  -c,--showColumns
                           Don't run the query, but show all available columns
                           in the input files.
  -t,--tabSep
                           Short for --sep $'\t'.
  --sep DELIM
                           Input file field delimiter. Will be automatically
                           detected if it's not specified.
  --noHeader
                           Does the input file have no column names? They will
                           be filled automatically with placeholders of the form
                           c1,c2,c3,...
                           Return the output table as tsv.
  --raw
  --noOutHeader
                           Remove the header line from the output.
```

- This help can be accessed with qjanno -h. Running qjanno without any parameters does not work: The QUERY
- 40 parameter is mandatory and the tool will fail with Query cannot be empty.

4.1 A basic example

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A basic, working qjanno query could look like this:

```
$ qjanno "SELECT package_title,Poseidon_ID,Country \
          FROM d(2010_RasmussenNature, 2012_MeyerScience)"
                        package_title
                              Poseidon_ID
                                                  Country
 2010_RasmussenNature | Inuk.SG
                                                | Greenland
| 2012 MeyerScience
                        | A Mbuti-5.DG
                                                | Congo
| 2012_MeyerScience
                        | A_Yoruba-4.DG
                                                | Nigeria
| 2012_MeyerScience
                        | A_Sardinian-4.DG
                                                | Italy
| 2012 MeyerScience
                        | A French-4.DG
                                                | France
| 2012_MeyerScience
                        | A_Dinka-4.DG
                                                | Sudan
| 2012_MeyerScience
                        | A_Ju_hoan_North-5.DG | Namibia
```

- Running qjanno with this query triggers the following process:
 - 1. With d(...) in the FROM field, qjanno searches recursively for package-defining POSEIDON.yml files in the given base directories 2010_RasmussenNature and 2012_MeyerScience.
 - 2. It finds the .yml files and reads some of their fields, including the title, the packageVersion and the jannoFile path. It then selects the latest version of each package.
 - 3. With the relevant .janno file paths available, qjanno reads them, appends the package_title, package_version and source_file columns, merges them (simple row-bind), and orders their columns.
 - 4. It then writes the resulting janno table to the SQLite database in memory.
 - 5. Now the actual query gets executed. In this case the SELECT statement includes three variables (column names): package_title, Poseidon_ID and Country. The database server returns these three columns for the merged .janno table.
 - 6. gianno finally prints the result in a neat, human readable format to the standard output.

55 4.2 The .janno-crawling pseudo-functions

- 56 d(...) is one of four mechanisms to search and load .janno files in the FROM field of the query:
 - d(<path_to_directory1>,<path_to_directory2>,...): With d(), qjanno (recursively) searches all package-defining POSEIDON.yml files in all listed directories and reads them to determine the latest package version. It then reads the .janno files associated with these latest package versions.
 - da(<path_to_directory1>,<path_to_directory2>,...): da() behaves just as d(), but it does not filter for the latest package version: It loads all packaged .janno files.
 - j(<path_to_directory1>,<path_to_directory2>,...): j() simply searches for files with the extension .janno in all listed directories and loads them regardless of whether they are part of a Poseidon package or not.
 - <path_to_one_janno_file>.janno: Specific .janno files can be listed individually. They are identified as such by their .janno extension.
- Multiple of these methods can be combined as a comma-separated list. Each respective mechanism then yields a list of janno file paths, and the list of lists is flattened to a simple list of paths. qjanno then reads all files in

- 69 this combined list, merges them and makes them available for querying in the in-memory SQLite database.
- 70 !> Note that FROM field should not include any spaces even in a comma-separated list, gjanno parses the QUERY
- 11 using space as a separator.

72 4.3 CLI details

qjanno can not just read .janno files, but also arbitrary .csv and .tsv files. This option is triggered by providing file names (relative paths) in the FROM field of the query, not d(...).

With these non-.janno files qjanno automatically tries to detect the relevant separator. With --sep a delimiter can be specified explicitly, and the shortcut -t sets --sep \$'\t' for tab-separated files.

The --noHeader option allows to read files without headers, so column names. The columns are then automatically named $c1, c2, \ldots cN$:

```
$ echo -e "Val1,Val2\nVal3,Val4\n" > test.csv
$ qjanno "SELECT c1,c2 FROM test.csv" --noHeader
.-----.
| c1 | c2 |
:====:=::
| Val1 | Val2 |
| Val3 | Val4 |
|-----'
```

The remaining options concern the output: --raw returns the output table not in the neat, human-readable

80 ASCII table layout, but in a simple .tsv format. --noOutHeader omits the header line in the output.

```
$ echo -e "Col1,Col2\nVal1,Val2\nVal3,Val4\n" > test.csv
$ qjanno "SELECT * FROM test.csv" --raw --noOutHeader
test.csv Val1 Val2
test.csv Val3 Val4
```

Note that these output options allow to directly prepare individual lists in trident's forgeScript selection language

82 format:

```
$ qjanno "SELECT '<'||Poseidon_ID||'>' FROM d(2012_MeyerScience)" --raw --noOutHeader
<A_Mbuti-5.DG>
<A_Yoruba-4.DG>
<A_Sardinian-4.DG>
<A_French-4.DG>
<A_Dinka-4.DG>
<A_Ju_hoan_North-5.DG>
```

83 4.4 The -c/--showColumns option

-c/--showColumns is a special option that, when activated, makes qjanno return not the result of a given query, but an overview table with the columns available in all loaded tables/files for said query. That is helpful to get

```
an overview what could actually be queried.
```

This summary also includes the artificial, structurally cleaned table names assigned by qjanno before writing to the SQLite database. Often we can not simply use the file names as table names, because SQLite has strict naming requirements. File names or relative paths are generally invalid as table names and need to be replaced with a tidy string. These artificially generated names are mostly irrelevant from a user perspective – except a query involves multiple files, e.g. in a JOIN operation. See below for an example.

⁹² 5 Query examples

- 93 The following examples show some of the functionality of the SQLite query language available through qjanno.
- See the SQLite syntax documentation for more details. They were prepared and tested in a clone of the Poseidon
- 95 community archive.

96 Sub-setting with WHERE

97 Get all individuals (rows) in two Poseidon packages where UDG is set to 'minus'.

```
$ qjanno " \
SELECT package_title,Poseidon_ID,UDG \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
WHERE UDG = 'minus' \
"
```

```
| Poseidon_ID | UDG |
:=====::
| Inuk.SG | minus |
'-----'
```

Get all individuals where Genetic_Sex is not 'F' and Country is 'Sudan'.

99 Get all individuals where the the UDG column is not NULL or the Country is 'Sudan'.

 $_{100}$ Get all individuals where Nr_SNPs is equal to or bigger than 600,000.

Ordering with ORDER BY

102 Order all individuals by Nr_SNPs.

```
$ qjanno " \
SELECT Poseidon_ID,Nr_SNPs \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
ORDER BY Nr_SNPs \
.----.
    Poseidon_ID
                 | Nr_SNPs |
| A_French-4.DG
                  | 592535 |
| A_Ju_hoan_North-5.DG | 593045 |
A_Mbuti-5.DG
                 | 593057 |
| A_Dinka-4.DG
                 | 593076 |
| A_Yoruba-4.DG
                 | 593097 |
| A_Sardinian-4.DG | 593109 |
| Inuk.SG
                 | 1101700 |
```

Order all individuals by Date_BC_AD_Median in a descending (DESC) order. Date_BC_AD_Median includes
NULL values.

$_{105}$ Reducing the number of return values with LIMIT

Only return the first three result individuals.

107 Combining tables with JOIN

For JOIN operations, SQLite requires table names to specify which columns are meant when combining multiple tables with overlapping column names. See the option -c/--showColumns to get the relevant table names as generated from the input paths.

```
$ echo -e "Poseidon_ID,MoreInfo\nInuk.SG,5\nA_French-4.DG,3\n" > test.csv
$ qjanno "SELECT * FROM d(2010_RasmussenNature,2012_MeyerScience)" -c
.-----.
         Column
                                     Path
| package_title
                      | d(2010_RasmussenNature,2012_MeyerScience) |
| package_version
                     | d(2010_RasmussenNature,2012_MeyerScience) |
| source_file
                      | d(2010_RasmussenNature,2012_MeyerScience) |
                      | d(2010_RasmussenNature,2012_MeyerScience) |
| Poseidon_ID
. . .
       qjanno Table name
d2010RasmussenNature2012MeyerScience |
d2010RasmussenNature2012MeyerScience |
d2010RasmussenNature2012MeyerScience |
d2010RasmussenNature2012MeyerScience |
$ qjanno "SELECT * FROM test.csv" -c
,-----,
  Column | Path | qjanno Table name |
| source_file | test.csv | test
| Poseidon_ID | test.csv | test
```

Join the .janno files with the information in the test.csv file (by the Poseidon_ID column).

```
$ qjanno " \
SELECT d2010RasmussenNature2012MeyerScience.Poseidon_ID,Country,MoreInfo \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
INNER JOIN test.csv \
ON d2010RasmussenNature2012MeyerScience.Poseidon_ID = test.Poseidon_ID \
"
```

112 Grouping data and applying aggregate functions

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SQLite provides a number of aggregation functions: avg(X), count(*), count(X), group_concat(X), group_concat(X), max(X), min(X), sum(X). See the documentation here. These functions can be well combined with the GROUP BY operation.

Determine the minimal number of SNPs across all individuals.

```
$ qjanno "SELECT min(Nr_SNPs) AS n FROM d(2010_RasmussenNature,2012_MeyerScience)"
.-----.
| n |
:======:
| 592535 |
'-----'
```

117 Count the number of individuals per Date_Type group and calculate the average Nr_SNPs for both groups.

119 [1] K. P. Gaffney, M. Prammer, L. Brasfield, D. R. Hipp, D. Kennedy, and J. M. Patel, "SQLite: Past, present, and future," *Proceedings of the VLDB Endowment*, vol. 15, no. 12, pp. 3535–3547, Aug. 2022, doi: 10.14778/3554821.3554842.