Guide for qjanno v1.0.0

Contents

1 Background

qjanno is a fork of the qhs software tool, which is, in turn, inspired by the CLI tool q. All of them enable SQL queries on delimiter-separated text files (e.g. .csv or .tsv). For qjanno we copied the source code of qhs v0.3.3 (MIT-License) and adjusted it to provide a smooth experience with a special kind of .tsv file: The Poseidon .janno file.

Unlike `trident` or `xerxes` qjanno does not have a complete understanding of the .janno-file structure, and (mostly) treats it like a normal .tsv file. It does not validate the files upon reading and takes them at face value. Still .janno files are given special consideration: With the `d(...)` pseudo-function they can be searched recursively and loaded together into one table.

qjanno still supports most features of qhs, so it can still read .csv and .tsv files independently or in conjunction with .janno files (e.g. for `JOIN` operations).

2 How does this work?

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

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 qjanno 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.

3 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,...
  --raw                    Return the output table as tsv.
  --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` parameter is mandatory and the tool will fail with `Query cannot be empty`.

A basic, working query could look like this:

```
$ qjanno "SELECT Poseidon_ID,Country FROM d(2010_RasmussenNature,2012_MeyerScience)"
.----------------------.-----------.
|     Poseidon_ID      |  Country  |
:======================:===========:
| Inuk.SG              | Greenland |
| A_Mbuti-5.DG         | Congo     |
| A_Yoruba-4.DG        | Nigeria   |
| A_Sardinian-4.DG     | Italy     |
| A_French-4.DG        | France    |
| A_Dinka-4.DG         | Sudan     |
| A_Ju_hoan_North-5.DG | Namibia   |
'----------------------'-----------'
```

qjanno is asked to run the query `SELECT ... FROM ...`, which triggers the following process:

1. As `d(...)` is provided in the table name field (`FROM`), qjanno searches recursively for .janno files in the provided base directories `2010_RasmussenNature` and `2012_MeyerScience`.
2. It finds the .janno files, reads them and merges them (simple row-bind).
3. It writes the resulting table to the SQLite database in memory.
4. Now the actual query gets executed. In this case the `SELECT` statement includes two variables (column names): `Poseidon_ID` and `Country`. The database server returns these two columns for the merged .janno table.
5. qjanno returns the resulting table in a neat, human readable format.

3.1 CLI details

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

```
$ echo -e "Col1,Col2\nVal1,Val2\nVal3,Val4\n" > test.csv
$ qjanno "SELECT Col2 FROM test.csv"
.------.
| Col2 |
:======:
| Val2 |
| Val4 |
'------'
```

qjanno automatically tries to detect the relevant separator of files. With `--sep` a delimiter can be specified explicitly, and the shortcut `-t` sets `--sep $'\t'` for tab-separated files. So a .janno file can also be read without `d(...)` using the following syntax:

```
$ qjanno "SELECT Poseidon_ID,Country FROM 2010_RasmussenNature/2010_RasmussenNature.janno" \
  -t # -t is optional
.-------------.-----------.
| Poseidon_ID |  Country  |
:=============:===========:
| Inuk.SG     | Greenland |
'-------------'-----------'
```

The `--noHeader` option allows to read files without headers, so column names. The columns are then automatically named *c1,c2,... 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 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
Val1  Val2
Val3  Val4
```

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

3.2 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.

```
$ echo -e "Col1,Col2\nVal1,Val2\nVal3,Val4\n" > test.csv
$ qjanno "SELECT * FROM test.csv" -c
.--------.----------.-------------------.
| Column |   Path   | qjanno Table name |
:========:==========:===================:
| Col1   | test.csv | test              |
| Col2   | test.csv | test              |
'--------'----------'-------------------'
```

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.

4 Query examples

The following examples show some of the functionality of the SQLite query language available through qjanno. See the SQLite syntax documentation for more details.

Sub-setting with `WHERE`

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

```
$ qjanno " \
SELECT 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'.

```
$ qjanno " \
SELECT Poseidon_ID,Country \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
WHERE Genetic_Sex <> 'F' AND Country = 'Sudan' \
"
.--------------.---------.
| Poseidon_ID  | Country |
:==============:=========:
| A_Dinka-4.DG | Sudan   |
'--------------'---------'
```

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

```
$ qjanno " \
SELECT Poseidon_ID,Country \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
WHERE UDG IS NOT NULL OR Country = 'Sudan' \
"
.--------------.-----------.
| Poseidon_ID  |  Country  |
:==============:===========:
| Inuk.SG      | Greenland |
| A_Dinka-4.DG | Sudan     |
'--------------'-----------'
```

Get all individuals where Nr_SNPs is equal to or bigger than 600,000.

```
$ qjanno " \
SELECT Poseidon_ID,Nr_SNPs \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
WHERE Nr_SNPs >= 600000 \
"
.-------------.---------.
| Poseidon_ID | Nr_SNPs |
:=============:=========:
| Inuk.SG     | 1101700 |
'-------------'---------'
```

Ordering with `ORDER BY`

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.

```
$ qjanno " \
SELECT Poseidon_ID,Date_BC_AD_Median \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
ORDER BY Date_BC_AD_Median DESC \
"
.----------------------.-------------------.
|     Poseidon_ID      | Date_BC_AD_Median |
:======================:===================:
| Inuk.SG              | -1935             |
| A_Sardinian-4.DG     |                   |
| A_Yoruba-4.DG        |                   |
| A_Dinka-4.DG         |                   |
| A_Mbuti-5.DG         |                   |
| A_Ju_hoan_North-5.DG |                   |
| A_French-4.DG        |                   |
'----------------------'-------------------'
```

Reducing the number of return values with `LIMIT`

Only return the first three result individuals.

```
$ qjanno " \
SELECT Poseidon_ID,Group_Name \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
LIMIT 3 \
"
.---------------.-----------------------------.
|  Poseidon_ID  |         Group_Name          |
:===============:=============================:
| Inuk.SG       | Greenland_Saqqaq.SG         |
| A_Mbuti-5.DG  | Ignore_Mbuti(discovery).DG  |
| A_Yoruba-4.DG | Ignore_Yoruba(discovery).DG |
'---------------'-----------------------------'
```

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                   |
:==============================:===========================================:
| Capture_Type                 | d(2010_RasmussenNature,2012_MeyerScience) | ->
...
--------------------------------------.
          qjanno Table name           |
======================================:
 d2010RasmussenNature2012MeyerScience |
...

$ qjanno "SELECT * FROM test.csv" -c
.-------------.----------.-------------------.
|   Column    |   Path   | qjanno Table name |
:=============:==========:===================:
| 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 \
"
.---------------.-----------.----------.
|  Poseidon_ID  |  Country  | MoreInfo |
:===============:===========:==========:
| Inuk.SG       | Greenland | 5        |
| A_French-4.DG | France    | 3        |
'---------------'-----------'----------'
```

Grouping data and applying aggregate functions

SQLite provides a number of aggregation functions: `avg(X)`, `count(*)`, `count(X)`, `group_concat(X)`, `group_concat(X,Y)`, `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 |
'--------'
```

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

```
$ qjanno " \
SELECT Date_Type,count(*),avg(Nr_SNPs) \
FROM d(2010_RasmussenNature,2012_MeyerScience) \
GROUP BY Date_Type \
"
.-----------.----------.--------------.
| Date_Type | count(*) | avg(Nr_SNPs) |
:===========:==========:==============:
| C14       | 1        | 1101700.0    |
| modern    | 6        | 592986.5     |
'-----------'----------'--------------'
```