

relsearch_0.1.0_manual

Sho Manabe (manabesh@hirakata.kmu.ac.jp)

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Information on relsearch

relsearch is open-source software for screening relatives between query and reference databases. The software is graphical-user-interface written in R language ($\geq 4.2.0$) with GNU General Public License v3.0. It can be applied to autosomal short tandem repeat (STR) markers, Y-STR markers, and mitochondrial DNA sequences commonly used in forensic genetics. For autosomal STR markers, likelihood ratios (LRs) of each query-reference pair are calculated considering mutation and drop-out. For Y-STR markers and mitochondrial DNA sequences, the software investigates the number of inconsistencies between query and reference haplotypes.

Getting started

1. Ensure that R ($\geq 4.2.0$) is installed. It is available from the R Development Core Team website (<http://www.R-project.org>).
2. Begin an R session.
3. Execute the following command in R to install required packages.

```
install.packages(c("Rcpp", "readr", "tcltk2"))
```

4. Go to <https://github.com/manabe0322/research/releases>.
5. Download "research_0.1.0.zip".
6. Execute the following commands in R to start GUI.

```
library(research)  
research()
```

Autosomal STR

Load query database

1. Press the 'Load' button for query database.

relsearch ver. 0.1.0

File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Input files

Query database **Load**

Reference database **Load**

Allele frequencies **Load**

Setting

Condition **Mutation rate** **IBD probabilities**

Screening

2. Select a .csv file for query database. Format of query database is shown in Fig. 1.

Sample Name	D3S1358	D3S1358	vWA	vWA	D16S539	D16S539	CSF1PO	CSF1PO	TPOX	TPOX
Q11	17	17	17		11		12			
Q12	17		18	18			10	11	8	
Q13	15	17	17	18	10	10	10	12	8	11
Q14	15	18	15	18	9	12	11	12	8	8
Q15	15	15	18		9	9	10		9	11
Q16	16	16	17				10	13	8	8
Q17	14	15	17	18	12				8	8
Q18	16	18	17	18	11		9	12	8	
Q19	15	15	16	18	11	11	12	12	8	11
Q20	15	16	18	19	9	10	12	12	8	11

Figure 1: Format of query database for autosomal STR

Note : format of query database

- This file must include information regarding ‘Sample Name’ and each marker.
- There are two columns in each marker.
- The marker with two empty cells (e.g., D16S539 of sample ‘Q12’ in Fig. 1) is ignored when calculating the LR.
- The marker with one empty cell (e.g., D3S1358 of sample ‘Q12’ in Fig. 1) can be regarded as allelic drop-out depending on the setting ‘Drop-out of query genotypes’ (see section 3.4).
- An example file named “str_query_example.csv” is located at `extdata > examples`.

Load reference database

1. Press the 'Load' button for reference database.

relsearch ver. 0.1.0

File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Input files

Query database Load

Reference database **Load**

Allele frequencies Load

Setting

Condition Mutation rate IBD probabilities

Screening

2. Select a .csv file for reference database. Format of reference database is shown in Fig. 2.

Sample Name	Relationship	D3S1358	D3S1358	vWA	vWA	D16S539	D16S539	CSF1PO	CSF1PO	TPOX	TPOX
R11	parent-child	17	17	14	17	10	10	12	12	8	8
R12	parent-child	17	18	17	18	12	13	10	11	11	11
R13	sibling	15	16	17	18	9	10	11	12	8	11
R14		15	15	15	19	11	12	11	12	8	8
R15	sibling	15	16	15	18	9	9	10	12	9	11
R16	sibling	16	17	17	17	10	12	13	13	8	11
R17	sibling	16	17	14	18	9	9	12	15	8	8
R18	sibling	15	16	17	18	9	12	11	12	8	9
R19	2nd-degree	15	15	14	16	10	11	11	12	8	9
R20		15	16	17	18	9	12	12	12	8	12

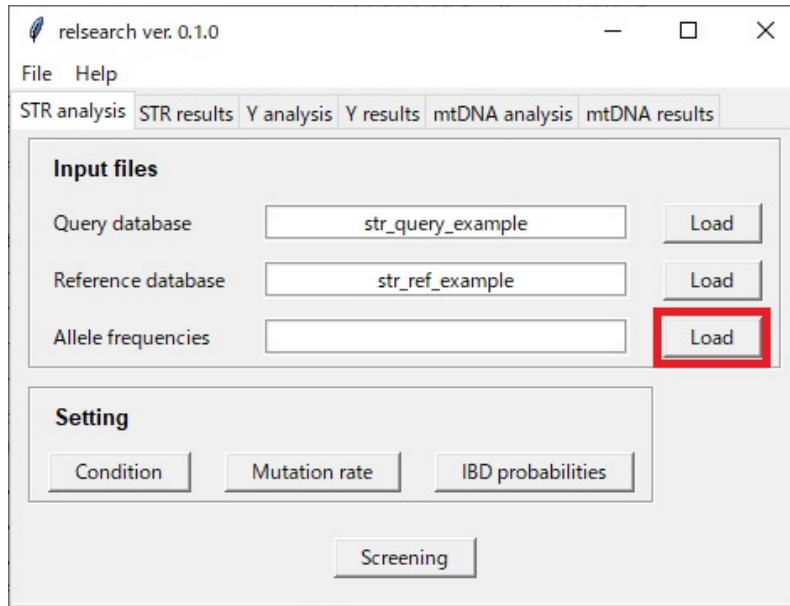
Figure 2: Format of reference database for autosomal STR

Note : format of reference database

- This file must include information regarding ‘Sample Name’, ‘Relationship’, and each marker.
- There are two columns in each marker.
- Names of the column ‘Relationship’ relate to those of relationships in the IBD probabilities (see section 3.6).
- If the relationship is not identified, leave the cell blank (e.g., samples ‘R14’ and ‘R20’). When calculating LR_s, all relationships for which the IBD probabilities is defined are considered.
- The marker with two empty cells is ignored when calculating the LR.
- The marker with one empty cell or with two same alleles (e.g., D3S1358 of sample ‘R11’ in Fig. 2) is regarded as the homozygotes.
- An example file named “str_ref_example.csv” is located at `extdata > examples`.

Load allele frequencies

1. Press the 'Load' button for allele frequencies.



2. Select a .csv file for allele frequencies. Format of allele frequencies is shown in Fig. 3.

Allele	D3S1358	vWA	D16S539	CSF1PO	TPOX	D8S1179
10			0.200266	0.215471	0.033566	0.128775
10.1						
10.2						
10.3						
11			0.186981	0.206175	0.356597	0.106206
11.1						
11.2						
11.3						
12	0.002324		0.172036	0.420983	0.038219	0.123133
12.2						
13	0.001328	0.000664	0.069744	0.069389	0.001329	0.225357
13.2						
14	0.02656	0.194887	0.008967	0.017596	0.000997	0.207766
14.1						

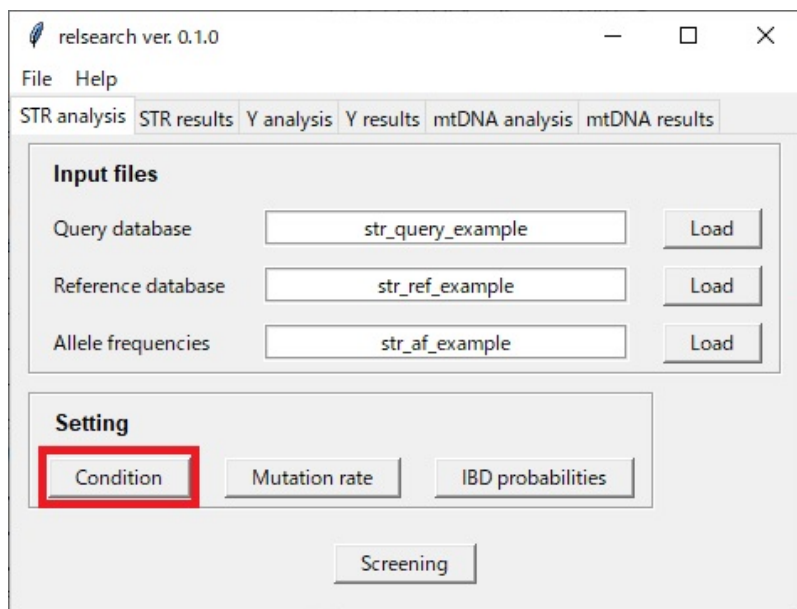
Figure 3: Format of allele frequencies for autosomal STR

Note : format of allele frequencies

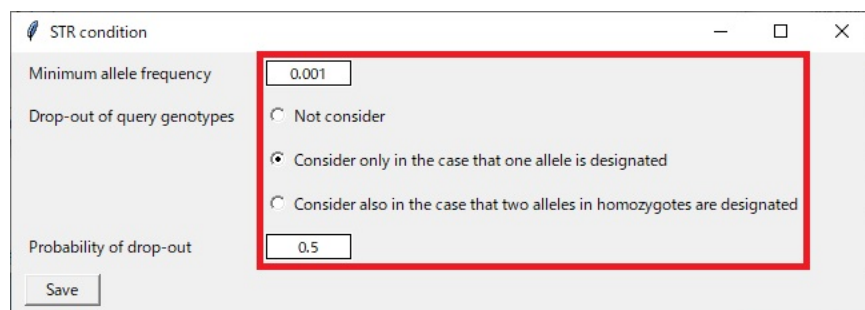
- This file must include information regarding 'Allele' and each marker.
- An example file named "str_af_example.csv" is located at extdata > examples.

Set conditions of calculation

1. Press the 'Condition' button.



2. Set conditions of 'Minimum allele frequency', 'Drop-out of query genotypes', and 'Probability of drop-out'.



Note : Drop-out of query genotypes

- **Not consider** : When calculating the LR, the marker with one empty cell in a query or reference database is regarded as the homozygote. The marker with two empty cells is ignored (i.e., $LR = 1$).
- **Consider only in the case that one allele is designated** : When calculating the LR, the marker with one empty cell in a query or reference database is regarded as both the homozygote (without drop-out) and the heterozygote (with drop-out). The marker with two empty cells is ignored (i.e., $LR = 1$).
- **Consider also in the case that two alleles in homozygotes are designated** : When calculating the LR, the marker with two same alleles in a query or reference database is also regarded as both the homozygote (without drop-out) and the heterozygote (with drop-out).

3. Press the 'Save' button.

STR condition

Minimum allele frequency

Drop-out of query genotypes

☐ Not consider

☒ Consider only in the case that one allele is designated

☐ Consider also in the case that two alleles in homozygotes are designated

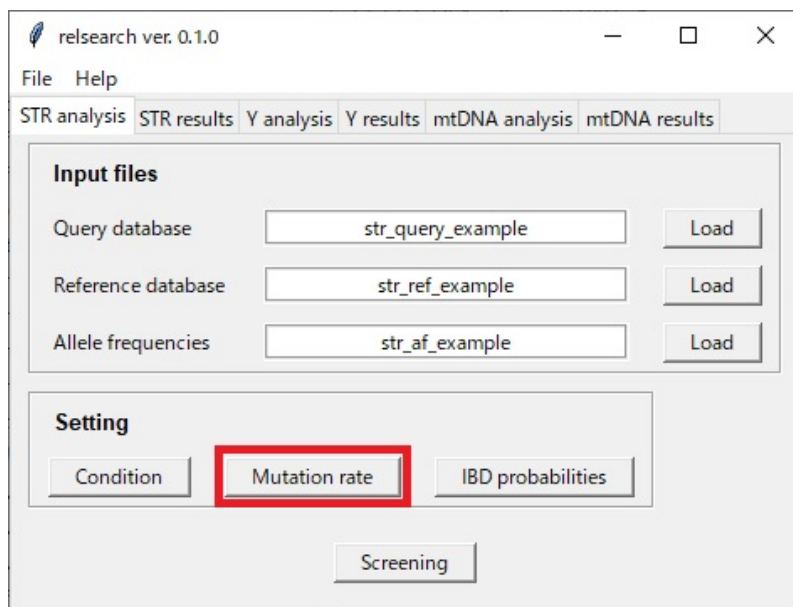
Probability of drop-out

Save

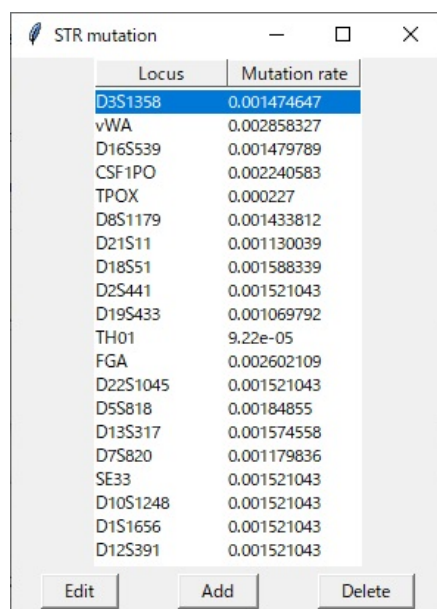
Set mutation rates

Edit mutation rates

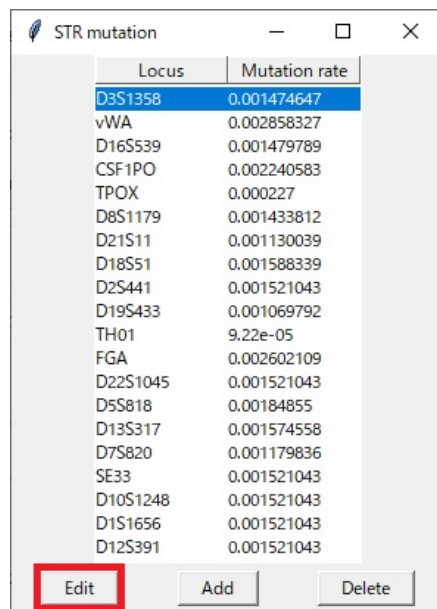
1. Press the 'Mutation rate' button. Then the window 'STR mutation' will be open.



2. Select a locus to change the mutation rate.



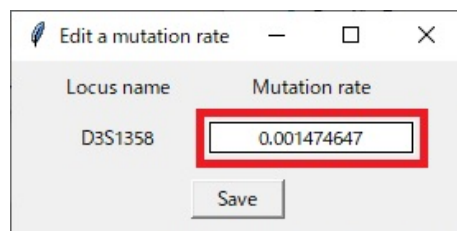
3. Press the 'Edit' button. Then the window 'Edit a mutation rate' will be open.



The 'STR mutation' window displays a table of loci and their corresponding mutation rates. The 'Edit' button at the bottom left is highlighted with a red box.

Locus	Mutation rate
D3S1358	0.001474647
vWA	0.002858327
D16S539	0.001479789
CSF1PO	0.002240583
TPOX	0.000227
D8S1179	0.001433812
D21S11	0.001130039
D18S51	0.001588339
D2S441	0.001521043
D19S433	0.001069792
TH01	9.22e-05
FGA	0.002602109
D22S1045	0.001521043
D5S818	0.00184855
D13S317	0.001574558
D7S820	0.001179836
SE33	0.001521043
D10S1248	0.001521043
D1S1656	0.001521043
D12S391	0.001521043

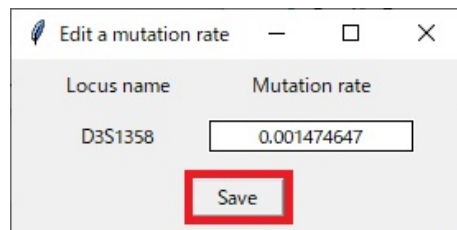
4. Enter an arbitrary mutation rate.



The 'Edit a mutation rate' window shows the 'Locus name' as 'D3S1358' and the 'Mutation rate' as '0.001474647'. The 'Mutation rate' input field is highlighted with a red box.

Locus name	Mutation rate
D3S1358	0.001474647

5. Press the 'Save' button.

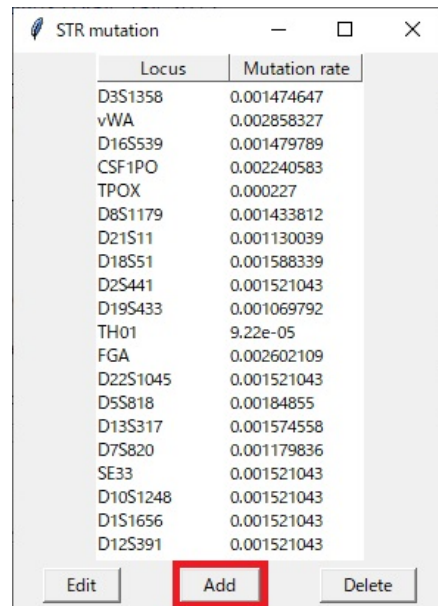


The 'Edit a mutation rate' window shows the 'Locus name' as 'D3S1358' and the 'Mutation rate' as '0.001474647'. The 'Save' button is highlighted with a red box.

Locus name	Mutation rate
D3S1358	0.001474647

Add a locus

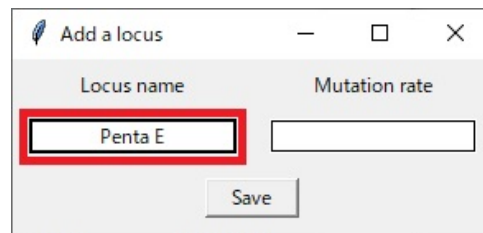
1. Press the 'Add' button in the window 'STR mutation'. Then the window 'Add a locus' will be open.



The 'STR mutation' window displays a table with two columns: 'Locus' and 'Mutation rate'. The table lists various loci and their corresponding mutation rates. At the bottom of the window, there are three buttons: 'Edit', 'Add', and 'Delete'. The 'Add' button is highlighted with a red box.

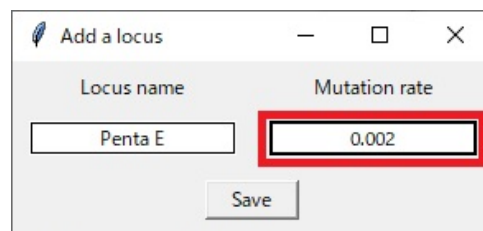
Locus	Mutation rate
D3S1358	0.001474647
vWA	0.002858327
D16S539	0.001479789
CSF1PO	0.002240583
TPOX	0.000227
D8S1179	0.001433812
D21S11	0.001130039
D18S51	0.001588339
D2S441	0.001521043
D19S433	0.001069792
TH01	9.22e-05
FGA	0.002602109
D22S1045	0.001521043
D5S818	0.00184855
D13S317	0.001574558
D7S820	0.001179836
SE33	0.001521043
D10S1248	0.001521043
D1S1656	0.001521043
D12S391	0.001521043

2. Enter an arbitrary locus name.



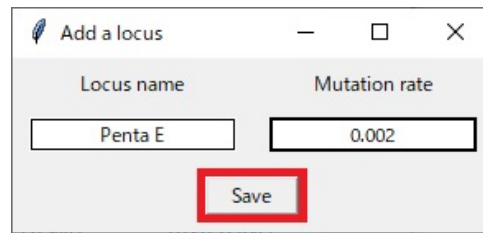
The 'Add a locus' window has two input fields: 'Locus name' and 'Mutation rate'. The 'Locus name' field is highlighted with a red box and contains the text 'Penta E'. Below the fields is a 'Save' button.

3. Enter an arbitrary mutation rate.



The 'Add a locus' window shows the 'Locus name' field with 'Penta E' and the 'Mutation rate' field highlighted with a red box, containing the value '0.002'. The 'Save' button is visible at the bottom.

4. Press the 'Save' button.



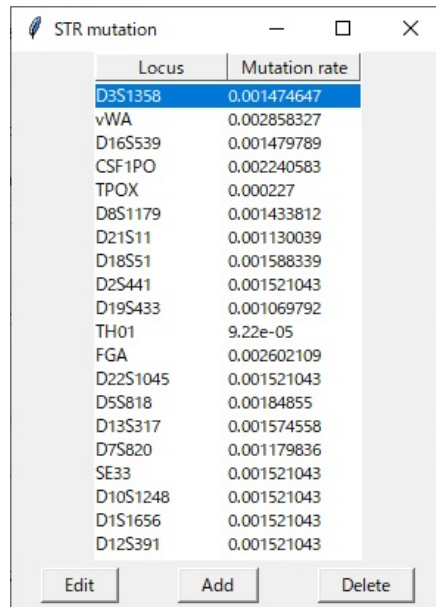
The image shows a software dialog box titled "Add a locus". It has a standard window header with a feather icon, a minus sign, a maximize button, and a close button. The dialog contains two input fields: "Locus name" with the text "Penta E" and "Mutation rate" with the value "0.002". Below these fields is a "Save" button, which is highlighted with a red rectangular border.

Locus name	Mutation rate
Penta E	0.002

Save

Delete a locus

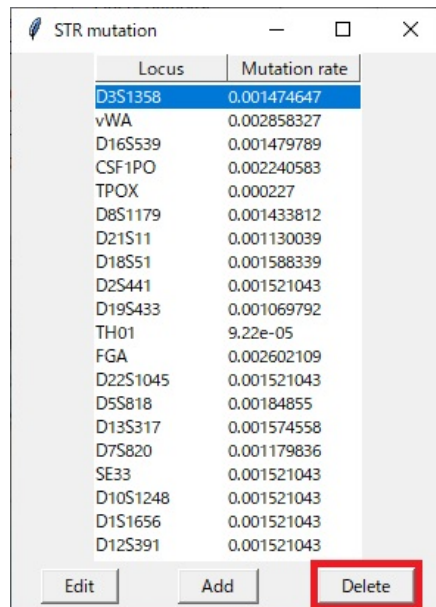
1. Select a locus in the window 'STR mutation'.



The screenshot shows a window titled 'STR mutation' with a table of loci and their mutation rates. The locus 'D3S1358' is selected, highlighted in blue. Below the table are three buttons: 'Edit', 'Add', and 'Delete'.

Locus	Mutation rate
D3S1358	0.001474647
vWA	0.002858327
D16S539	0.001479789
CSF1PO	0.002240583
TPOX	0.000227
D8S1179	0.001433812
D21S11	0.001130039
D18S51	0.001588339
D2S441	0.001521043
D19S433	0.001069792
TH01	9.22e-05
FGA	0.002602109
D22S1045	0.001521043
D5S818	0.00184855
D13S317	0.001574558
D7S820	0.001179836
SE33	0.001521043
D10S1248	0.001521043
D1S1656	0.001521043
D12S391	0.001521043

2. Press the 'Delete' button. Then the selected locus will be deleted.



This screenshot is identical to the previous one, but the 'Delete' button at the bottom right is highlighted with a red rectangle, indicating the next step in the process.

Locus	Mutation rate
D3S1358	0.001474647
vWA	0.002858327
D16S539	0.001479789
CSF1PO	0.002240583
TPOX	0.000227
D8S1179	0.001433812
D21S11	0.001130039
D18S51	0.001588339
D2S441	0.001521043
D19S433	0.001069792
TH01	9.22e-05
FGA	0.002602109
D22S1045	0.001521043
D5S818	0.00184855
D13S317	0.001574558
D7S820	0.001179836
SE33	0.001521043
D10S1248	0.001521043
D1S1656	0.001521043
D12S391	0.001521043

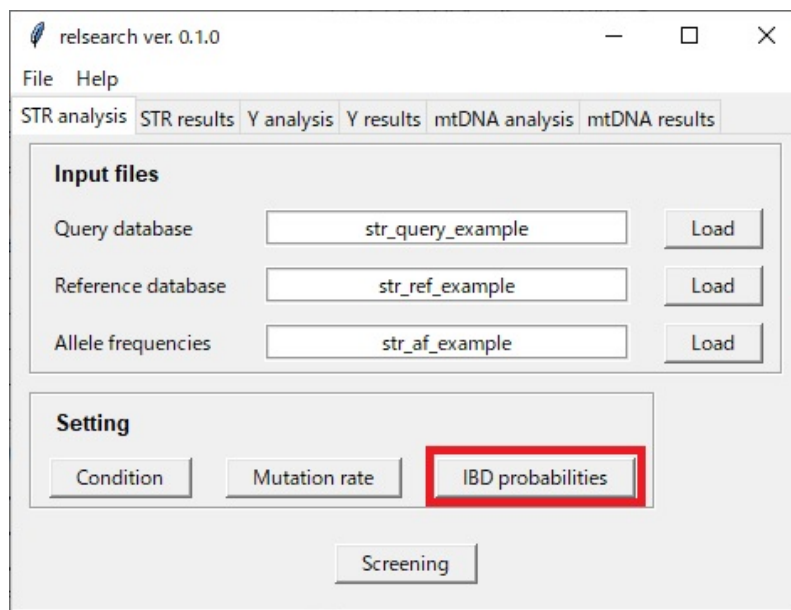
Set IBD probabilities

Note :

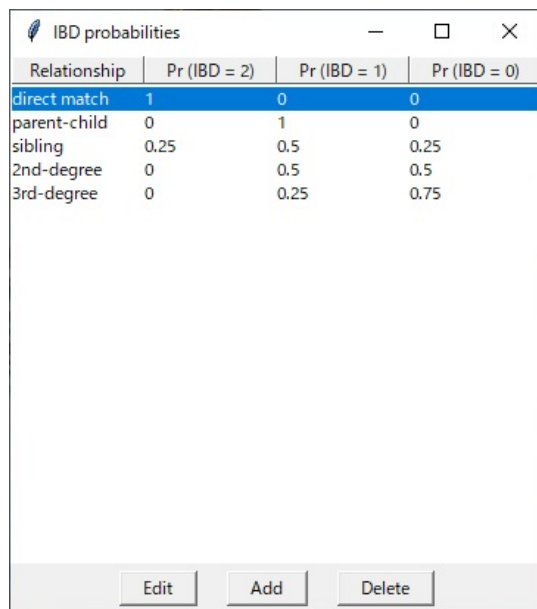
- When the relationship of each reference profile is not identified, all relationships for which the IBD probabilities is defined are considered when calculating the LR.

Edit IBD probabilities

1. Press the 'IBD probabilities' button. Then the window 'IBD probabilities' will be open.



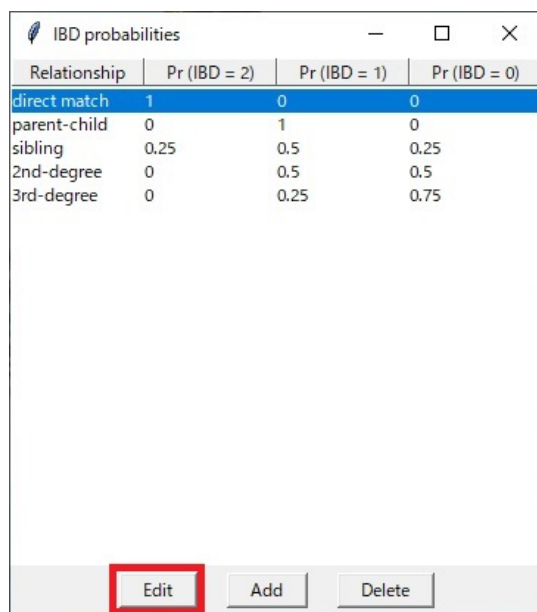
2. Select a relationship to change the IBD probabilities.



Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
direct match	1	0	0
parent-child	0	1	0
sibling	0.25	0.5	0.25
2nd-degree	0	0.5	0.5
3rd-degree	0	0.25	0.75

Edit Add Delete

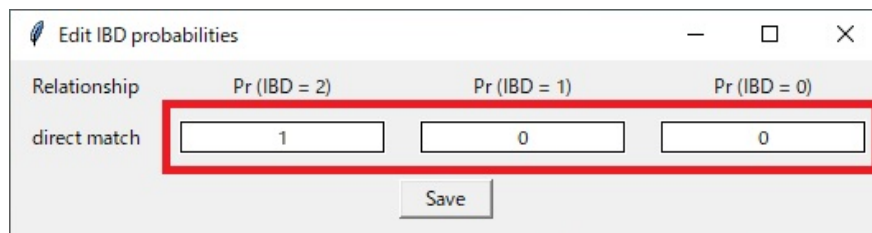
3. Press the 'Edit' button. Then the window 'Edit IBD probabilities' will be open.



Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
direct match	1	0	0
parent-child	0	1	0
sibling	0.25	0.5	0.25
2nd-degree	0	0.5	0.5
3rd-degree	0	0.25	0.75

Edit Add Delete

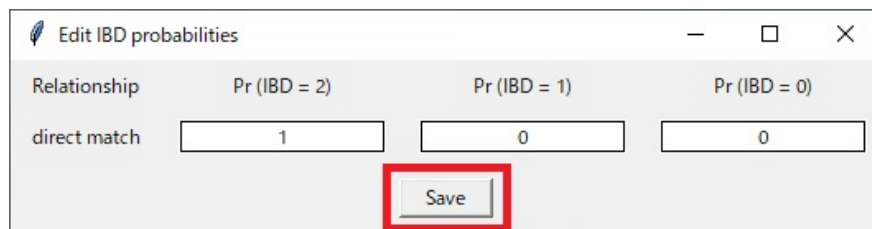
4. Enter arbitrary probabilities of $IBD = 2$, $IBD = 1$, and $IBD = 0$.



Relationship	Pr ($IBD = 2$)	Pr ($IBD = 1$)	Pr ($IBD = 0$)
direct match	1	0	0

Save

5. Press the 'Save' button.

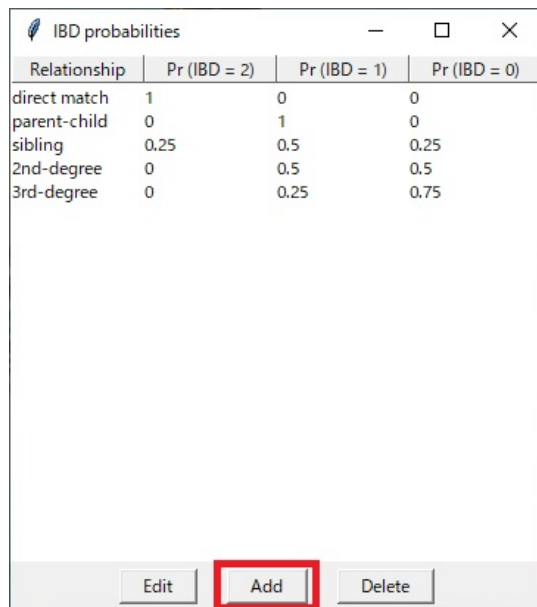


Relationship	Pr ($IBD = 2$)	Pr ($IBD = 1$)	Pr ($IBD = 0$)
direct match	1	0	0

Save

Add a relationship

1. Press the 'Add' button in the window 'IBD probabilities'. Then the window 'Add a relationship' will be open.



Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
direct match	1	0	0
parent-child	0	1	0
sibling	0.25	0.5	0.25
2nd-degree	0	0.5	0.5
3rd-degree	0	0.25	0.75

Buttons: Edit, Add, Delete

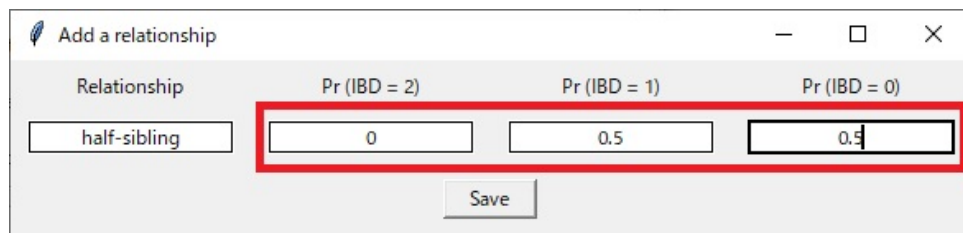
2. Enter an arbitrary relationship.



Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
half-sibling			

Save

3. Enter arbitrary probabilities of $IBD = 2$, $IBD = 1$, and $IBD = 0$.



Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
half-sibling	0	0.5	0.5

Save

4. Press the 'Save' button.

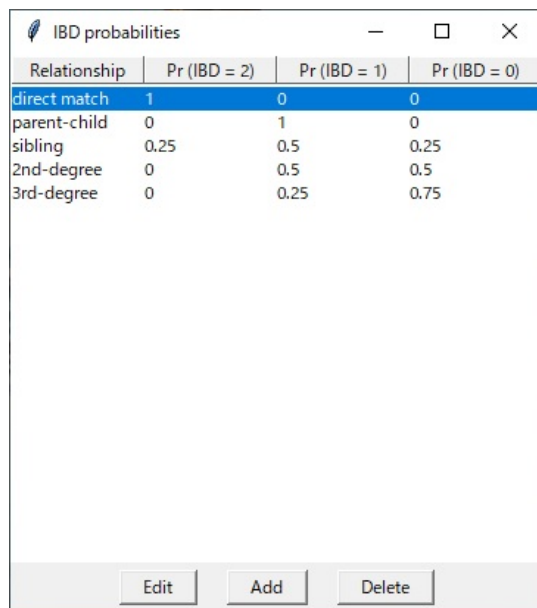


Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
half-sibling	0	0.5	0.5

Save

Delete a relationship

1. Select a relationship in the window 'IBD probabilities'.

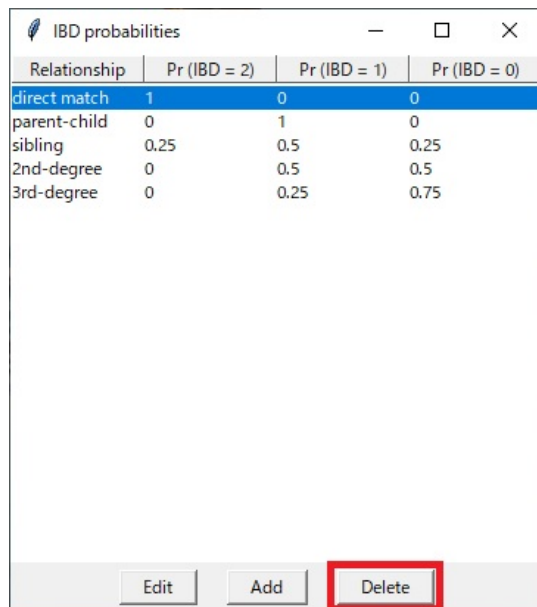


The screenshot shows a window titled 'IBD probabilities' with a table of relationships and their probabilities. The 'direct match' row is highlighted in blue.

Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
direct match	1	0	0
parent-child	0	1	0
sibling	0.25	0.5	0.25
2nd-degree	0	0.5	0.5
3rd-degree	0	0.25	0.75

Buttons at the bottom: Edit, Add, Delete.

2. Press the 'Delete' button. Then the selected relationship will be deleted.



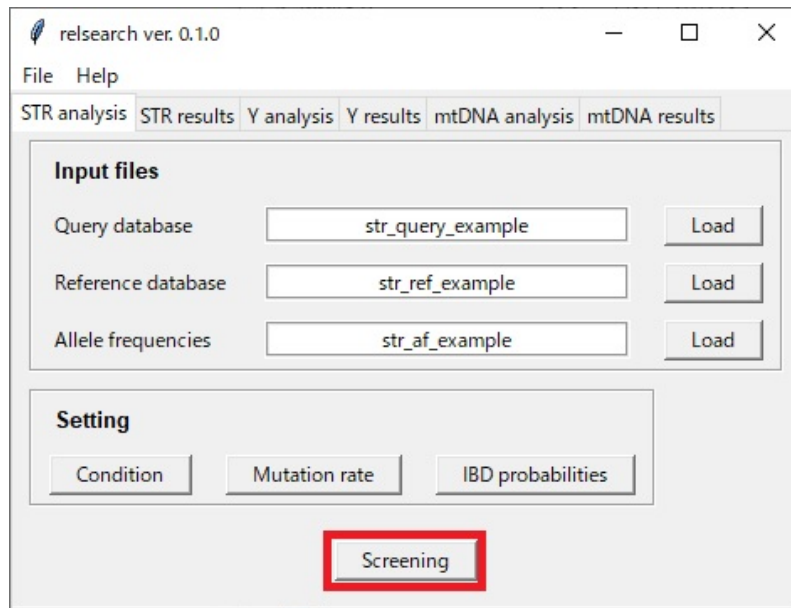
The screenshot shows the same 'IBD probabilities' window as before, but the 'Delete' button at the bottom is highlighted with a red rectangular box.

Relationship	Pr (IBD = 2)	Pr (IBD = 1)	Pr (IBD = 0)
direct match	1	0	0
parent-child	0	1	0
sibling	0.25	0.5	0.25
2nd-degree	0	0.5	0.5
3rd-degree	0	0.25	0.75

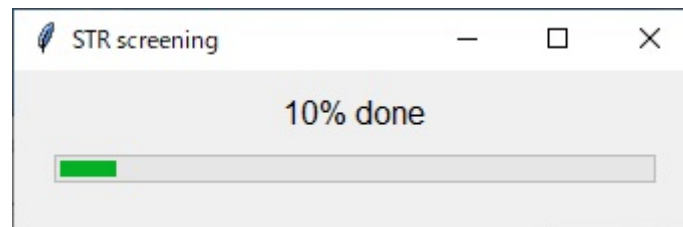
Buttons at the bottom: Edit, Add, Delete.

Perform screening

1. Press the 'Screening' button.

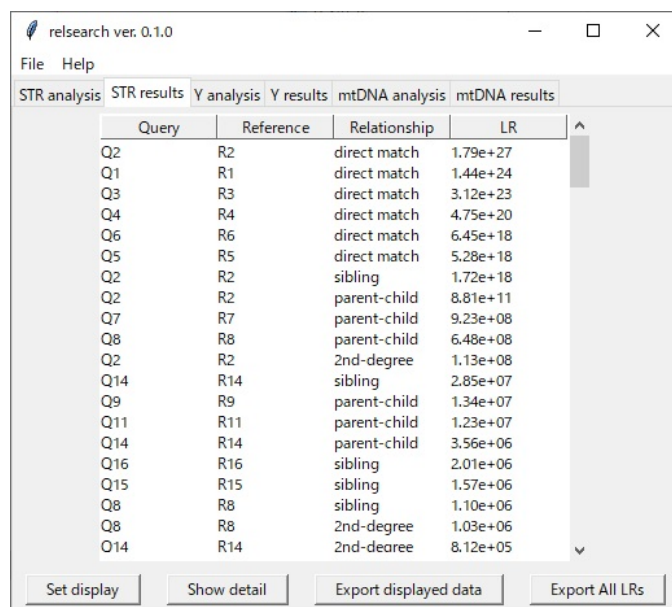


2. Wait until the screening finishes.



Check the result of screening

1. After finishing the screening, the result of the screening will be displayed.



The screenshot shows the 'relsearch ver. 0.1.0' application window. It has a menu bar with 'File' and 'Help'. Below the menu bar are tabs for 'STR analysis', 'STR results', 'Y analysis', 'Y results', 'mtDNA analysis', and 'mtDNA results'. The 'STR results' tab is active, displaying a table with four columns: 'Query', 'Reference', 'Relationship', and 'LR'. The table lists 20 results, sorted by LR in descending order. At the bottom of the window are four buttons: 'Set display', 'Show detail', 'Export displayed data', and 'Export All LRs'.

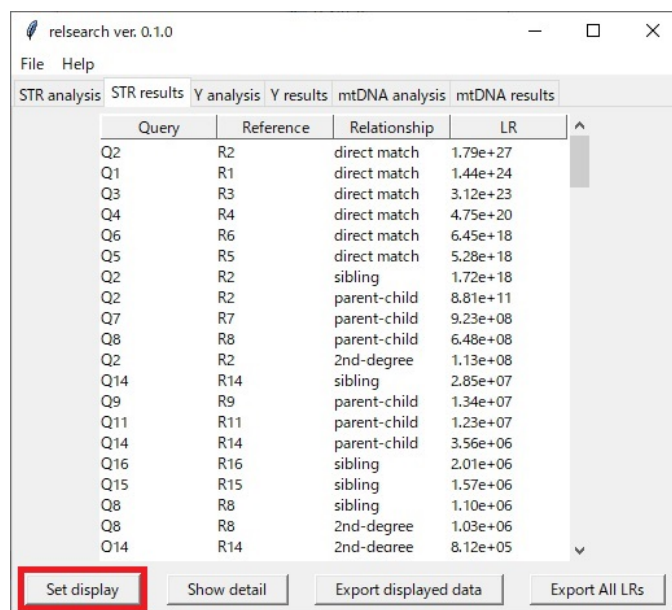
Query	Reference	Relationship	LR
Q2	R2	direct match	1.79e+27
Q1	R1	direct match	1.44e+24
Q3	R3	direct match	3.12e+23
Q4	R4	direct match	4.75e+20
Q6	R6	direct match	6.45e+18
Q5	R5	direct match	5.28e+18
Q2	R2	sibling	1.72e+18
Q2	R2	parent-child	8.81e+11
Q7	R7	parent-child	9.23e+08
Q8	R8	parent-child	6.48e+08
Q2	R2	2nd-degree	1.13e+08
Q14	R14	sibling	2.85e+07
Q9	R9	parent-child	1.34e+07
Q11	R11	parent-child	1.23e+07
Q14	R14	parent-child	3.56e+06
Q16	R16	sibling	2.01e+06
Q15	R15	sibling	1.57e+06
Q8	R8	sibling	1.10e+06
Q8	R8	2nd-degree	1.03e+06
Q14	R14	2nd-degree	8.12e+05

Note : Default display

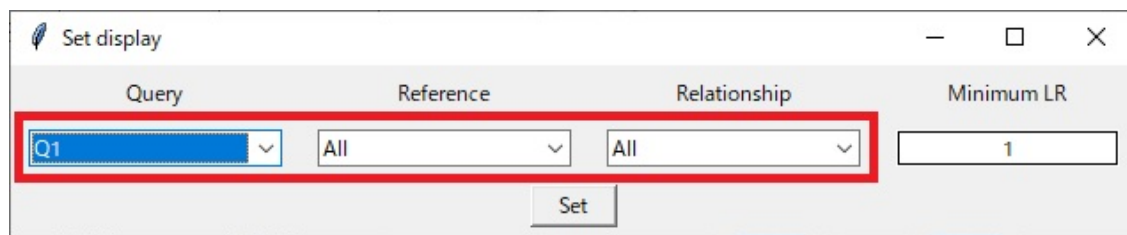
- All query names
- All reference names
- All relationships
- Likelihood ratio (LR) > 1
- Descending order of LR

Change the displayed information

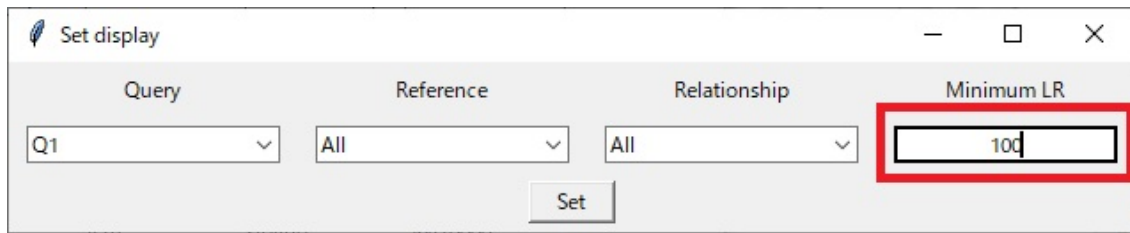
1. Press the 'Set display' button. Then the window 'Set display' will be open.



2. Select a query name, a reference name, and a relationship.



3. Enter the minimum LR.

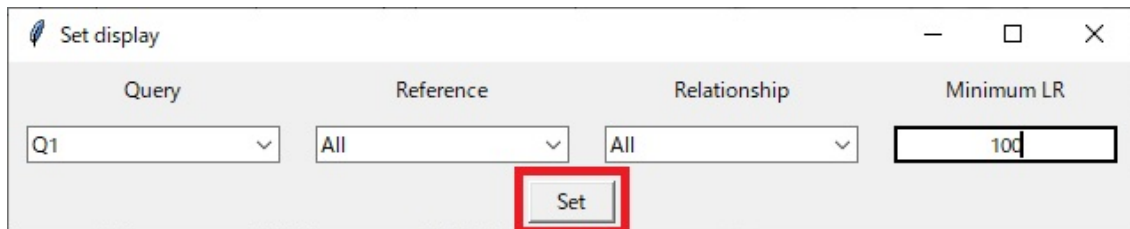


The 'Set display' dialog box is shown with the following fields and values:

Query	Reference	Relationship	Minimum LR
Q1	All	All	100

A red rectangle highlights the 'Minimum LR' input field, which contains the value '100'. A 'Set' button is located below the fields.

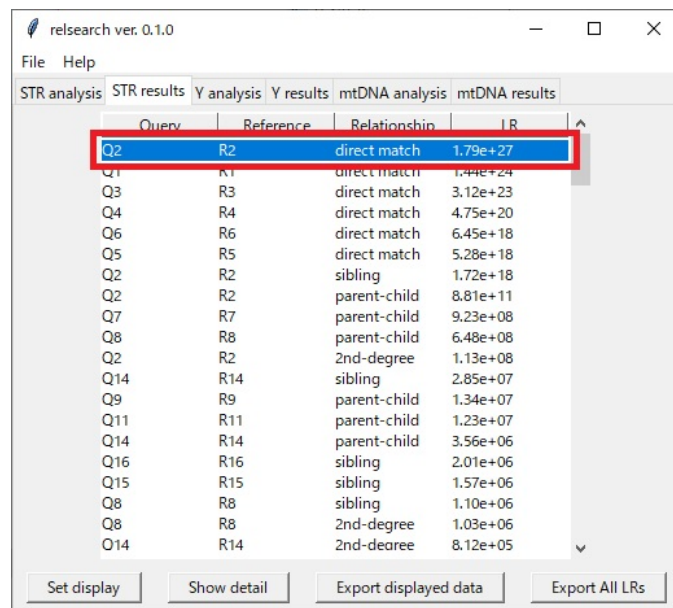
4. Press the 'Set' button.



The 'Set display' dialog box is shown with the same field values as the previous screenshot. A red rectangle highlights the 'Set' button, which is positioned below the 'Reference' and 'Relationship' fields.

Show the result of a query-reference pair in detail

1. Select a query-reference pair.



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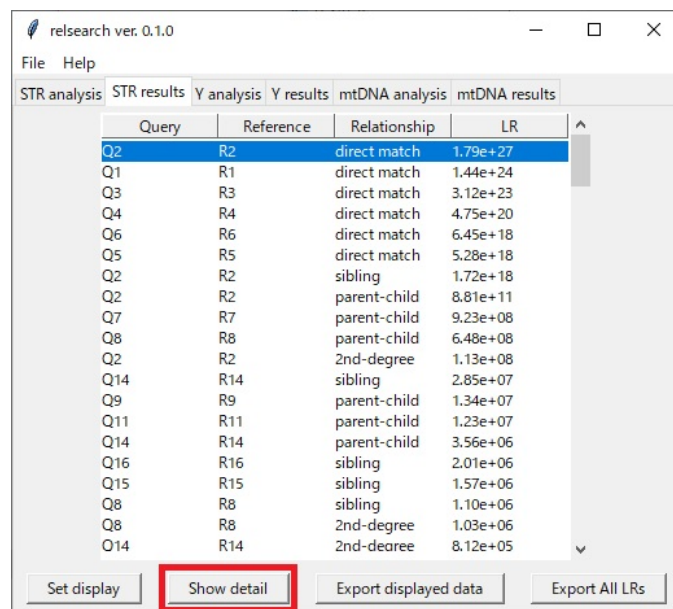
File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Query	Reference	Relationship	LR
Q2	R2	direct match	1.79e+27
Q1	R1	direct match	1.44e+24
Q3	R3	direct match	3.12e+23
Q4	R4	direct match	4.75e+20
Q6	R6	direct match	6.45e+18
Q5	R5	direct match	5.28e+18
Q2	R2	sibling	1.72e+18
Q2	R2	parent-child	8.81e+11
Q7	R7	parent-child	9.23e+08
Q8	R8	parent-child	6.48e+08
Q2	R2	2nd-degree	1.13e+08
Q14	R14	sibling	2.85e+07
Q9	R9	parent-child	1.34e+07
Q11	R11	parent-child	1.23e+07
Q14	R14	parent-child	3.56e+06
Q16	R16	sibling	2.01e+06
Q15	R15	sibling	1.57e+06
Q8	R8	sibling	1.10e+06
Q8	R8	2nd-degree	1.03e+06
Q14	R14	2nd-degree	8.12e+05

Set display Show detail Export displayed data Export All LRs

2. Press the 'Show detail' button. Then the window 'STR result in detail' will be open.



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File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Query	Reference	Relationship	LR
Q2	R2	direct match	1.79e+27
Q1	R1	direct match	1.44e+24
Q3	R3	direct match	3.12e+23
Q4	R4	direct match	4.75e+20
Q6	R6	direct match	6.45e+18
Q5	R5	direct match	5.28e+18
Q2	R2	sibling	1.72e+18
Q2	R2	parent-child	8.81e+11
Q7	R7	parent-child	9.23e+08
Q8	R8	parent-child	6.48e+08
Q2	R2	2nd-degree	1.13e+08
Q14	R14	sibling	2.85e+07
Q9	R9	parent-child	1.34e+07
Q11	R11	parent-child	1.23e+07
Q14	R14	parent-child	3.56e+06
Q16	R16	sibling	2.01e+06
Q15	R15	sibling	1.57e+06
Q8	R8	sibling	1.10e+06
Q8	R8	2nd-degree	1.03e+06
Q14	R14	2nd-degree	8.12e+05

Set display Show detail Export displayed data Export All LRs

3. Press the 'Export' button to save the displayed data.

STR result in detail

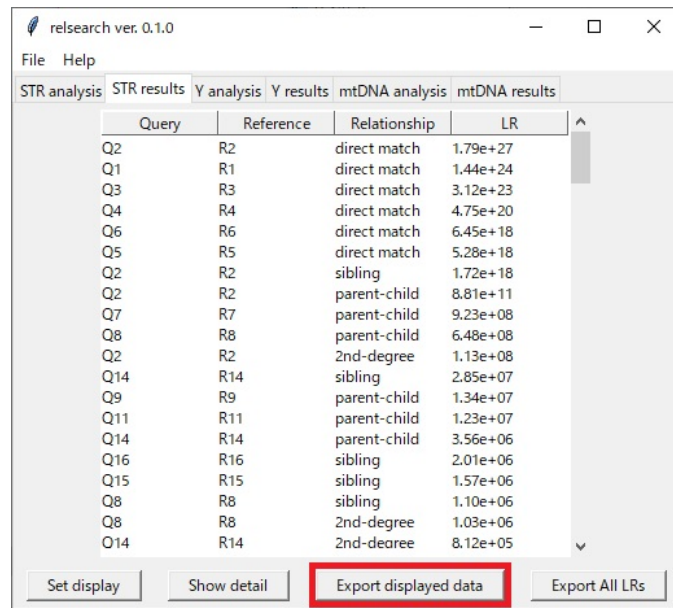
Relationship : direct match

locus	Query genotype (Q2)	Reference genotype (R2)	likelihood (related)	likelihood (unrelated)	LR
D3S1358	15, 17	15, 17	0.1602	0.02565	6.243
vWA	17, 18	17, 18	0.1284	0.0165	7.786
D16S539	11, 11	11, 11	0.03496	0.001222	28.6
CSF1PO	12, 13	12, 13	0.05842	0.003413	17.12
TPOX	11, 11	11, 11	0.1272	0.01617	7.864
D8S1179	14, 15	14, 15	0.05668	0.003213	17.64
D21S11	30, 30	30, 30	0.116	0.01346	8.619
D18S51	13, 18	13, 18	0.01851	0.0003426	54.03
D2S441	9, 1, 14	9, 1, 14	0.007302	5.332e-05	136.9
D19S433	13, 14	13, 14	0.1995	0.0398	5.013
TH01	7, 9	7, 9	0.2136	0.04564	4.681
FGA	24, 25	24, 25	0.02416	0.0005838	41.39
D22S1045	15, 16	15, 16	0.1542	0.02377	6.486
D5S818	11, 11	11, 11	0.08272	0.006843	12.09
D13S317	12, 12	12, 12	0.04091	0.001674	24.44
D7S820	12, 12	12, 12	0.05195	0.002699	19.25
SE33	16, 17	16, 17	0.00189	3.573e-06	529
D10S1248	13, 16	13, 16	0.05039	0.002539	19.85
D1S1656	14, 15	14, 15	0.03888	0.001512	25.72
D12S391	18, 20	18, 20	0.08656	0.007493	11.55

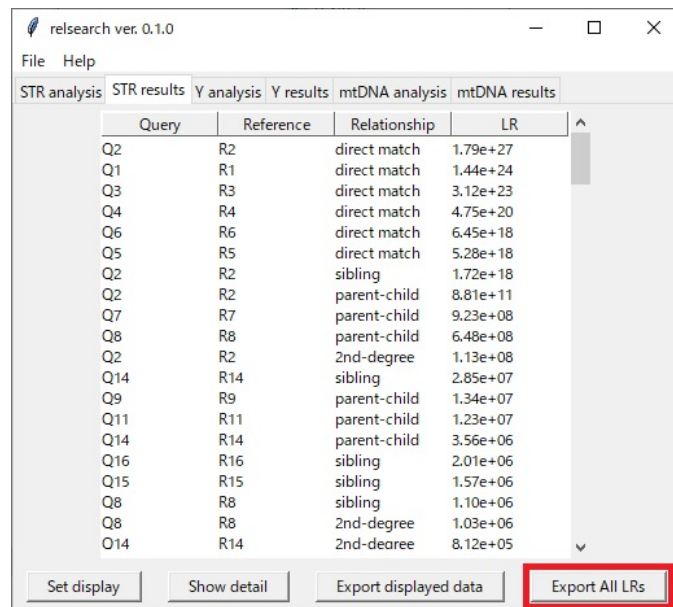
Export

Export data

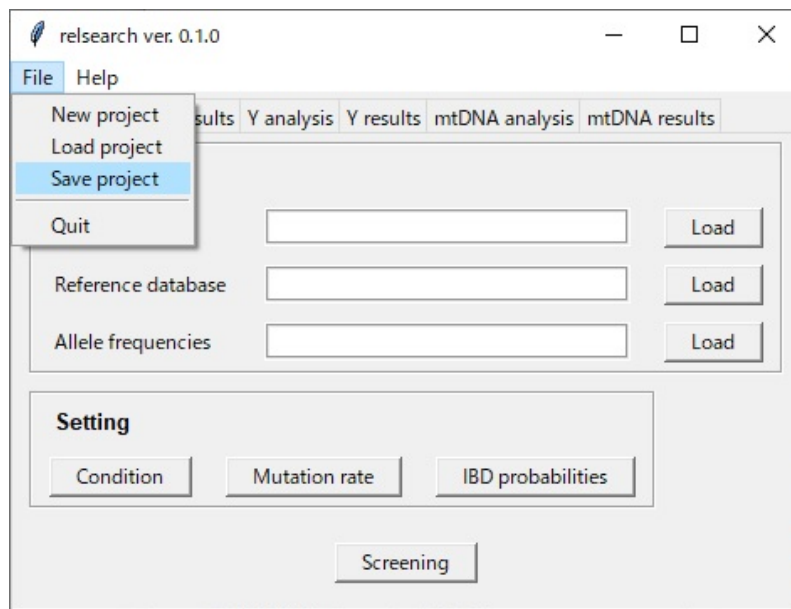
1. Press the 'Export displayed data' to save the displayed data in the tab 'STR results'.



2. Press the 'Export All LRs' to save all LR values.



3. Select **File -> Save project** to save current project data.



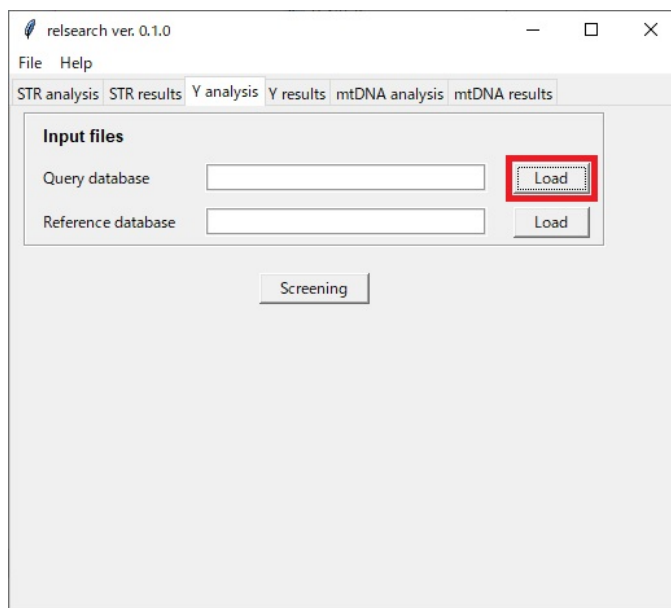
Note

- Users can load the saved project data. Select **File -> Load project**.
- Users can start a new project. Select **File -> New project**.

Y-chromosomal STR

Load query database

1. Press the 'Load' button for query database.



2. Select a .csv file for query database. Format of query database is shown in Fig. 4.

Sample Name	DYS456	DYS390	DYS438	DYS392	DYS518	DYS570	DYS437	DYS385
Q1	15	24	10	11	37	17	14	13,17
Q2	15	22	13	13	38	19	14	10,20
Q3	15	25	10	11	37	17	14	14
Q4	15	26	11	11	37	16	14	13
Q5	15	23	11			18		13,17
Q6				11	40		14	14
Q7	15	23	10	11	38	16	14	11,19
Q8	16	24	10	11	38	19	14	13,15
Q9	14	24	11	14	37	18	14	13,18
Q10	16	24	11	14		16	15	13,17

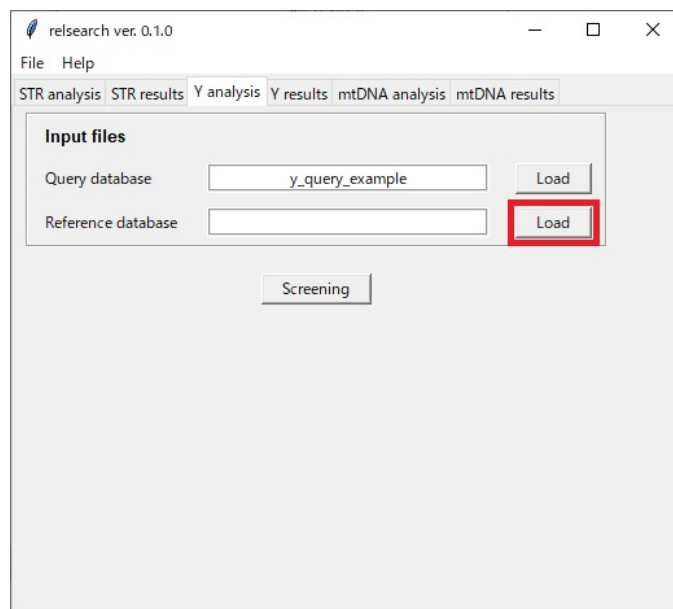
Figure 4: Format of query database for Y-STR

Note : format of query database

- This file must include information regarding ‘Sample Name’ and each marker.
- There is one column in each marker.
- In the marker with more than one allele, each allele must be separated by a comma (e.g., DYS385).
- The marker with an empty cell (e.g., DYS456 of sample ‘Q6’ in Fig. 4) is regarded as the ‘ignored loci’.
- An example file named “y_query_example.csv” is located at `extdata > examples`.

Load reference database

1. Press the 'Load' button for reference database.



2. Select a .csv file for reference database. Format of reference database is shown in Fig. 5.

Sample Name	DYS456	DYS390	DYS438	DYS392	DYS518	DYS570	DYS437	DYS385
R1	15	24	10	11	37	17	14	13,17
R2	15	22	13	13	38	19	14	10,20
R3	15	25	10	11	37	17	14	14,17
R4	15	26	11	11	37	16	14	13,16
R5	15	23	11	12	42	18	14	13,17
R6	16	26	10	11	40	20	14	14
R7	15	23	10	11	38	16	14	11,19
R8	16	24	10	11	38	19	14	13,15
R9	14	24	11	14	37	18	14	13,18
R10	16	24	11	14	34	16	15	13,17

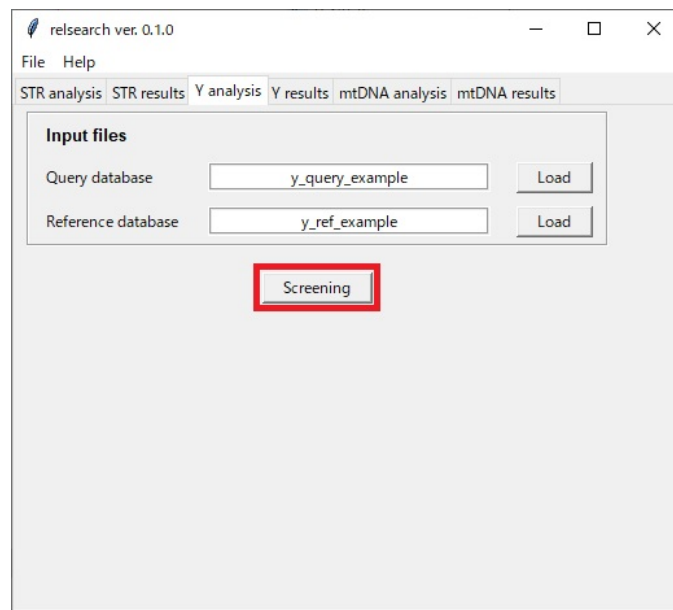
Figure 5: Format of reference database for Y-STR

Note : format of reference database

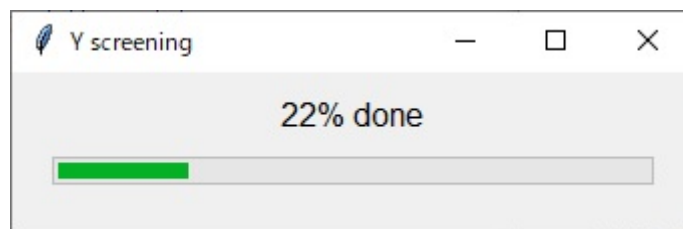
- This file must include information regarding ‘Sample Name’ and each marker.
- There is one column in each marker.
- In the marker with more than one allele, each allele must be separated by a comma (e.g., DYS385).
- An example file named “y_ref_example.csv” is located at extdata > examples.

Perform screening

1. Press the 'Screening' button.

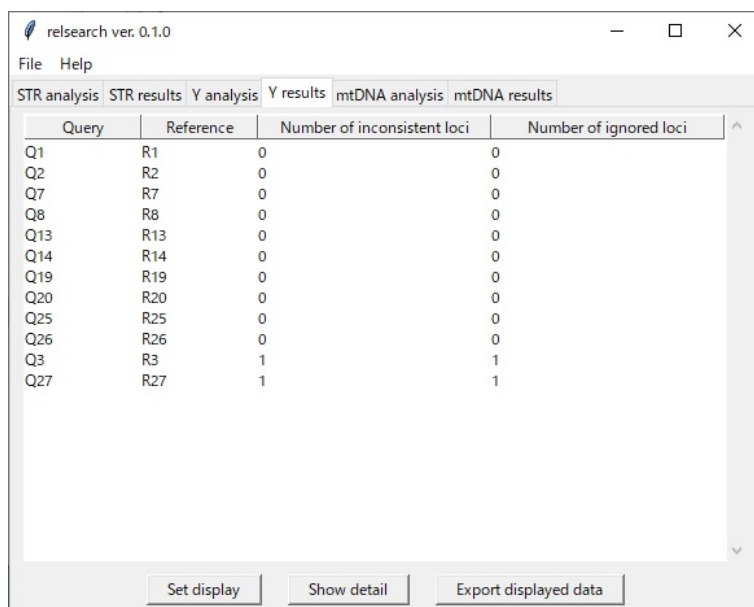


2. Wait until the screening finishes.



Check the result of screening

1. After finishing the screening, the result of the screening will be displayed.



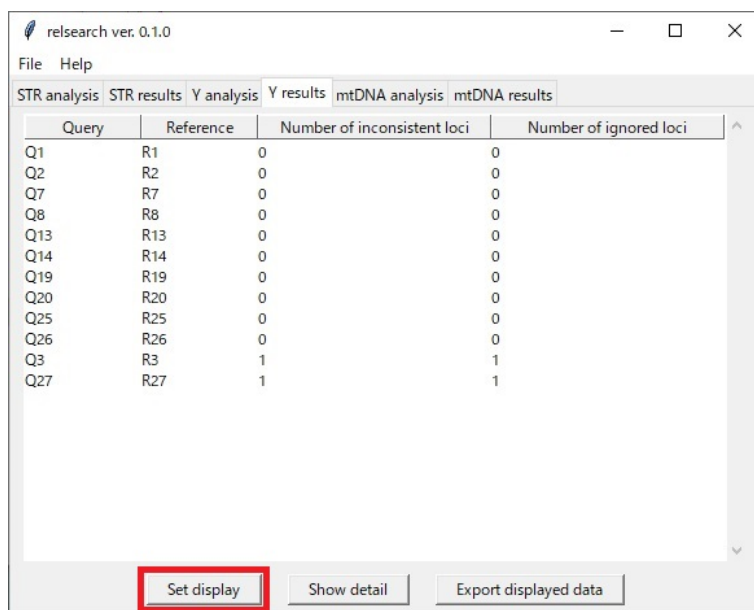
Query	Reference	Number of inconsistent loci	Number of ignored loci
Q1	R1	0	0
Q2	R2	0	0
Q7	R7	0	0
Q8	R8	0	0
Q13	R13	0	0
Q14	R14	0	0
Q19	R19	0	0
Q20	R20	0	0
Q25	R25	0	0
Q26	R26	0	0
Q3	R3	1	1
Q27	R27	1	1

Note : Default display

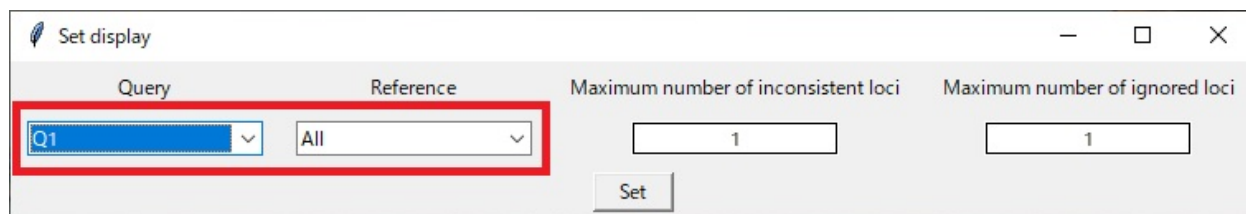
- All query names
- All reference names
- Number of inconsistent loci : 0 or 1
- Number of ignored loci : 0 or 1
- Ascending order of the number of inconsistent loci and the number of ignored loci

Change the displayed information

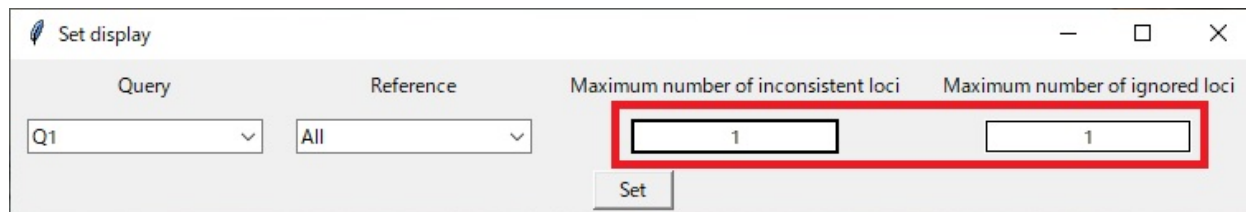
1. Press the 'Set display' button. Then the window 'Set display' will be open.



2. Select a query name and a reference name.



3. Enter the minimum number of inconsistent loci and the maximum number of loci explained by drop-outs.

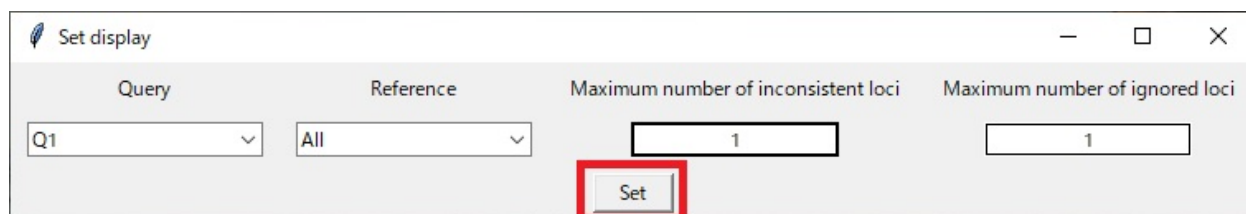


The 'Set display' dialog box is shown with the following fields and values:

Query	Reference	Maximum number of inconsistent loci	Maximum number of ignored loci
Q1	All	1	1

A red rectangle highlights the two input fields for the maximum number of inconsistent and ignored loci, both of which contain the value '1'. A 'Set' button is located below these fields.

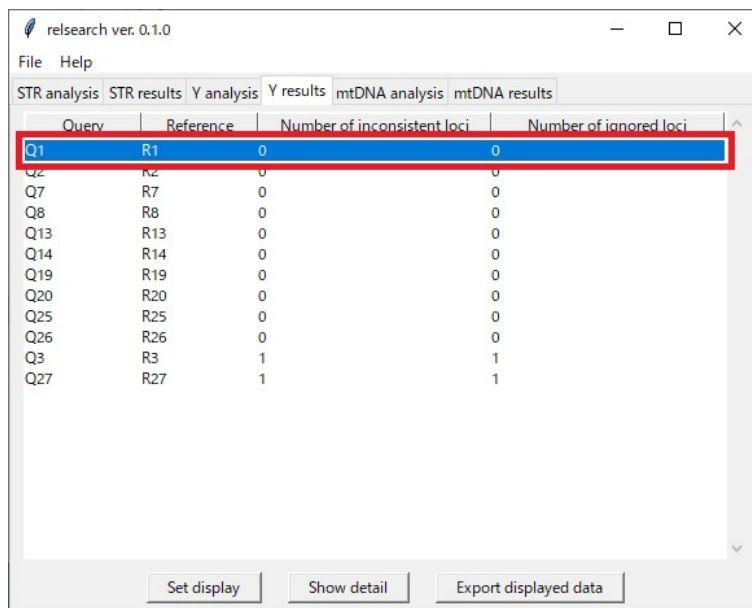
4. Press the 'Set' button.



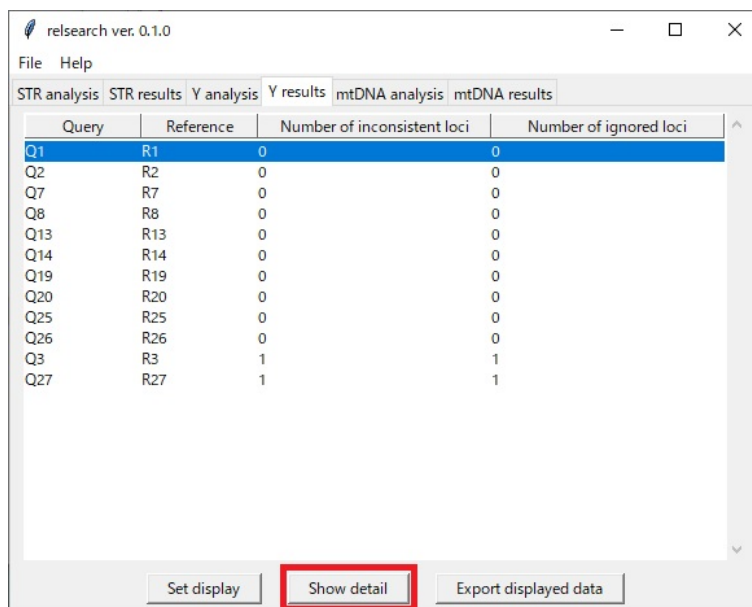
The 'Set display' dialog box is shown with the same field values as the previous screenshot. In this view, the 'Set' button is highlighted with a red rectangle, indicating it should be pressed.

Show the result of a query-reference pair in detail

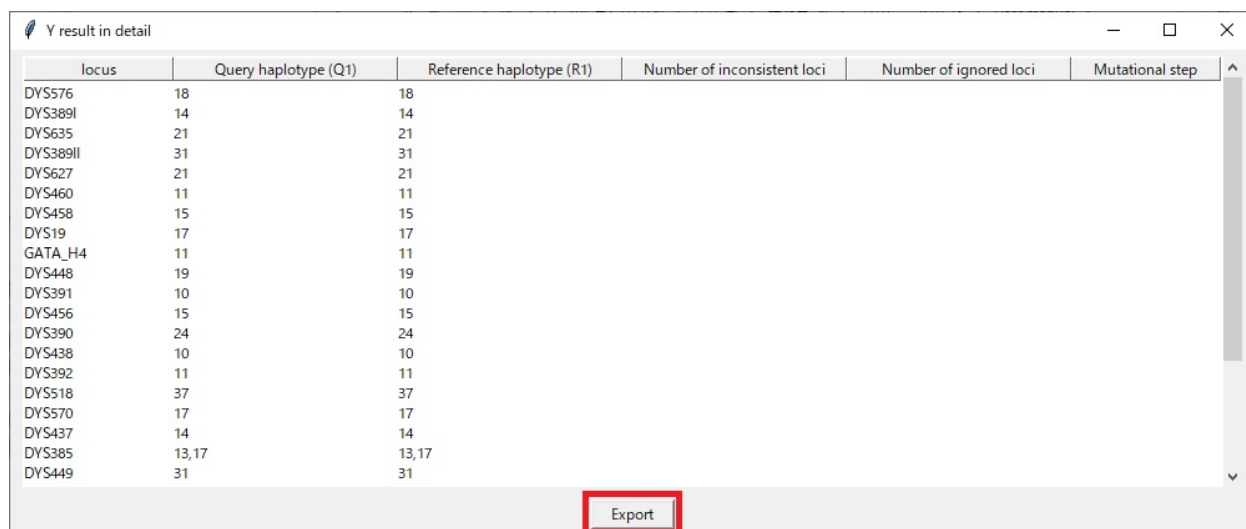
1. Select a query-reference pair.



2. Press the 'Show detail' button. Then the window 'Y result in detail' will be open.



3. Press the 'Export' button to save the displayed data.



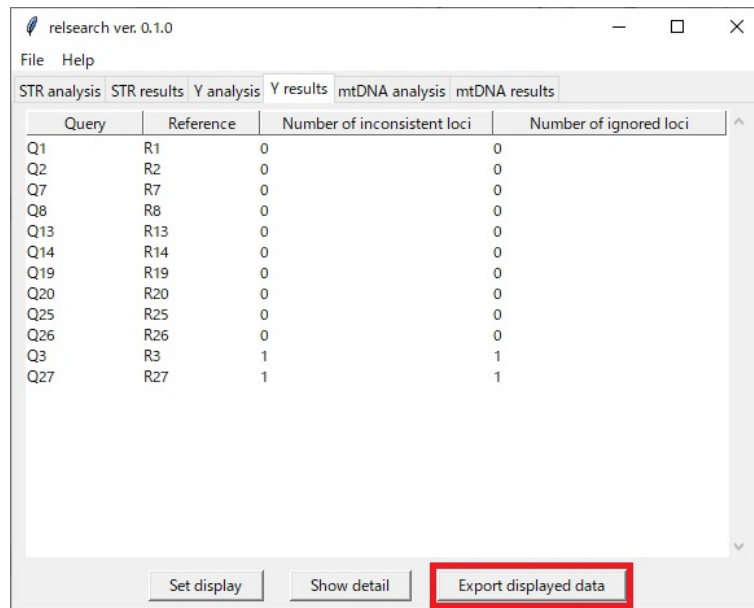
Y result in detail

locus	Query haplotype (Q1)	Reference haplotype (R1)	Number of inconsistent loci	Number of ignored loci	Mutational step
DYS576	18	18			
DYS389I	14	14			
DYS635	21	21			
DYS389II	31	31			
DYS627	21	21			
DYS460	11	11			
DYS458	15	15			
DYS19	17	17			
GATA_H4	11	11			
DYS448	19	19			
DYS391	10	10			
DYS456	15	15			
DYS390	24	24			
DYS438	10	10			
DYS392	11	11			
DYS518	37	37			
DYS570	17	17			
DYS437	14	14			
DYS385	13,17	13,17			
DYS449	31	31			

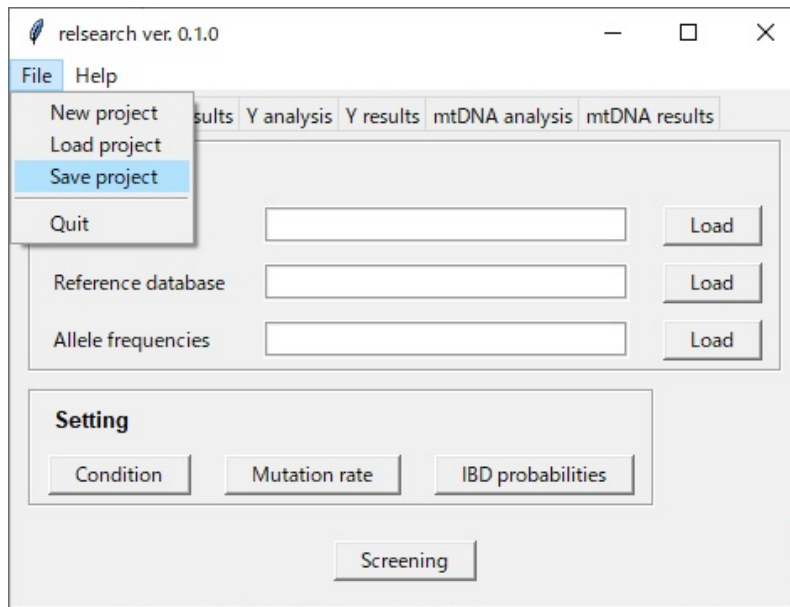
Export

Export data

1. Press the 'Export displayed data' to save the displayed data in the tab 'Y results'.



2. Select **File -> Save project** to save current project data.



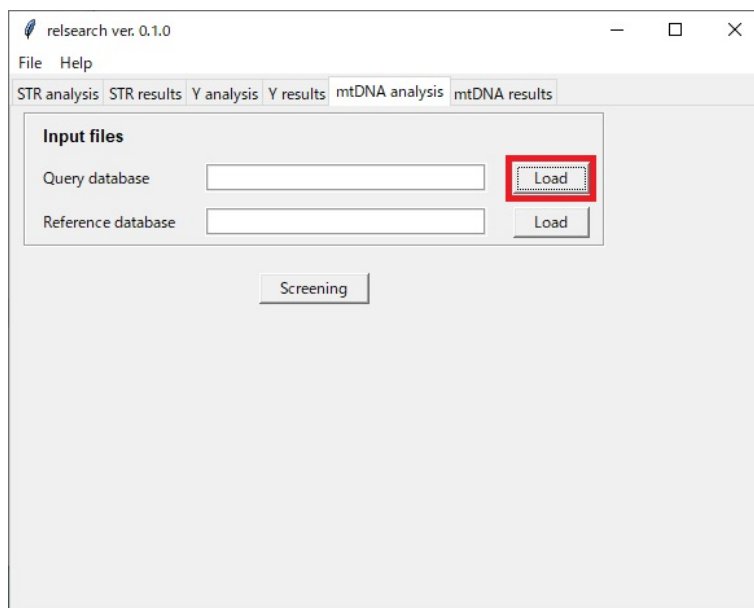
Note

- Users can load the saved project data. Select **File -> Load project**.
- Users can start a new project. Select **File -> New project**.

Mitochondrial DNA

Load query database

1. Press the 'Load' button for query database.



2. Select a .csv file for query database. Format of query database is shown in Fig. 6.

Sample Name	Range	Haplotype
Q1	73-340 16024-16365	16183C 16189C 16217C 16311C 73G 263G 309.1C 315.1C
Q2	73-340 16024-16365	16093C 16114A 16223T 16362C 73G 191.1A 194T 263G 309.1C 315.1C
Q3	73-167 240-340 16024-16365	16095T 16189C 16223T 16265C 16274A 16362C 73G 143A 152C 263G 315.1C
Q4	73-265 16024-16284	16223T 73G 152C 263G
Q5	16117-16365	16140C 16182C 16183C 16189C 16234T 16243C 16291T
Q6	73-167 16117-16209	16172C 73G 150T
Q7	73-340 16024-16365	16172C 16223T 16257A 16261T 73G 150T 263G 309.1C 309.2C 315.1C
Q8	73-340 16024-16365	16129A 16183C 16189C 16223T 16297C 16298C 16311C 73G 150T 199C 263G 309.1C 309.2C 315.1C
Q9	73-340 16024-16132 16179-16365	16183C 16189C 16209C 16223T 16324C 73G 207A 263G 284G 309.1C 309.2C 315.1C
Q10	73-265 16024-16209 16266-16365	16362C 73G 263G

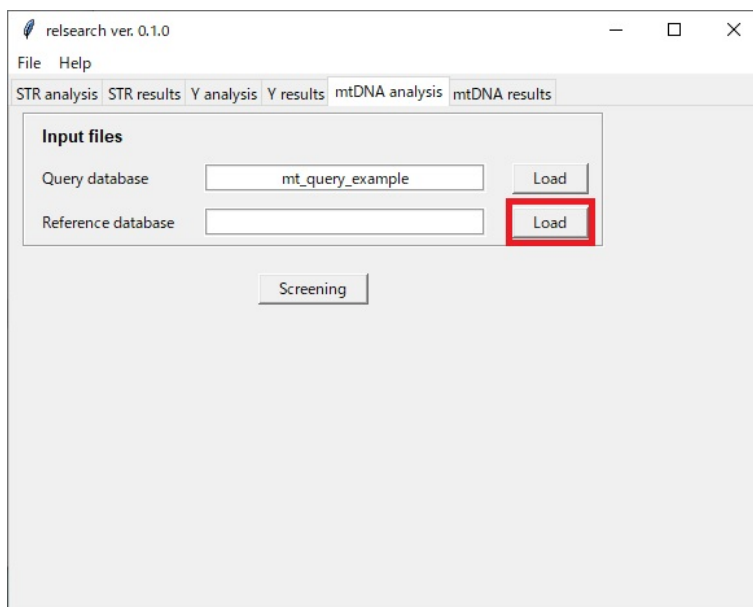
Figure 6: Format of query database for mtDNA

Note : format of query database

- This file must include information regarding 'Sample Name', 'Range', and 'Haplotype'.
- An example file named "mt_query_example.csv" is located at extdata > examples.

Load reference database

1. Press the 'Load' button for reference database.



2. Select a .csv file for reference database. Format of reference database is shown in Fig. 7.

Sample Name	Range	Haplotype
R1	16024-16365 73-340	16183C 16189C 16217C 16311C 73G 263G 309.1C 315.1C
R2	16024-16365 73-340	16093C 16114A 16223T 16362C 73G 191.1A 194T 263G 309.1C 315.1C
R3	16024-16365 73-340	16095T 16189C 16223T 16265C 16274A 16362C 73G 143A 152C 263G 315.1C
R4	16024-16365 73-340	16223T 16319A 16362C 73G 152C 263G 309.1C 315.1C
R5	16024-16365 73-340	16111T 16140C 16182C 16183C 16189C 16234T 16243C 16291T 73G 131C 195C 204C 263G 309.1C 309.2C 315.1C
R6	16024-16365 73-340	16172C 16223T 16250T 16257A 16261T 73G 150T 263G 309.1C 315.1C
R7	16024-16365 73-340	16172C 16189C 16223T 16355T 16362C 73G 150T 263G 309.1C 315.1C
R8	16024-16365 73-340	16223T 16362C 73G 263G 315.1C
R9	16024-16365 73-340	16183G 16223T 16274A 16290T 16319A 16362C 73G 195C 263G 309.1C 315.1C
R10	16024-16365 73-340	16111T 16140C 16154C 16183C 16189C 16217C 16261T 16274A 73G 263G 315.1C

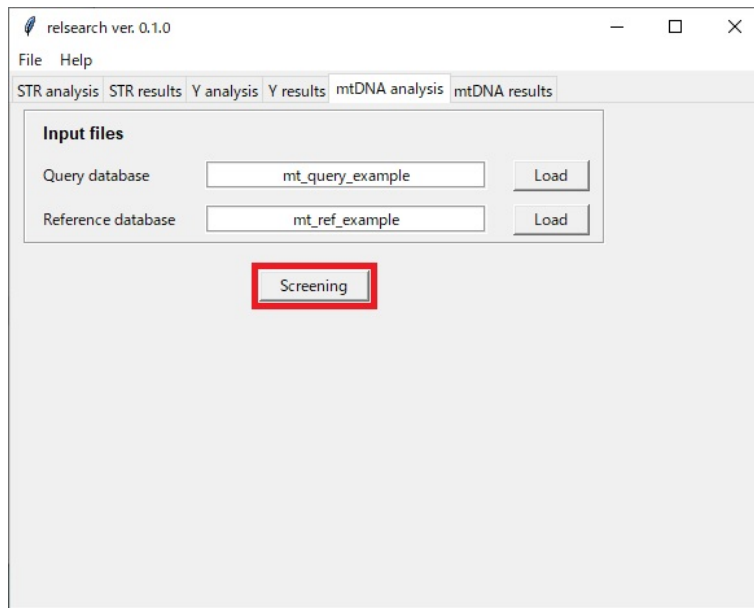
Figure 7: Format of reference database for mtDNA

Note : format of reference database

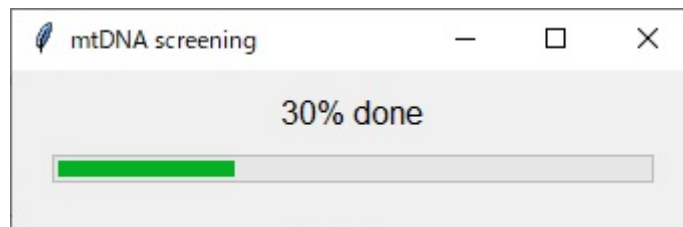
- This file must include information regarding 'Sample Name', 'Range', and 'Haplotype'.
- An example file named "mt_ref_example.csv" is located at `extdata > examples`.

Perform screening

1. Press the 'Screening' button.



2. Wait until the screening finishes.



Check the result of screening

1. After finishing the screening, the result of the screening will be displayed.

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File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Query	Reference	Shared range	Shared length	Number of inconsistency
Q1	R1	73-340 16024-16365	610	0
Q2	R2	73-340 16024-16365	610	0
Q13	R13	73-340 16024-16365	610	0
Q14	R14	73-340 16024-16365	610	0
Q26	R15	73-340 16024-16365	610	0
Q11	R16	73-340 16024-16209 16266-16365	554	0
Q3	R3	73-167 240-340 16024-16365	538	0
Q15	R15	73-167 240-340 16024-16209 16266-16365	482	0
Q10	R8	73-265 16024-16209 16266-16365	479	0
Q10	R18	73-265 16024-16209 16266-16365	479	0
Q4	R4	73-265 16024-16284	454	0
Q16	R16	73-340 16024-16209	454	0
Q17	R17	142-340 16024-16132 16179-16284	414	0
Q18	R8	142-265 16117-16209 16266-16365	317	0
Q18	R18	142-265 16117-16209 16266-16365	317	0
Q7	R19	73-340 16024-16365	610	1
Q12	R18	142-340 16024-16209 16266-16365	485	1
Q10	R11	73-265 16024-16209 16266-16365	479	1
Q10	R14	73-265 16024-16209 16266-16365	479	1
Q10	R17	73-265 16024-16209 16266-16365	479	1

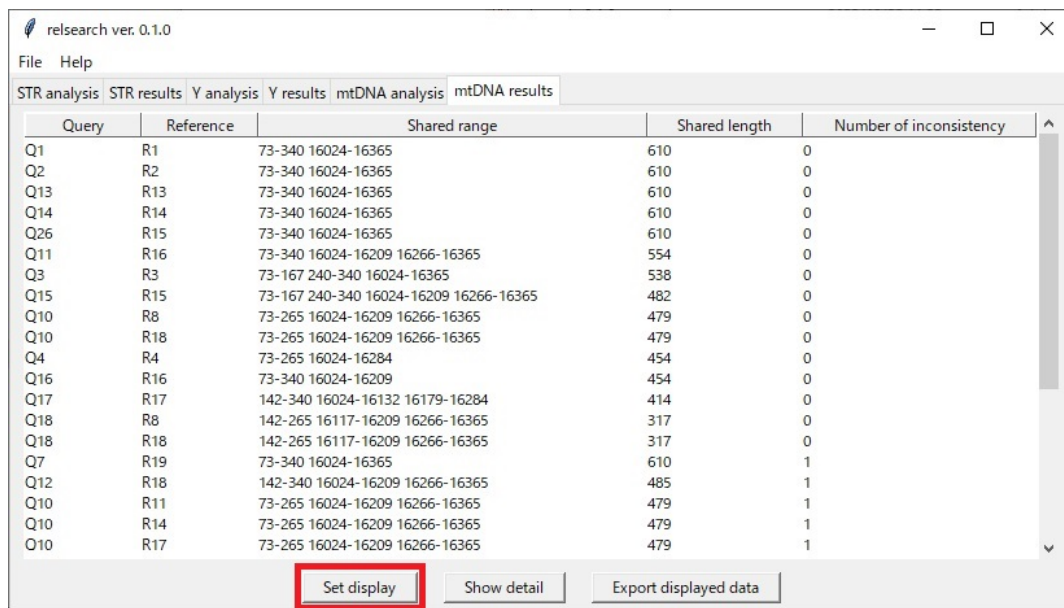
Set display Show detail Export displayed data

Note : Default display

- All query names
- All reference names
- Shared length > 300
- Number of inconsistency : 0 or 1
- Descending order of the shared length
- Ascending order of the number of inconsistency

Change the displayed information

1. Press the 'Set display' button. Then the window 'Set display' will be open.



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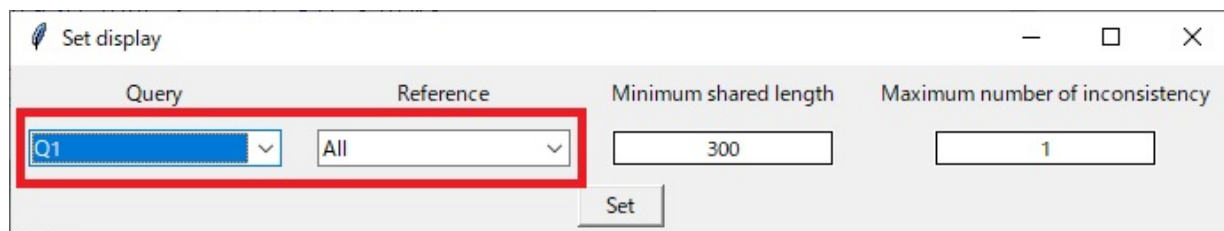
File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Query	Reference	Shared range	Shared length	Number of inconsistency
Q1	R1	73-340 16024-16365	610	0
Q2	R2	73-340 16024-16365	610	0
Q13	R13	73-340 16024-16365	610	0
Q14	R14	73-340 16024-16365	610	0
Q26	R15	73-340 16024-16365	610	0
Q11	R16	73-340 16024-16209 16266-16365	554	0
Q3	R3	73-167 240-340 16024-16365	538	0
Q15	R15	73-167 240-340 16024-16209 16266-16365	482	0
Q10	R8	73-265 16024-16209 16266-16365	479	0
Q10	R18	73-265 16024-16209 16266-16365	479	0
Q4	R4	73-265 16024-16284	454	0
Q16	R16	73-340 16024-16209	454	0
Q17	R17	142-340 16024-16132 16179-16284	414	0
Q18	R8	142-265 16117-16209 16266-16365	317	0
Q18	R18	142-265 16117-16209 16266-16365	317	0
Q7	R19	73-340 16024-16365	610	1
Q12	R18	142-340 16024-16209 16266-16365	485	1
Q10	R11	73-265 16024-16209 16266-16365	479	1
Q10	R14	73-265 16024-16209 16266-16365	479	1
Q10	R17	73-265 16024-16209 16266-16365	479	1

Set display Show detail Export displayed data

2. Select a query name and a reference name.



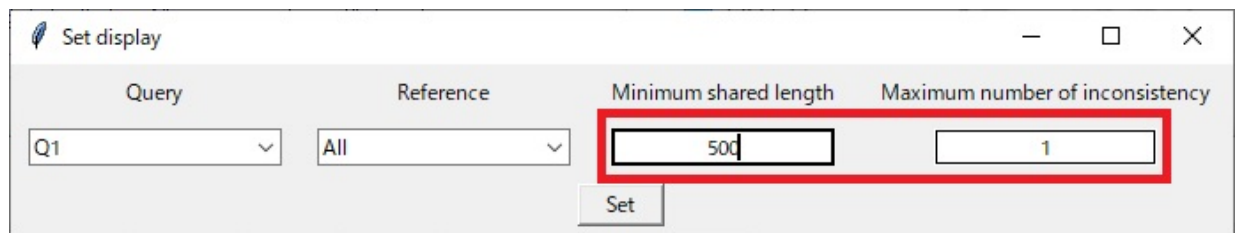
Set display

Query Reference Minimum shared length Maximum number of inconsistency

Q1 All 300 1

Set

3. Enter the minimum shared length and the maximum number of inconsistency.



The screenshot shows a 'Set display' dialog box with a title bar containing a feather icon, the text 'Set display', and standard window controls (minimize, maximize, close). The dialog has four input fields: 'Query' (dropdown menu with 'Q1' selected), 'Reference' (dropdown menu with 'All' selected), 'Minimum shared length' (text box with '500'), and 'Maximum number of inconsistency' (text box with '1'). A red rectangular box highlights the 'Minimum shared length' and 'Maximum number of inconsistency' fields. Below these fields is a 'Set' button.

4. Press the 'Set' button.



This screenshot is identical to the previous one, showing the 'Set display' dialog box with the same settings. However, a red rectangular box now highlights the 'Set' button located below the input fields.

Show the result of a query-reference pair in detail

1. Select a query-reference pair.

relsearch ver. 0.1.0

File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Query	Reference	Shared range	Shared length	Number of inconsistency
Q1	R1	73-340 16024-16365	610	0
Q2	R2	73-340 16024-16365	610	0
Q13	R13	73-340 16024-16365	610	0
Q14	R14	73-340 16024-16365	610	0
Q26	R15	73-340 16024-16365	610	0
Q11	R16	73-340 16024-16209 16266-16365	554	0
Q3	R3	73-167 240-340 16024-16365	538	0
Q15	R15	73-167 240-340 16024-16209 16266-16365	482	0
Q10	R8	73-265 16024-16209 16266-16365	479	0
Q10	R18	73-265 16024-16209 16266-16365	479	0
Q4	R4	73-265 16024-16284	454	0
Q16	R16	73-340 16024-16209	454	0
Q17	R17	142-340 16024-16132 16179-16284	414	0
Q18	R8	142-265 16117-16209 16266-16365	317	0
Q18	R18	142-265 16117-16209 16266-16365	317	0
Q7	R19	73-340 16024-16365	610	1
Q12	R18	142-340 16024-16209 16266-16365	485	1
Q10	R11	73-265 16024-16209 16266-16365	479	1
Q10	R14	73-265 16024-16209 16266-16365	479	1
Q10	R17	73-265 16024-16209 16266-16365	479	1

Set display Show detail Export displayed data

2. Press the 'Show detail' button. Then the window 'mtDNA result in detail' will be open.

relsearch ver. 0.1.0

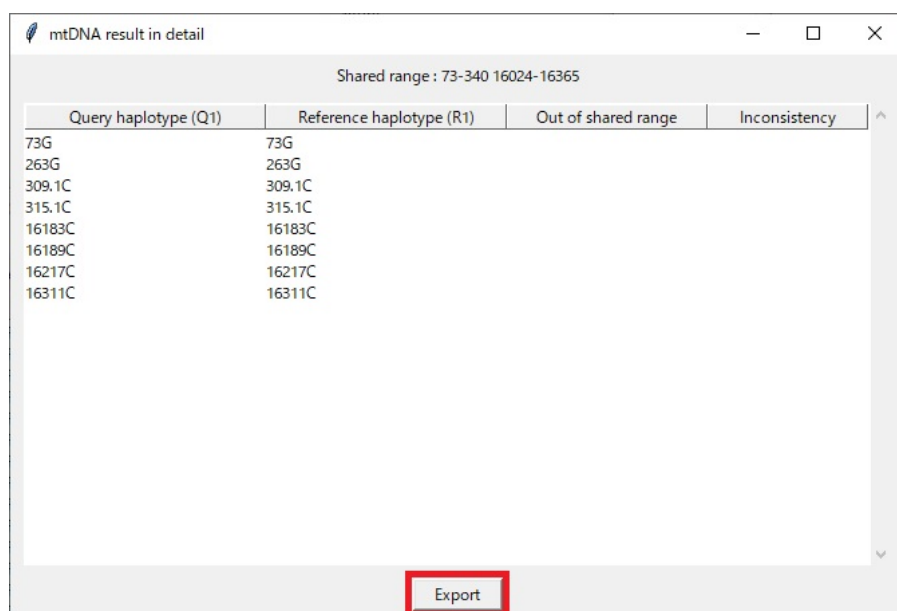
File Help

STR analysis STR results Y analysis Y results mtDNA analysis mtDNA results

Query	Reference	Shared range	Shared length	Number of inconsistency
Q1	R1	73-340 16024-16365	610	0
Q2	R2	73-340 16024-16365	610	0
Q13	R13	73-340 16024-16365	610	0
Q14	R14	73-340 16024-16365	610	0
Q26	R15	73-340 16024-16365	610	0
Q11	R16	73-340 16024-16209 16266-16365	554	0
Q3	R3	73-167 240-340 16024-16365	538	0
Q15	R15	73-167 240-340 16024-16209 16266-16365	482	0
Q10	R8	73-265 16024-16209 16266-16365	479	0
Q10	R18	73-265 16024-16209 16266-16365	479	0
Q4	R4	73-265 16024-16284	454	0
Q16	R16	73-340 16024-16209	454	0
Q17	R17	142-340 16024-16132 16179-16284	414	0
Q18	R8	142-265 16117-16209 16266-16365	317	0
Q18	R18	142-265 16117-16209 16266-16365	317	0
Q7	R19	73-340 16024-16365	610	1
Q12	R18	142-340 16024-16209 16266-16365	485	1
Q10	R11	73-265 16024-16209 16266-16365	479	1
Q10	R14	73-265 16024-16209 16266-16365	479	1
Q10	R17	73-265 16024-16209 16266-16365	479	1

Set display Show detail Export displayed data

3. Press the 'Export' button to save the displayed data.



mtDNA result in detail

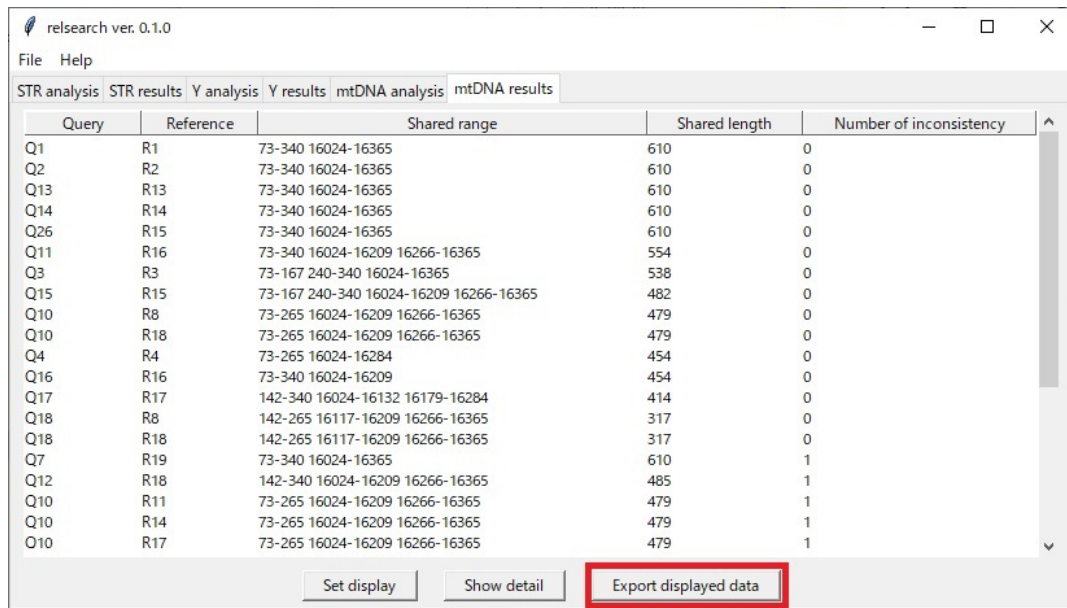
Shared range : 73-340 16024-16365

Query haplotype (Q1)	Reference haplotype (R1)	Out of shared range	Inconsistency
73G	73G		
263G	263G		
309.1C	309.1C		
315.1C	315.1C		
16183C	16183C		
16189C	16189C		
16217C	16217C		
16311C	16311C		

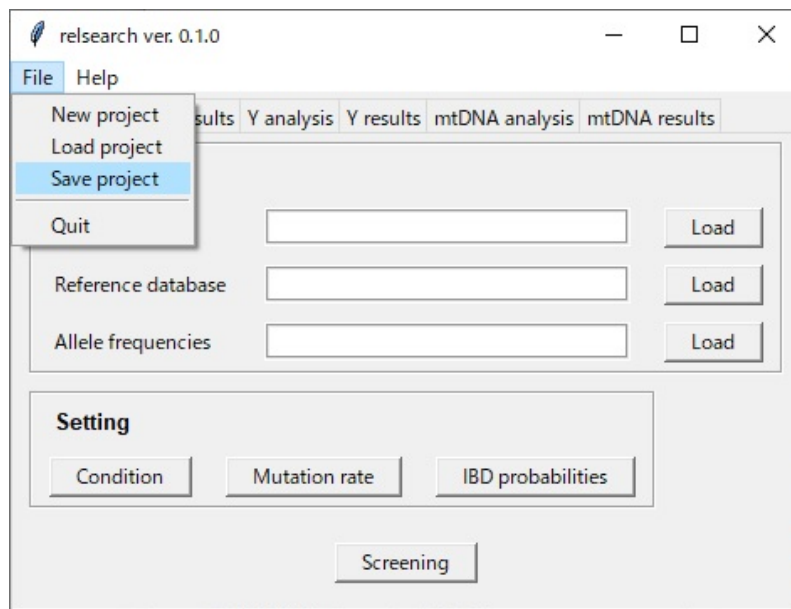
Export

Export data

1. Press the 'Export displayed data' to save the displayed data in the tab 'mtDNA results'.



2. Select **File -> Save project** to save current project data.



Note

- Users can load the saved project data. Select **File -> Load project**.
- Users can start a new project. Select **File -> New project**.