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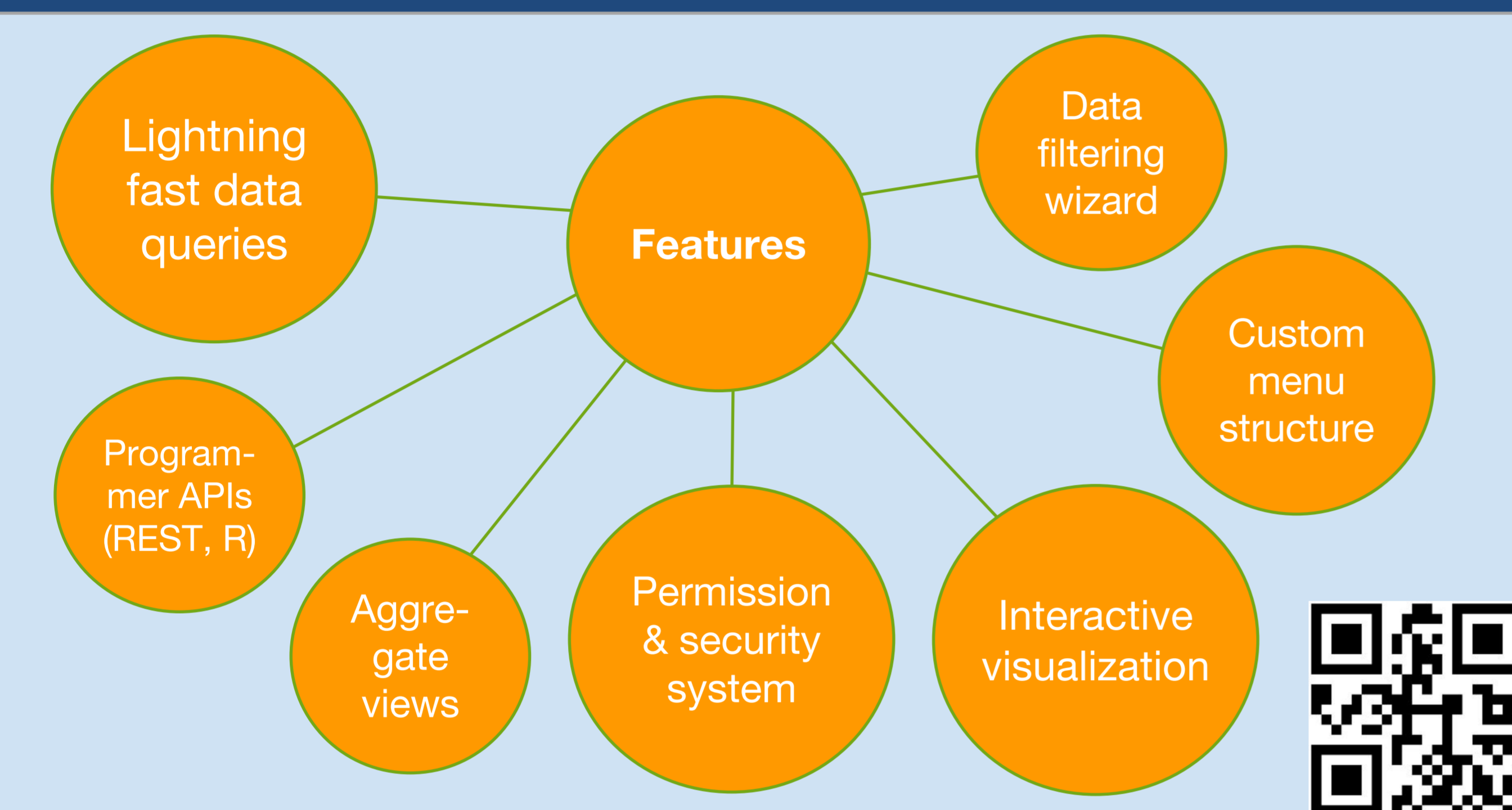
Biobanking and
BioMolecular resources
Research Infrastructure
The Netherlands

Towards a personalized genomics workbench using MOLGENIS

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For multi-omics and personalized medicine with BBMRI-NL, VKGL, BioShare, RD-Connect, LifeLines, BioMedBridges, CTMM/TraIT, et al.



Use simple Excel/CSV/VCF files to upload your molecular, phenotypical, and environmental data with a step-by-step wizard

Search data values

Data Item selection

| Identifier | mutation | mutationPosition | cdna_position | gfa_position | exon_pos | length | strand | type | exchange |
|------------|----------|------------------|---------------|--------------|----------|--------|--------|----------------|----------------------|
| M153 | 111 | 111 | 48621938 | 38 | 1 | 1 | + | insertion | nonense mutation |
| M204 | 112 | 112 | 48621938 | 38 | 1 | 1 | + | point mutation | missense mutation |
| M58 | 1264 | 1264 | 48629424 | 422 | 1 | 1 | + | duplication | nonense mutation |
| M354 | 1332 | 1332 | 48629356 | 444 | 1 | 1 | + | point mutation | nonense mutation |
| M241 | 1263 | 1263 | 48629200 | 453 | 1 | 1 | + | point mutation | nonense mutation |
| M714 | 143 | 143 | 48621938 | 48 | 1 | 1 | + | point mutation | missense mutation |
| M200 | 1452 | 1452 | 48629161 | 464 | 2 | 1 | + | deletion | nonense mutation |
| M188 | 1474 | 1474 | 48629139 | 482 | 11 | 1 | + | deletion | nonense mutation |
| M216 | 1478 | 1478 | 48629125 | 483 | 8 | 1 | + | deletion | nonense mutation |
| M202 | 1521 | 1521 | 48629168 | 5 | 1 | 1 | + | point mutation | splice-site mutation |
| M3 | 151 | 151 | 48621918 | 51 | 1 | 1 | + | point mutation | missense mutation |
| M22 | 1573 | 1573 | 48628860 | 525 | 1 | 1 | + | point mutation | missense mutation |
| M23 | 1732 | 1732 | 48628154 | 578 | 1 | 1 | + | point mutation | missense mutation |

login: admin/admin

Enrich your data with information from public molecular, genetic, clinical and pathogenicity knowledge bases using 'annotators'

Annotations available

- KEGG
- HGNC-Symbol
- Clinical Genomic Database
- OmimHpo

Annotations not available

- CADD
- Cosmic
- dbNSFP-Variant
- Clinvar
- dbNSFP-Gene
- EBI-CHEMBL

Run annotation Copy before annotating

Plug-ins for detailed research and clinic use: WikiPathways, candidate gene reporting, patient symptom-to-disease matching, etc.

Monogenic disease candidate report for

Candidate genes

High impact

WikiPathways

Clinical Synopsis

Symptoms: HP:000707, HP:0100022, HP:0001300, HP:0007325, HP:0002015, HP:0001336, HP:000496, HP:0001250

Add your statistics: create and use 'on the spot' R analyses with built-in R framework and R-API (example: A.S.E. @ www.molgenis.org/ase)

Add Script

```

1 if (require("KColorbrewer")) {
2   install.packages("KColorbrewer", dependencies = TRUE)
3 }
4 library(KColorbrewer)
5
6 ## B) Reading in data and transform 1s into matrix format
7 #####
8 data <- read.csv("../datasets/heatmaps_1s_r.csv", comment.char="#")
9
10 #names <- data[,1] # assign labels in column 1 to 'm
11 #names
12 #names <- data.matrix(data[,2:ncol(data)]) # transform column 2-5 into a matr
13 #names(names_data) <- #names # assign row names
14
15
16
17
18
19
20
21
22
23

```

Name: ASE plot

Parameters: Select ScriptParam

Result file extension: resultfileExtension

Percentage alternative

Reference allele count (G)

Total number

Query all shared variant data from e.g. VKGL* diagnostic labs in one genome browser

Search data values

Genome Browser

Human GRCh37/hg19

Variant details

| CHROM | POS | ID | REF | ALT | QUAL | FILTER | INFO |
|-------|---------|-----------|-----|-----|------|--|--|
| 1 | 7848217 | rs1782455 | G | A | 100 | PASS | AC=1;AN=2;DP=10;GQ=10;MQ=10;PL=0,100,100;R=100;S=100;T=100 |
| 1 | 7848201 | G | A | 100 | PASS | AC=1;AN=2;DP=10;GQ=10;MQ=10;PL=0,100,100;R=100;S=100;T=100 | |
| 1 | 7848045 | G | A | 100 | PASS | AC=1;AN=2;DP=10;GQ=10;MQ=10;PL=0,100,100;R=100;S=100;T=100 | |
| 1 | 7848045 | rs1160088 | G | A | 100 | PASS | AC=48;AN=48;DP=10;GQ=10;MQ=10;PL=0,100,100;R=100;S=100;T=100 |
| 1 | 7848013 | A | C | 100 | PASS | AC=1;AN=2;DP=10;GQ=10;MQ=10;PL=0,100,100;R=100;S=100;T=100 | |

GTC(UMCG) - 0,10,0

GTC(UMCG) - 138,8,0

0,10,1

0,10,2

203,12,0

203,12,0

0,10,2

203,12,0

0,10,2

179,11,0



umcg



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Project website: https://github.com/molgenis

