TEAM:

A **web tool** for the design, analysis and management of **panels of genes** for clinical applications

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TEAM: Targeted Enrichment Analysis and Management

Introduction

- **Development of high throughput sequencing technologies:**
  - Rapid and economical genome sequencing.
  - Disease targeted sequencing: powerful and cost-effective application.

- **Vast amount of biological knowledge available:**
  - HGMD-public, HUMSAVAR, ClinVar, COSMIC.

- We need a tool to connect **sequencing data and biological knowledge for diagnostic:**
  - **TEAM** (**T**argeted **E**nrichment **A**nalysis and **M**anagement).

TEAM | **Targeted Enrichment Analysis and Management**

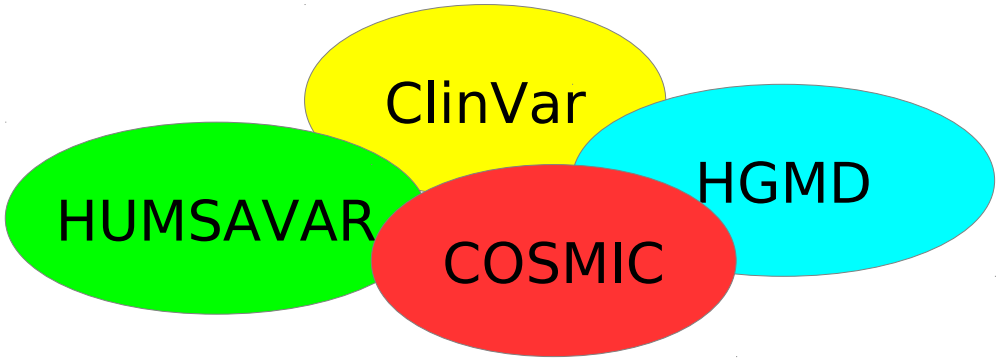
How does TEAM work?

1. VCF files
2. Gene panel

TEAM

Diagnostic: Retinitis

	Chromosome	Position	SNP Id	Ref	Alt	Gene	Con
1	3	129247734	.	T	C	.	exon ator
2	3	129247734	.	T	C	RHO	exon ator
3	3	129247734	.	T	C	RHO	exon ator
4	3	129247734	.	T	C	RHO	exon ator



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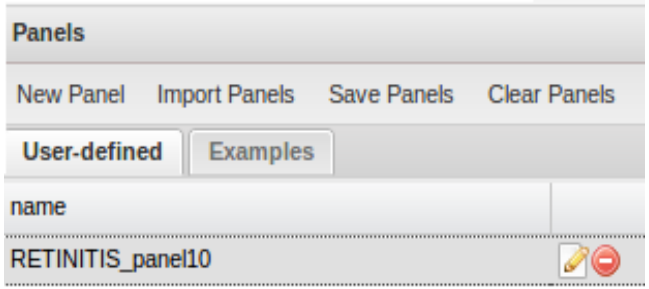
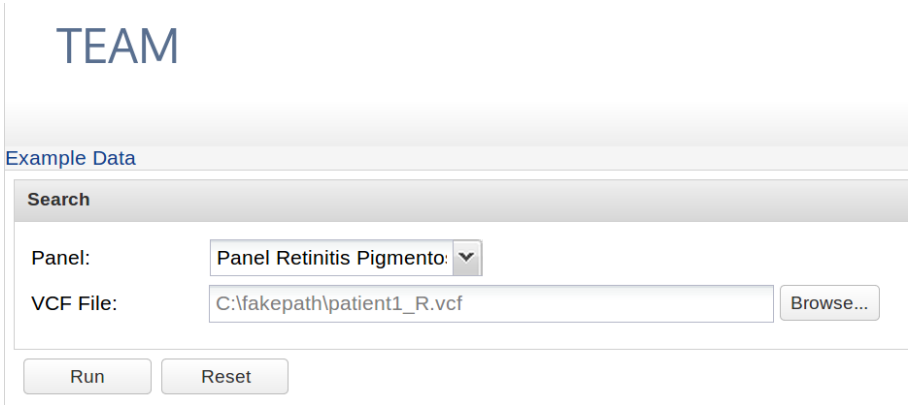
How does TEAM work?

http://team.babelomics.org/

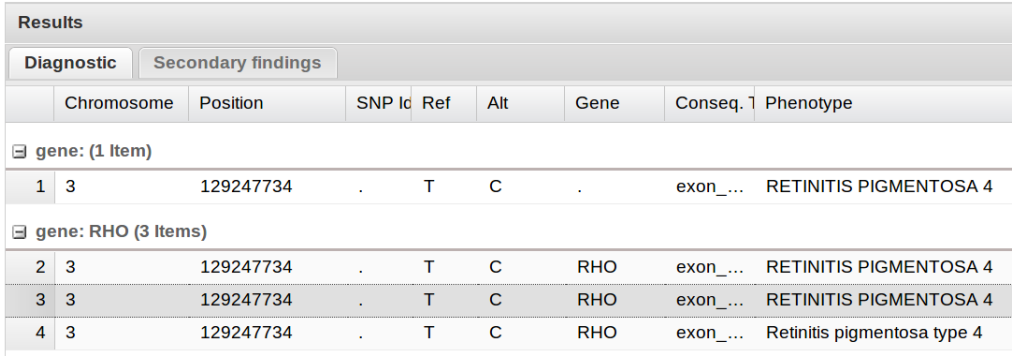
1. Defining panel



2. Uploading input data



3. Getting results



	Chromosome	Position	SNP Id	Ref	Alt	Gene	Conseq. T	Phenotype
gene: (1 Item)								
1	3	129247734	.	T	C	.	exon_...	RETINITIS PIGMENTOSA 4
gene: RHO (3 Items)								
2	3	129247734	.	T	C	RHO	exon_...	RETINITIS PIGMENTOSA 4
3	3	129247734	.	T	C	RHO	exon_...	RETINITIS PIGMENTOSA 4
4	3	129247734	.	T	C	RHO	exon_...	Retinitis pigmentosa type 4

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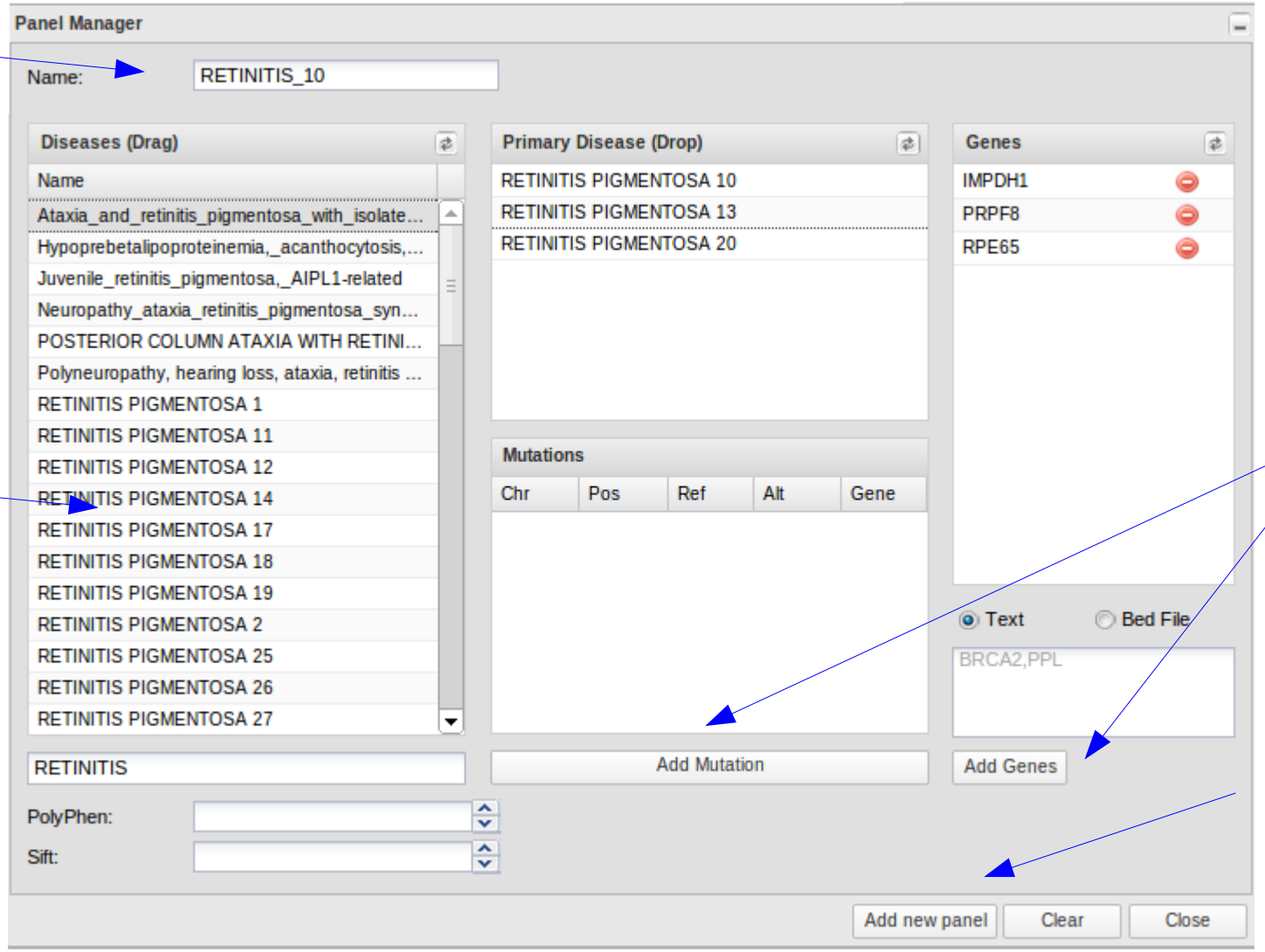
How to define a panel?

1. Name of panel

2. Diseases

3. Adding:
- more genes
- mutations

4. Save panel



How to define a panel?

Adding
new mutations

Checking
mutations from
Genome Viewer

Results

Results

Diagnostic | Secondary findings

	Chromosome	Position	SNP Id	Ref	Alt	Gene	Conseq. Type	Phenotype
gene: (1 Item)								
1	3	129247734	.	T	C	.	exon_vari...	RETINITIS PIGMENTOSA 4
gene: RHO (3 Items)								

Variant Effect - 3:129247734 T>C

	Position chr:start:end (strand)	SNP Id	Conseq. Type	Aminoacid Change
1	3:129247734-129247734 (+)	CM920608	SNP (SO:0000694)	.
2	3:129247483-129247937 (+)		synonymous_codon (SO:00...	P/P - CCC/CCC (53)
3	3:129245550-129248350		regulatory_region_variant (...	.
4	3:129247734-129247734 (+)	rs28933395	SNP (SO:0000694)	.

A. Web results

B. PDF report

Diagnostic: Retinitis

	Chromosome	Position	SNP Id	Ref	Alt	Gene	Con
1	3	129247734	.	T	C	.	exon ator
2	3	129247734	.	T	C	RHO	exon ator
3	3	129247734	.	T	C	RHO	exon ator
4	3	129247734	.	T	C	RHO	exon ator

Conclusions

- TEAM is an **free and easy-to-use web tool** that fills the gap between the enormous amounts of data in targeted enrichment sequencing analysis and the **biological knowledge** available.

- TEAM **provides an intuitive environment for the clinician** in which unprocessed data on patient’s genomic variation can easily be transformed in a **diagnostic**.

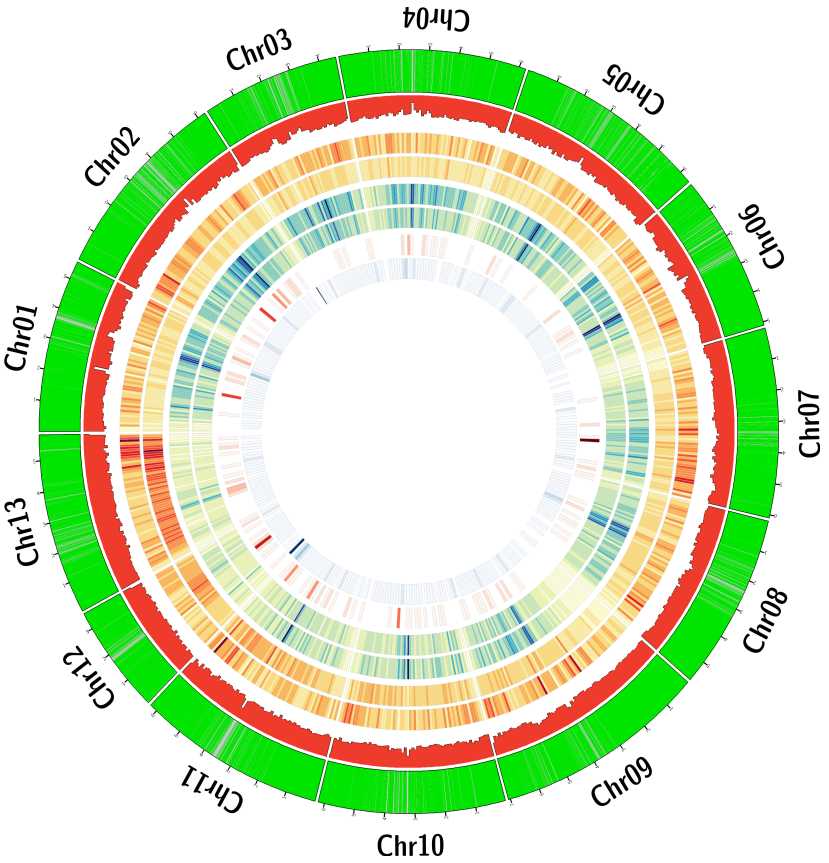
- The entire patient's sequencing information is managed locally thus avoiding any problem of data **privacy or confidentiality**.

Next improvements:

- Inclusion of a **database with public panels genes** of various diseases.

- **Comparative Analysis** for groups of panels.

- **Visualization results**.



More info + questions

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A web tool for the design and management of panels of genes for targeted enrichment and massive sequencing for clinical applications

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