

TEAM:

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



Computational • Genomics



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

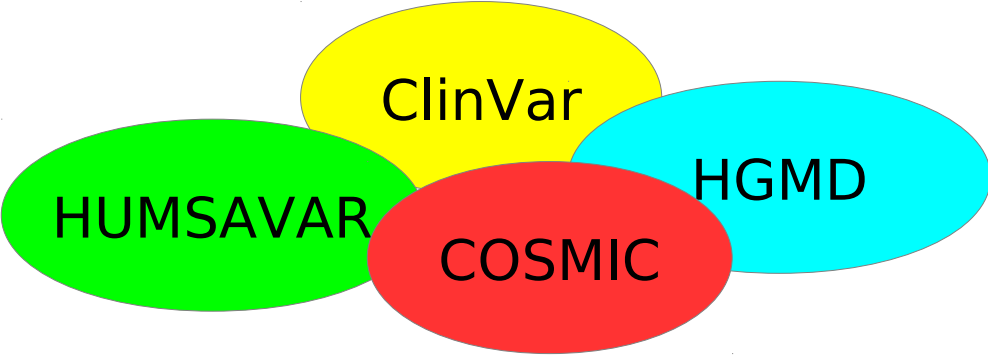
How does TEAM work?

- 1. VCF files
- 2. Gene panel

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Diagnostic: Retinitis

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



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How does TEAM work?

<http://team.babelomics.org/>

1. Defining panel

Panels

New Panel Import Panels Save Panels Clear Panels

User-defined Examples

name

RETINITIS_panel10

2. Uploading input data

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Example Data

Search

Panel: Panel Retinitis Pigmento: ▾

VCF File: C:\fakepath\patient1_R.vcf Browse...

Run Reset

3. Getting results

Results

Diagnostic Secondary findings

	Chromosome	Position	SNP Id	Ref	Alt	Gene	Conseq. 1	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

The screenshot shows the 'Panel Manager' window with the following components:

- Name:** A text input field containing 'RETINITIS_10'.
- Diseases (Drag):** A list of diseases including 'Ataxia_and_retinitis_pigmentosa_with_isolate...', 'Hypoprebetalipoproteinemia,_acanthocytosis,...', 'Juvenile_retinitis_pigmentosa,_AIPL1-related', 'Neuropathy_ataxia_retinitis_pigmentosa_syn...', 'POSTERIOR COLUMN ATAXIA WITH RETINI...', 'Polyneuropathy, hearing loss, ataxia, retinitis ...', 'RETINITIS PIGMENTOSA 1', 'RETINITIS PIGMENTOSA 11', 'RETINITIS PIGMENTOSA 12', 'RETINITIS PIGMENTOSA 14', 'RETINITIS PIGMENTOSA 17', 'RETINITIS PIGMENTOSA 18', 'RETINITIS PIGMENTOSA 19', 'RETINITIS PIGMENTOSA 2', 'RETINITIS PIGMENTOSA 25', 'RETINITIS PIGMENTOSA 26', and 'RETINITIS PIGMENTOSA 27'.
- Primary Disease (Drop):** A list containing 'RETINITIS PIGMENTOSA 10', 'RETINITIS PIGMENTOSA 13', and 'RETINITIS PIGMENTOSA 20'.
- Genes:** A list containing 'IMPDH1', 'PRPF8', and 'RPE65', each with a red minus sign to its right.
- Mutations:** A table with columns 'Chr', 'Pos', 'Ref', 'Alt', and 'Gene'. Below the table is a text input field containing 'BRCA2,PPL' and radio buttons for 'Text' (selected) and 'Bed File'.
- Buttons:** 'Add Mutation' and 'Add Genes' buttons are located below the mutations and genes sections, respectively.
- Footer:** 'RETINITIS' text, 'PolyPhen:' and 'Sift:' dropdown menus, and 'Add new panel', 'Clear', and 'Close' buttons.

2. Diseases

3. Adding:
- more genes
- mutations

4. Save panel

How to define a panel?

Adding new mutations

Checking mutations from Genome Viewer

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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								
3		Position chr:start:end (strand)	SNP Id				Conseq. Type	Aminoacid Change
4	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

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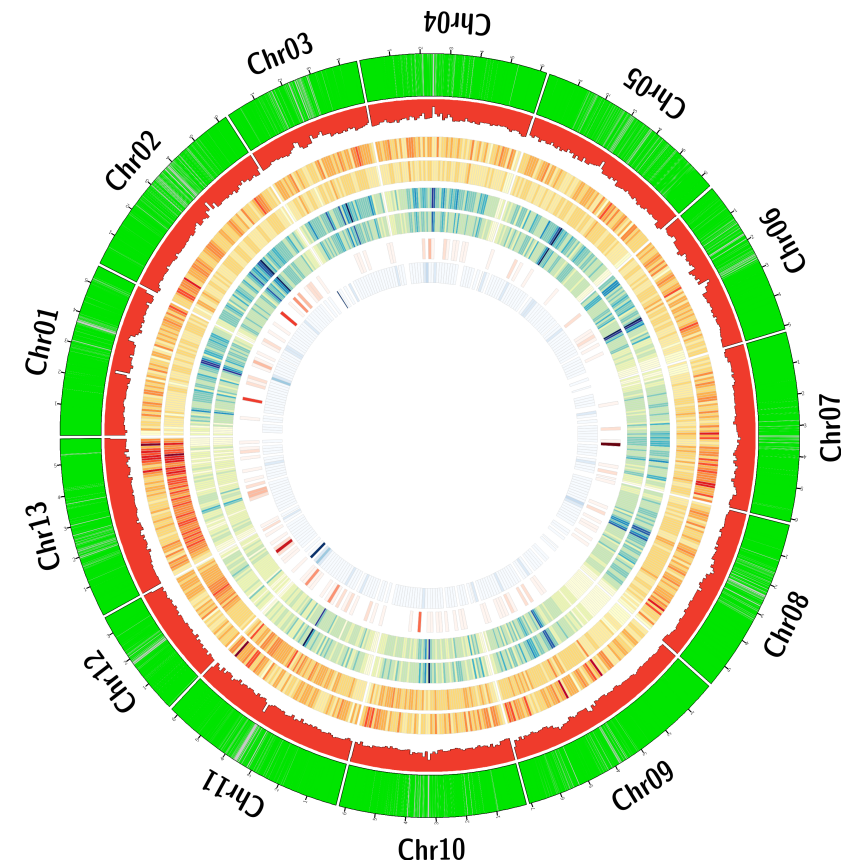
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Conclusions

- 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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