HUMAN GENETIC VARIATION VIEWER

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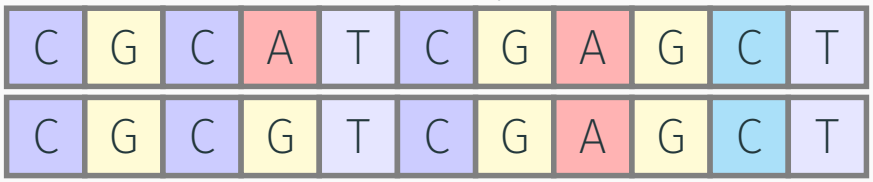

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OUTLINE

- Motivation
- Solution
- Demo and Use-Cases
- Implementation
- Future Work

MOTIVATION

VISUALIZATIONS ARE POWERFUL!

The power of the unaided mind is highly overrated. The real powers come from devising external aids that enhance cognitive abilities. – Donald Norman

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- Exploratory visualization is the first step towards discovering patterns
- Variation viewers are *absent*, if not, provide limited flexibility

SOLUTION

- A graphical hub to present annotated variants from different sources

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- Scalable, and Interactive exploration on the browser

Demo

http://saketkc.github.io/biojs

DETAILS

IMPLEMENTATION

- Entirely written in javascript using the *d3js* library

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- Events system that triggers events on user actions, allows cross-component communication

WHY BIOJS

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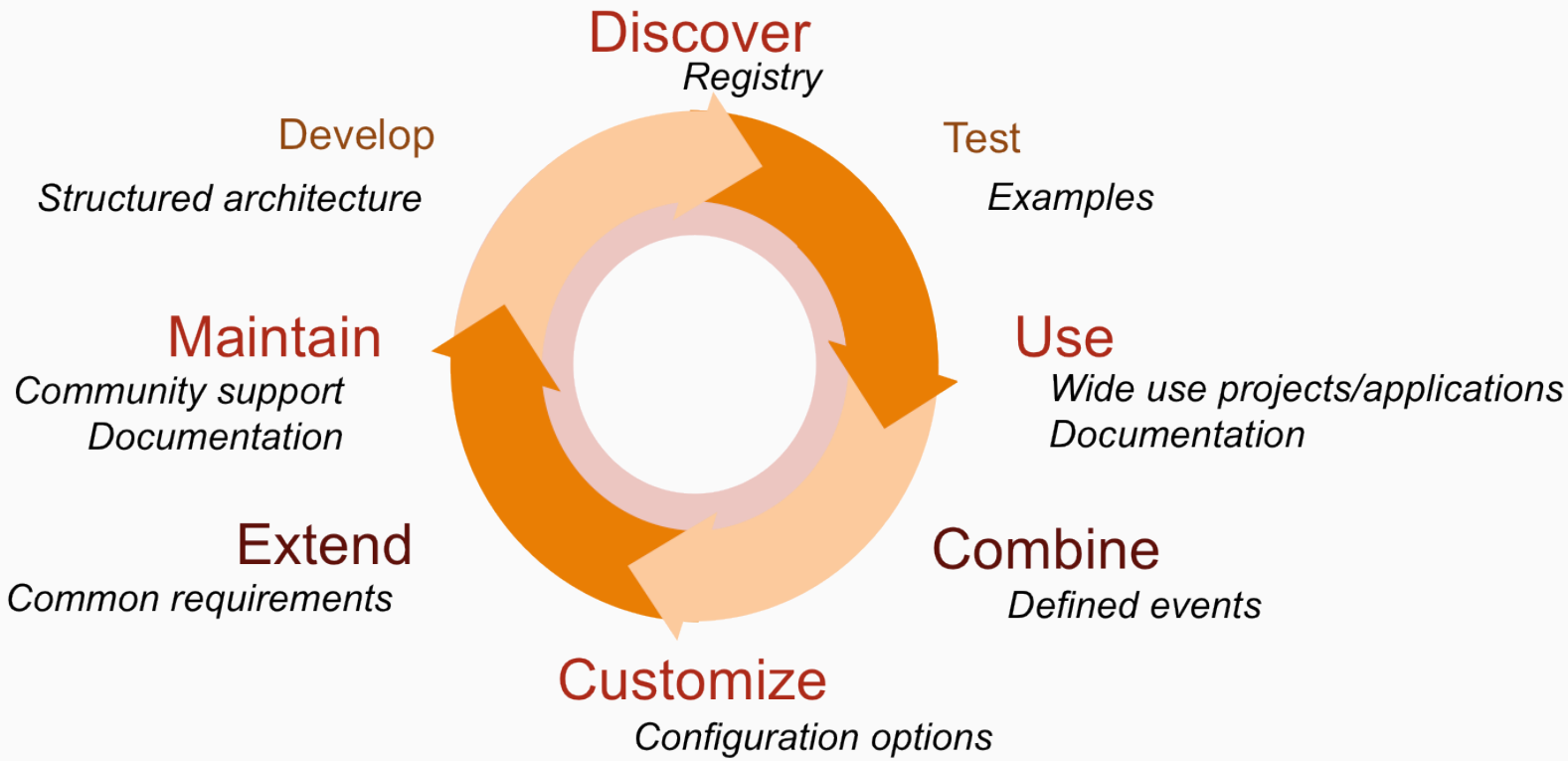
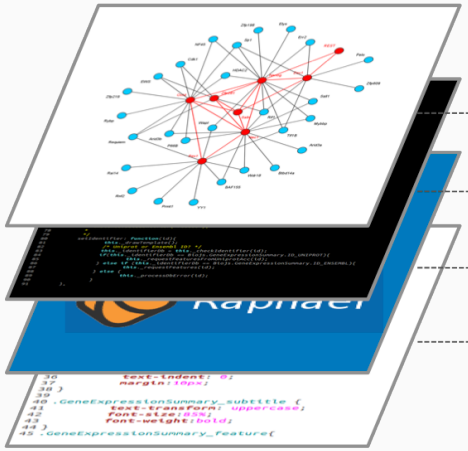


Reusable components that can talk to each other



Data representation

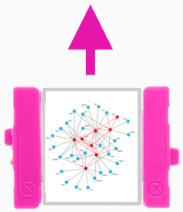
JavaScript code

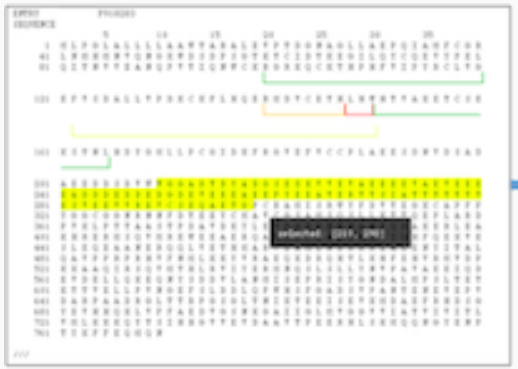
- Constructor
- **Options**
- **Methods**
- **Events**
- Comments
- Examples
- Dependencies
- Documentation

Dependencies

Style

Registry + API doc





onSelectionChanged

setSelection

```
{
"id":"P00533_variant226",
"sourceIds":["COSM1090877","COSM1090879"],
"position":541,
"wild_type":"L",
"mutation":"I",
"frequency":0.0,
"polyphenPrediction":"benign",
"polyphenScore":0.0,
"siftPrediction":"tolerated",
"siftScore":0.86,
"somaticStatus":1,
"consequenceTypes":"missense variant",
"cytogeneticBand":"7p11.2",
"genomicLocation":"7:g.55229314C>A"
}
```

DATA INPUT

- Pre-generated JSON files
- Current version uses files generated by an unpublished webservice at EBI
- Protein variants, though not specific to it

FEATURES

- Supports JSON formatted files, alpha VCF support
- User defined scoring criteria
- Different levels of information
  - Overview: Condensed information
  - Zoomed View: All annotations
- Loading proteins through URL parameters
- SIFT, Polyphen, ...

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- Benchmarking predictions

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- Performance improvements
- Interaction with 3D Protein viewer to highlight domains

CONCLUSION

SUMMARY

- A tool for visualizing genetic variants
- Supports visualization of different levels of information
- Cross component talks
- User defined and user controlled

ACKNOWLEDGEMENTS

Google, for running the Google Summer of Code 2014.

Questions?