HUMAN GENETIC VARIATION VIEWER

Saket Choudhary [1], Leyla Garcia[2] and Andrew Nightingale[2]

October 30, 2014

[1]University of Southern California and [2]EMBI-EBI

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

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

· NGS has given rise to catalog of genetic variants: dbSNP, COSMIC...

- NGS has given rise to catalog of genetic variants: dbSNP, COSMIC...
- Loads of data, but limited relevant information: Benign, Damaging, Intermediate

- NGS has given rise to catalog of genetic variants: dbSNP, COSMIC...
- Loads of data, but limited relevant information: Benign, Damaging, Intermediate
- Lots of mutations $\implies$ Loads of *differing* predictions

- NGS has given rise to catalog of genetic variants: dbSNP, COSMIC...
- Loads of data, but limited relevant information: Benign, Damaging, Intermediate
- Lots of mutations $\implies$ Loads of *differing* predictions
- Non consensus scoring mechanisms: SIFT and Polyphen predictions often disagree

- NGS has given rise to catalog of genetic variants: dbSNP, COSMIC...
- Loads of data, but limited relevant information: Benign, Damaging, Intermediate
- Lots of mutations $\implies$ Loads of *differing* predictions
- Non consensus scoring mechanisms: SIFT and Polyphen predictions often disagree
- Exploratory visualization is the first step towards discovering patterns

- NGS has given rise to catalog of genetic variants: dbSNP, COSMIC...
- Loads of data, but limited relevant information: Benign, Damaging, Intermediate
- Lots of mutations $\implies$ Loads of *differing* predictions
- Non consensus scoring mechanisms: SIFT and Polyphen predictions often disagree
- Exploratory visualization is the first step towards discovering patterns
- Variation viewers are *absent*, if not, provide limited flexibility

· A graphical hub to present annotated variants from different sources

- A graphical hub to present annotated variants from different sources
- Present information at different levels in a coherent manner

- A graphical hub to present annotated variants from different sources
- Present information at different levels in a coherent manner
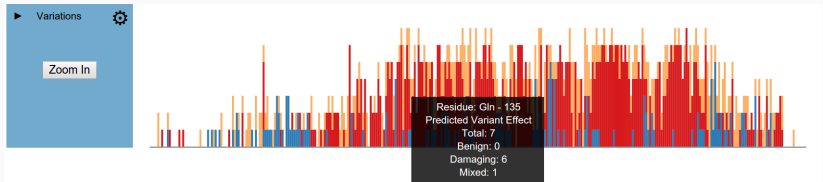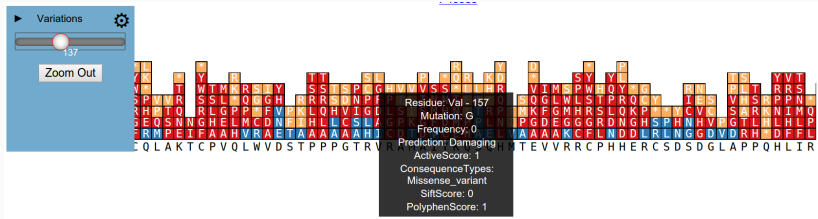- Scalable, and Interactive exploration on the browser

Figure: Overview

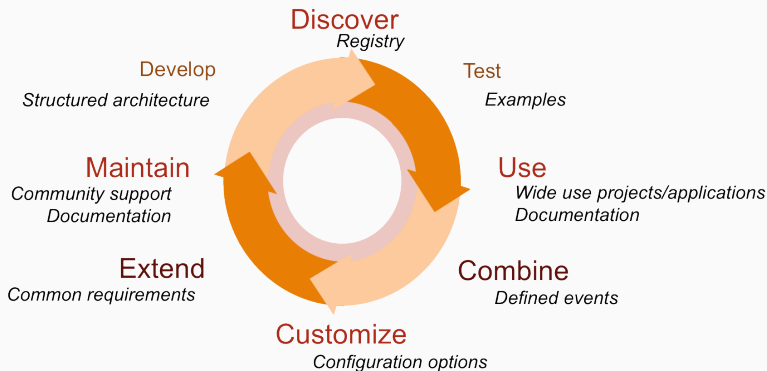Figure: Zoomed View

DEMO
http://saketkc.github.io/biojs

· Entirely written in javascript using the *d3js* library

· Entirely written in javascript using the *d3js* library
· Deployed as a BioJS component

- Entirely written in javascript using the *d3js* library
- Deployed as a BioJS component
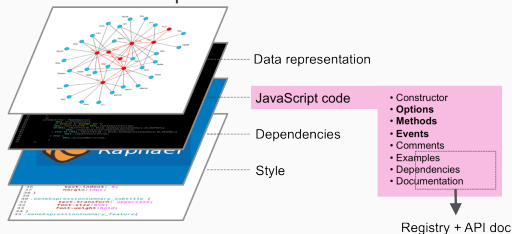- Events system that triggers events on user actions, allows cross-component communication

- BioJS is a javascript library for developing visualization of the biological data

- BioJS is a javascript library for developing visualization of the biological data

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

12

```
{
"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"
}
```

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

- Supports JSON formatted files, alpha VCF support

- Supports JSON formatted files, alpha VCF support
- User defined scoring criteria

- Supports JSON formatted files, alpha VCF support
- User defined scoring criteria
- Different levels of information, tooltips

- Supports JSON formatted files, alpha VCF support
- User defined scoring criteria
- Different levels of information, tooltips
  - Overview: Condensed information

- Supports JSON formatted files, alpha VCF support
- User defined scoring criteria
- Different levels of information, tooltips
  - Overview: Condensed information
  - Zoomed View: All annotations

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

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

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

· Identifying most or least mutated sites across proteins

- Identifying most or least mutated sites across proteins
- Discover differences between different scoring criteria

· Identifying most or least mutated sites across proteins
· Discover differences between different scoring criteria
· Benchmarking predictions

- VCF support(almost there)

- VCF support(almost there)
- Integration with Galaxy, web based bioinformatics workflows

- VCF support(almost there)
- Integration with Galaxy, web based bioinformatics workflows
- Performance improvements

· VCF support(almost there)
· Integration with Galaxy, web based bioinformatics workflows
· Performance improvements
· Interaction with 3D Protein viewer to highlight domains