HUMAN GENETIC VARIATION VIEWER

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- Motivation
- Solution
- Demo and Use-Cases
- Implementation
- Improvements

MOTIVATION

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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- Variation viewers are *absent*, if not, provide limited flexibility

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- Scalable, and Interactive exploration on the browser
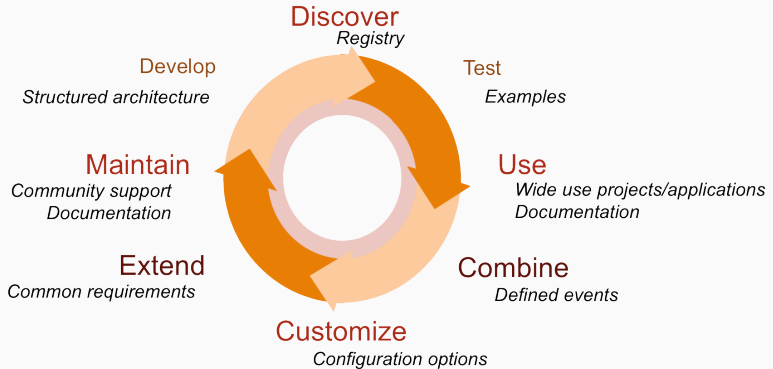
DEMO
http://saketkc.github.io/biojs

DETAILS

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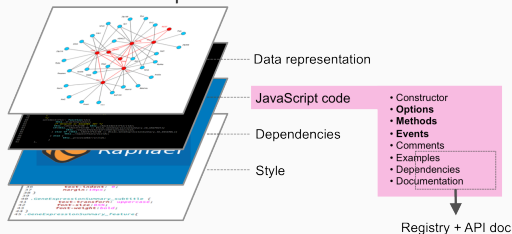· Events system that triggers events on user actions, allows cross-component communication

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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

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```json
{
"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"
}
```

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- 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- Current version uses files generated by an unpublished webservice at EBI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
- 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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- Interaction with 3D Protein viewer to highlight domains

CONCLUSION

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

QUESTIONS?