

# HUMAN GENETIC VARIATION VIEWER

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

# MOTIVATION

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*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

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- Scalable, and Interactive exploration on the browser

DEMO

## DETAILS

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- Entirely written in javascript using the *d3js* library

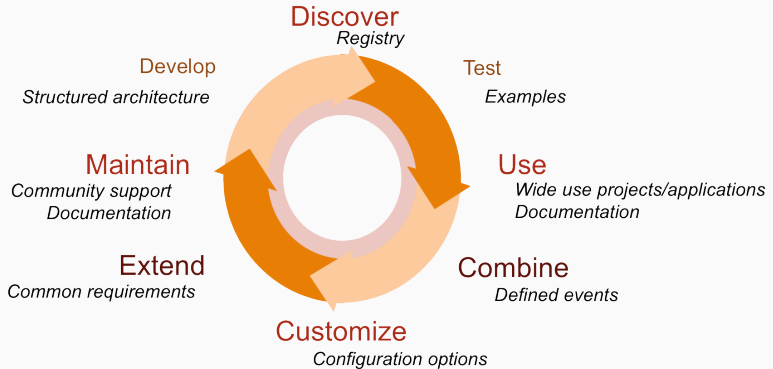


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- Deployed as a BioJS component
- Events system that triggers events on user actions, allows cross-component communication

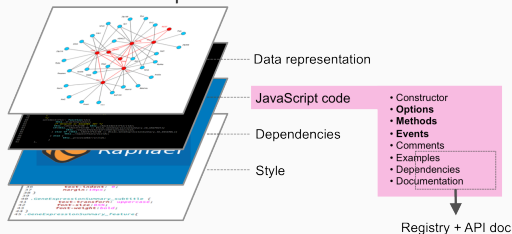
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# WHY BIOJS

Reusable components that can talk to each other



```
{  
  "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- Current version uses files generated by an unpublished webservice at EBI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
- User defined scoring criteria
- Different levels of information
  - Overview: Condensed information
  - Zoomed View: All annotations
- Loading proteins through URL parameters
- SIFT, Polyphen, ....



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

- VCF support, almost there

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- Integration with Galaxy, web based bioinformatics workflows

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- Performance improvements

## CONCLUSION

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

Google, for running the Google Summer of Code 2014.

QUESTIONS?