

HUMAN GENETIC VARIATION VIEWER

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

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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- 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

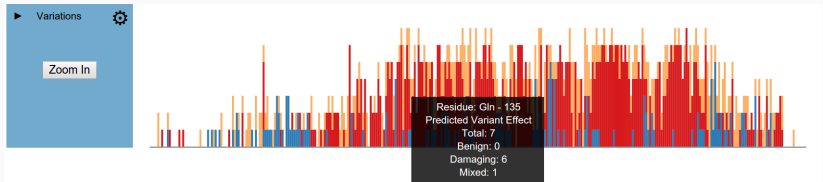


Figure: Overview

ZOOMED VIEW

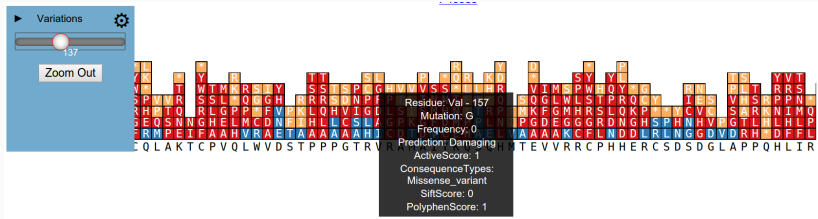


Figure: Zoomed View

DEMO

<http://saketkc.github.io/biojs>

DETAILS

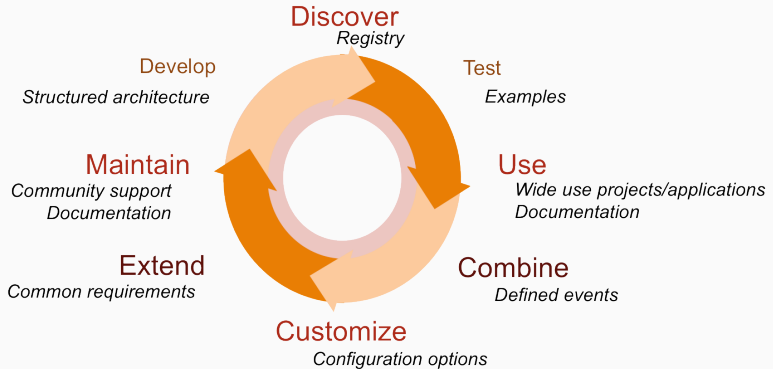
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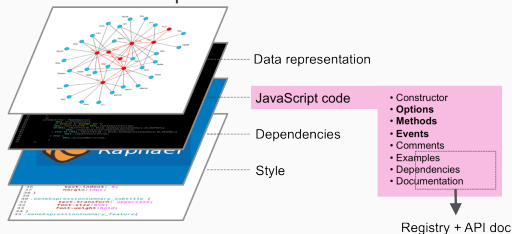
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WHY BIOJS

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- Current version uses files generated by an unpublished webservice at EBI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

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- Scalable, adaptable to new scores, mutation categories

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- Benchmarking predictions

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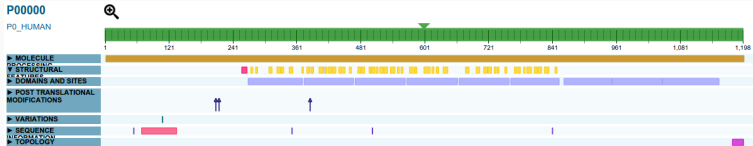
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- Interaction with 3D Protein viewer to highlight domains

CONCLUSION

SUMMARY

- A tool for visualizing genetic variants
- Limited applications as a standalone tool, more usable with Protein Features Viewer



- Supports visualization of different levels of information
- Cross component talks
- User defined and user controlled
- Open Sourced(MIT License): <https://github.com/saketkc/biojs-genetic-variation-viewer>

Google | Google Summer of Code 2014
BioJS Community
USC MCB

QUESTIONS?