

# Visualizing consequences of genetic variation in biological networks

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## Summary

The Variation app for Cytoscape 3 establishes a genomic location for genes on nodes in a biological network, associates variations with those genes from a variety of local or remote resources, and then sources or predicts consequences of those variations for further analysis and visual mapping.

## Features

- Feature view
- Variation view
- Variation consequence view
- Visual mappings
- Discrete color mapping for Sequence Ontology (SO)-annotated consequence terms

## Resources

- Ensembl REST APIs
- SnpEff-annotated VCF files
- Ensembl VEP-annotated VCF files
- Global Alliance for Genomics and Health APIs
- Google Genomics APIs

## Google Summer of Code (GSoC) 2014

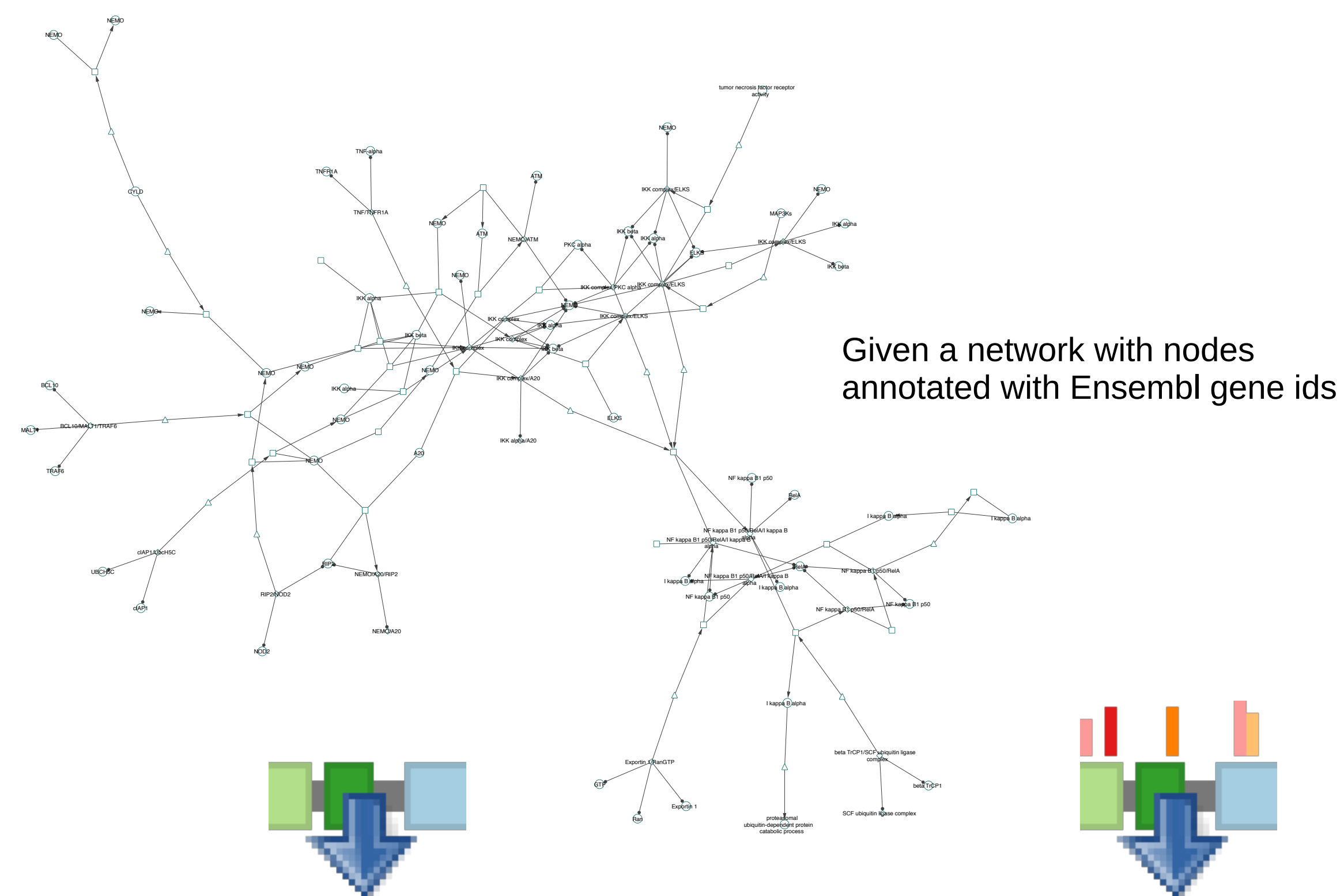
Under the 2014 National Resource for Network Biology (NRNB) GSoC program:

- Support for ADAM genome analysis system
- Additional compound visual mappings

## Availability

The Variation app for Cytoscape 3 is licensed GNU Lesser General Public License (LGPL), version 3 or later.

Versions 1.0 and later are available on the Cytoscape 3 App Store at <http://apps.cytoscape.org/apps/variation>.

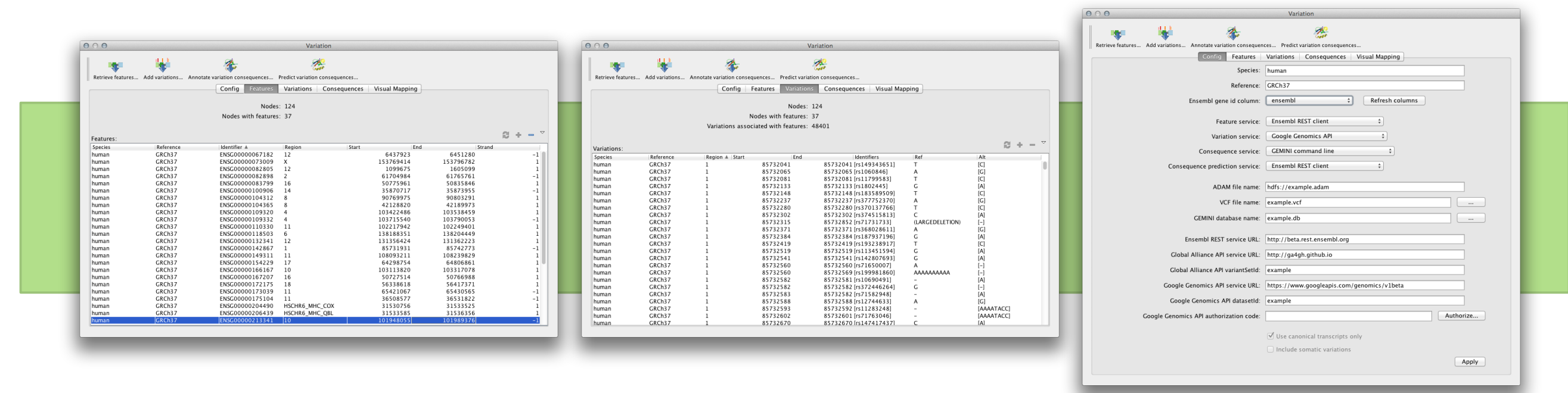


### Retrieve features

- All genomic features associated with the nodes in the current network are displayed in the Feature view

### Add variations

- All variations associated with the genomic features associated with the nodes in the current network are displayed in the Variation view
- A new column 'variation\_count' is added to the node table with the aggregate count of variations per node

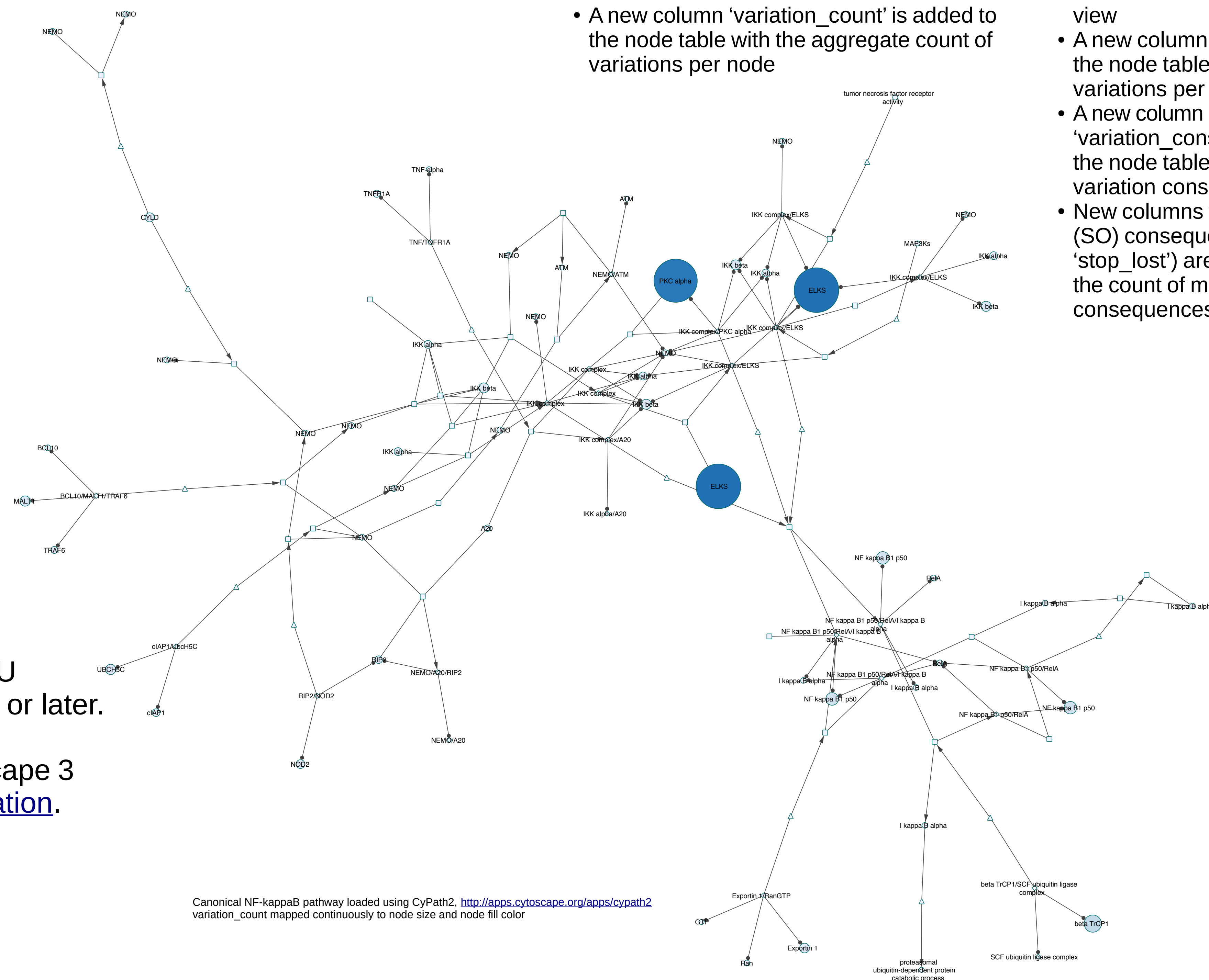


### Annotate variation consequences

- All variation consequences for all variations associated with the genomic features associated with the nodes in the current network are displayed in the Consequence view
- A new column 'variation\_count' is added to the node table with the aggregate count of variations per node
- A new column 'variation\_consequence\_count' is added to the node table with the aggregate count of variation consequences per node
- New columns for each Sequence Ontology (SO) consequence term (e.g. 'stop\_gained', 'stop\_lost') are added to the node table with the count of matching variation consequences per node

### Predict variation consequences

- All newly predicted variation consequences for all variations associated with the genomic features associated with the nodes in the current network are displayed in the Consequence view
- A new column 'variation\_count' is added to the node table with the aggregate count of variations per node
- A new column 'variation\_consequence\_count' is added to the node table with the aggregate count of newly predicted variation consequences per node
- New columns for each Sequence Ontology (SO) consequence term (e.g. 'stop\_gained', 'stop\_lost') are added to the node table with the count of matching newly predicted variation consequences per node



Canonical NF-kappaB pathway loaded using CyPath2, <http://apps.cytoscape.org/apps/cyath2>  
variation\_count mapped continuously to node size and node fill color

Species	Reference	Region	Start	End	Identifiers	Ref	Alt	Consequence A
human	GRCh37	1	85735998	85735999	CG	T	C	intron_variant
human	GRCh37	1	85735981	85735982	CG	G	A	intron_variant
human	GRCh37	1	85735991	85735992	CG	G	A	intron_variant
human	GRCh37	1	85735999	85736000	CG	G	C	intron_variant
human	GRCh37	1	85736021	85736022	CG	C	T	intron_variant
human	GRCh37	1	85736045	85736046	CG	C	T	intron_variant
human	GRCh37	1	85736062	85736063	CG	C	T	intron_variant
human	GRCh37	1	85736064	85736065	CG	C	T	intron_variant
human	GRCh37	1	85736071	85736072	CG	C	T	intron_variant
human	GRCh37	1	85736087	85736088	CG	T	G	intron_variant
human	GRCh37	1	85736097	85736098	CG	G	C	intron_variant
human	GRCh37	1	85736138	85736139	CG	A	G	intron_variant
human	GRCh37	1	85736152	85736153	CG	G	A	intron_variant
human	GRCh37	1	85736211	85736212	CG	C	T	intron_variant
human	GRCh37	1	85736216	85736217	CG	A	G	intron_variant
human	GRCh37	1	85736291	85736292	CG	A	G	intron_variant
human	GRCh37	1	85733316	85733317	CG	C	T	missense_variant
human	GRCh37	1	85733365	85733366	CG	C	T	missense_variant
human	GRCh37	1	85733378	85733379	CG	C	T	missense_variant
human	GRCh37	1	85733443	85733444	CG	T	C	missense_variant
human	GRCh37	1	85733526	85733527	CG	G	A	missense_variant
human	GRCh37	1	85733530	85733531	CG	T	C	missense_variant
human	GRCh37	1	85733490	85733491	CG	A	T	stop_gained
human	GRCh37	1	85733522	85733523	CG	C	T	synonymous_variant
human	GRCh37	1	85733525	85733526	CG	C	T	synonymous_variant