Variant Analysis™

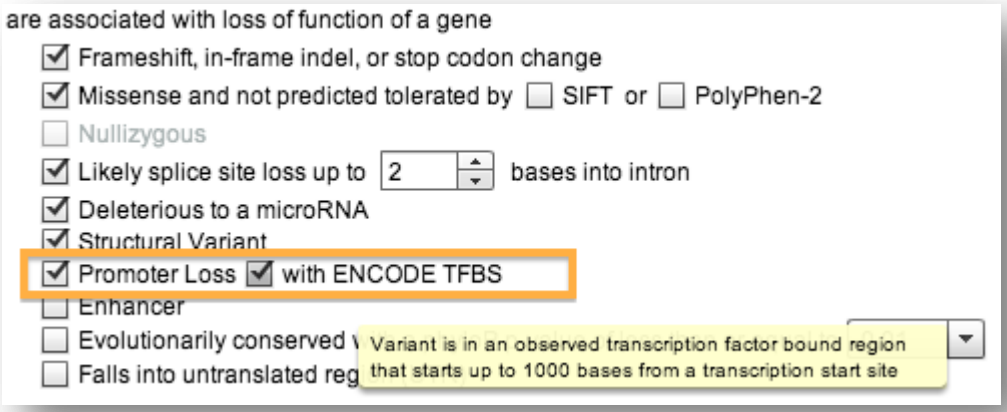
Feature Highlight

ENCODE content

Ingenuity® Variant Analysis™ incorporates content from a number of sources, now including ENCODE. With this addition, researchers can now filter for variants that fall within ENCODE transcription factor binding sites, providing enhanced capabilities to identify biologically relevant variants.

ENCODE, which stands for the Encyclopedia Of DNA Elements[i], is a public research consortium that was launched by the National Human Genome Research Institute (NHGRI) in September 2003 to identify all functional elements in the human genome. After an initial pilot phase, ENCODE scientists started applying their methods in a production phase to the entire genome in 2007. In September 2012, that led to the publication of 30 papers in leading journals. Scientists continue their efforts to complete the mapping, and ENCODE is committed to rapidly releasing data into the public domain.

Now researchers working with Ingenuity Variant Analysis can access ENCODE findings directly within the application through the "Predicted Deleterious Filter," as shown below.



Users of Variant Analysis can also see ENCODE annotations in the "Regulatory Site" and "Regulator" columns of the "Variants" details view, as shown.

Regulatory Site	Regulator
ENCODE TFBS, Promoter Loss	CEBPB, CTCF, ELF1, F
Promoter Loss	GATA3
Enhancer Binding Site, Enhancer TFBS Loss	Pax2

This content enhancement in Variant Analysis reflects Ingenuity's commitment to providing a world-class Knowledge Base. The Ingenuity Knowledge Base is supported by a robust set of people, processes, and technology for curating high-quality scientific relationships from peer-reviewed journals and both public and private biomedical databases.

For any additional questions, please contact Ingenuity support at support@ingenuity.com
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[i] http://www.genome.gov/10005107

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