
Title	Building a community menagerie of automated variant validations
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License	MIT

bcbio (<https://github.com/chapmanb/bcbio-nextgen>) is an open, community effort to develop validated and scalable variant calling, RNA-seq, ChIP-seq and small RNA analyses. bcbio runs across a wide variety of platforms from full stack cloud providers to local high performance computing environments by leveraging Common Workflow Language (CWL: <http://www.commonwl.org/>) and the GA4GH interoperability standards (<https://github.com/ga4gh/wiki/wiki>),

bcbio integrates production ready analysis pipelines with automated validations using reference materials developed by communities like Genome in a Bottle (<http://jimb.stanford.edu/giab/>) and the International Cancer Genome Consortium (<https://icgc.org/>). This creates a multi-platform test suite for method comparisons across a wide variety of biological analyses, as well as a baseline for adjusting and improving existing methods.

We'll highlight four useful validations from our collection of workflows:

- GATK4: an evaluation of the new open source GATK4 variant caller against GATK3 and other recent methods like strelka2. https://github.com/bcbio/bcbio_validations/tree/master/gatk4
- DeepVariant and CHM reference materials: compares Google's Neural Network based caller against other standard methods, including comparisons on an orthogonal haploid/diploid based truth set to evaluate training bias. https://github.com/bcbio/bcbio_validations/tree/master/deepvariant
- Value of trimming in low frequency somatic variant detection: explores the impact of 3' quality and low complexity trimming on runtime and quality, helping remove bottlenecks in whole genome variant calling. https://github.com/bcbio/bcbio_validations/tree/master/somatic_trim
- Structural variant detection sensitivity for long and short reads: identifies limits of detection for short read methods based on comparisons with resolvable large scale events in long reads. https://github.com/bcbio/bcbio_validations/tree/master/NA24385_sv

The goal of this work is to coordinate with the bioinformatics community to build automated variant test suites that run on any platform of choice. Regular automated builds: ensure that tools are always functional across platforms, create a historical account of method performance, evaluate new methods, and give researchers a robust and validated way to run variant analyses to answer difficult biological questions.