

In this tutorial, we collect data from normal and tumor tissues to discover what spectrum of mutations distinguishes this tumor from healthy tissue. Our goal was to report the variant sites, and the genes affected by them, annotated with the content of general human genetic and cancer-specific databases.

The Tumor/Normal data analysis workflow, consisting of Alignment and VarScan Somatic variant calling, demonstrates excellent performance for the detection of somatic variants. Somatic variant calling not only calls variants but also distinguishes Cancer-specific variants (Somatic mutations) found only in tumor tissues from germline mutations that are shared by tumor and healthy tissue, and loss-of-heterozygosity events; that is, the absence of one of two alleles found at a biallelic site of healthy tissues, in tumor tissues. This makes it a more optimal approach for applications requiring high precision such as novel mutation detection and mutation signature analysis.

The interpretation of any list of variants (somatic, germline or LOH) almost always depends crucially on rich genetic and cancer-specific variants and gene annotations which can be inferred from the analysis.

Ideally, this may provide insight into the genetic events driving tumor formation and growth in the patient, and might be of prognostic and even therapeutic value by revealing variants known to affect drug resistance/sensitivity, tumor aggressiveness, etc.