

The screenshot displays the PhenGenVar Browser interface with the following components and annotations:

- (1)** Points to the **File** menu in the top-left corner.
- (2)** Points to the **Reference** dropdown menu, which is set to **Human hg19**.
- (3)** Points to the **dbSNP Version** dropdown menu, which is set to **dbSNP150**.
- (4)** Points to the **Gene Filter** section on the left, which includes options like **View only genes with variations** and **Allele Frequency**.
- (5)** Points to the **Gene/Transcript View** area, which displays a list of genes and transcripts. The **Gene** dropdown is set to **ATR**.
- (6)** Points to the **Exon View Panel** on the right, which shows a detailed view of the selected variant (rs11201840) and its surrounding genomic context.

### Supplementary Figure S1. Simple steps to use PhenGenVar Browser.

(1) Load a BAM file and a VCF file containing variant calling results (optional) by selecting them using the File menu. Note: select only BAM and VCF files that have index files (\*.bai/\*.idx). (2) Select a reference sequence version and a dbSNP version (optional) which are necessary for BAM/VCF file analysis. (3) Select a Gene\_Group or Phenotype to analyze. The related gene sets are displayed in the Gene/Transcript View area. (4) Double click a random gene output in the Gene/Transcript View area to obtain the transcript data of the gene in a child node. If a random transcript is double-clicked, the variant information areas are activated to provide the variant list identified on the transcript of interest. (5) Select and click a random variant to display an exon area including the position of the selected variant in the Main Exon View Panel. (6) Select a variant and double click with a mouse to call a Genome Browser centering at the position of the related variant and use it for a detailed analysis of that area.