

The screenshot displays the PhenGenVar Browser interface. At the top, the 'File' menu is highlighted (1), and the 'Reference' dropdown is set to 'Human hg19' (2). The 'dbSNP Version' is set to 'dbSNP150' (3). The 'Phenotype / Gene Group' is set to '11 pairs of ribs'. A table of VCF entries is shown, with the first row selected (4). The table has columns: Exon, Chrom, POS, ID, REF, ALT, QUAL, FILTER, Strand, ref#CBI, ref#CSC, Observ..., Function, MolTy, Class, allele, allele..., INFO, FORM., and Sequ... The selected row is: 5, 3, 142,168,331, rs1229204, C, T, 305.77, ., -, C, C, A, A, C, coding, gene, single, A,G, 0.0555, ABSO, GT,A, T/T,2. Below the table, a list of genes is shown, with 'NM_001184' selected (4). A detailed view of the transcript 'NM_001184' is shown, including the exon structure and a variant call (5). A 'Genome Browser' window is open, showing the genomic context of the variant (6).

Exon	Chrom	POS	ID	REF	ALT	QUAL	FILTER	Strand	ref#CBI	ref#CSC	Observ...	Function	MolTy	Class	allele	allele...	INFO	FORM.	Sequ...
5	3	142,168,331	rs1229204	C	T	305.77	.	-	C	C	A	coding	gene	single	A,G	0.0555	ABSO	GT,A	T/T,2
14	3	142,215,178	rs7820648	A	C	489.77	.	.	A	A	A								
14	3	142,215,233	rs112018640	G	A	948.77	.	.	G	G	A								
14	3	142,215,305	rs797045404	AAAG	A	1027.73	.	.	A	A	A								
14	3	142,215,376	rs587783335	C	A	325.77	.	.	C	C	A								
33	3	142,288,372	rs28910272	C	T	379.77	.	-	C	C	A								

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.