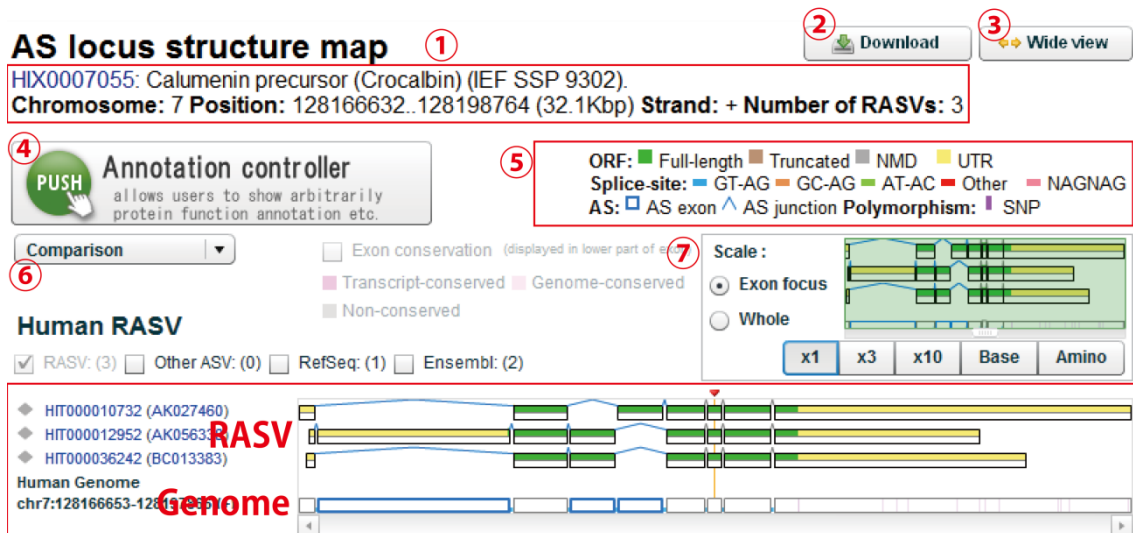


Operating manual of AS locus structure map in H-DBAS

1. Whole explanation



⑧ Main view

① Information of locus

There is locus information of H-InvDB. HIX ID is linked to the locus of H-InvDB.

② Download (human locus only)

UCSC refFlat format file of representative AS variants (RASVs) is offered.

③ Wide view

This is a button to adjust the viewer size in accordance with the screen size of browser.

④ Annotation controller (human locus only)

This is a gadget to display annotation information like protein motif on Main and Scale views (**Details are given in 2. Annotation controller**).

⑤ Explanation of color

Simple explanation of colors used in Main and Scale views.

⑥ Comparison (human locus only)

When other species locus corresponded to human one by comparative genomics exists, selected the other species locus is shown under the Main view (**Details are given in 3. Comparison**) .

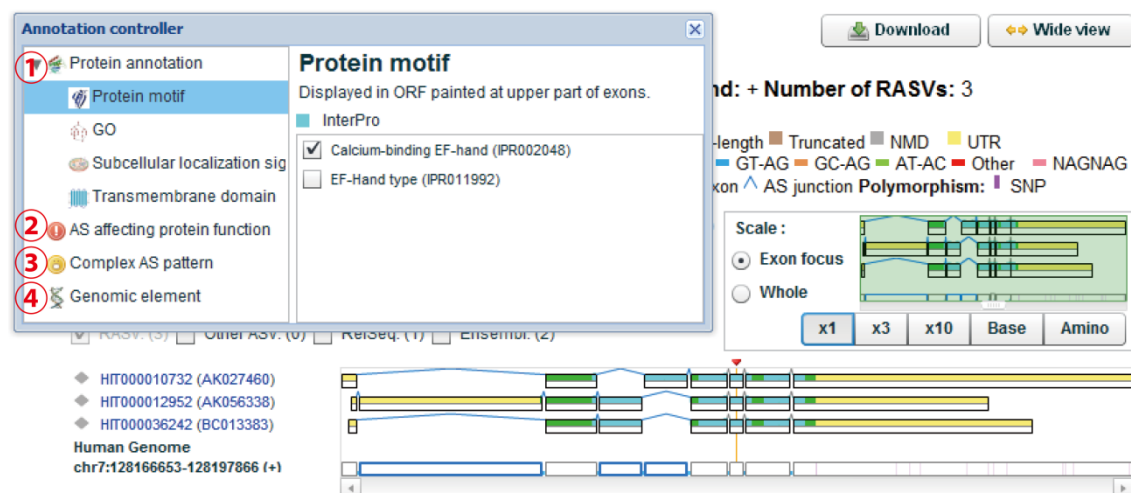
⑦ Scale view

This is a gadget to select the display form of RASVs and display the zoom of RASVs and genome on Main view (**Details are given in 4. Scale view**) .

⑧ Main view

RASVs and genome are displayed. The operation results of Annotation controller, Comparison and Scale view are displayed. By checking the checkboxes in the superior region, AS variants except for RASVs (Other ASVs), RefSeq and Ensembl transcripts are displayed (**Details are given in 5. Main view**) .

2. Annotation controller



① Protein annotation

Protein motif and transmembrane domain are displayed in the ORF of RASV with different from ORF color by checking the checkbox.

GO and Subcellular localization signal are displayed as the highlighted background of RASV by checking the checkbox.

② AS affecting protein function

RASV pair which affect above mentioned four protein annotations are displayed as the

highlighted backgrounds of them. After selecting an annotation item, RASV pair can be selected from the underneath list. The display are cleared by clicking Clear button.

③ Complex AS pattern

RASV pair of complex AS pattern are displayed as the highlighted backgrounds of them. After selecting a pattern item, RASV pair can be selected from the underneath list. The display are cleared by clicking Clear button.

④ Genomic element

ESE, ESS and retrotransposon are displayed in the lower part of exons of RASVs. Fox and Nova are displayed in the genome within 300bp from exon terminals.

3. Comparison

AS locus structure map Download Wide view

HIX0007055: Calumenin precursor (Crocabin) (IEF SSP 9302).
Chromosome: 7 Position: 128166632..128198764 (32.1Kbp) Strand: + Number of RASVs: 3

Annotation controller
 PUSH allows users to show arbitrarily protein function annotation etc.

ORF: ■ Full-length ■ Truncated ■ NMD ■ UTR
 Splice-site: ■ GT-AG ■ GC-AG ■ AT-AC ■ Other ■ NAGNAG
 AS: ■ AS exon ▲ AS junction Polymorphism: ■ SNP

Mouse: 06-0000255 ①

ESV cluster: 2 (There is Conserved AS)

Human RASV

☒ RASV: (3) ☐ Other ASV: (0) ☐ RefSeq: (1) ☐ Ensembl: (2)

☒ Exon conservation (displayed in lower part of exon)
☐ Transcript-conserved ☐ Genome-conserved
☐ Non-conserved ②

Scale: ☒ Exon focus ☐ Whole
 x1 x3 x10 Base Amino

③

HIT000010732 (AK027460) ESV 1
 HIT000012952 (AK056338)
 HIT000036242 (BC013383) ESV 2
 Human Genome
 chr7:128166653-128197866 (+)
 chr6:29298154-29326675 (+)
 Mouse Genome
 AK147756 ESV 1
 AK148178 ESV 2
 AK161807

HIT000010732 (7/7)
 chr7:128196353-128197866(+)
 Genome alignment sequence ④

☐ RASV: (3) ☐ Other ASV: (4) ☐ RefSeq: (2) ☐ Ensembl: (2) ⑤

Mouse RASV

① Selection of other species (and the locus ID)

Selected other species locus is displayed under the Main view. By selecting Default, the display is cleared. In a state that other species is selected, the function of Base and Amino in Scale view is unavailable.

② Exon conservation

When the other species locus is displayed, the checkbox of Exon conservation is available. By checking it, the lower part of human exons are color-coded according to Transcript-, Genome- and Non-conserved.

③ Display of ESV

When the equally-spliced variants (ESVs) among interspecies exist, the word "ESV" is displayed in the right side of transcript ID. The backgrounds of their transcript ID are highlighted by same color and they are also linked by the same color line. When two or more ESVs which have different ESV number exist, the word "ESV" is colored in red and the sentence "There is Conserved AS" is displayed also in red.

④ Exon pop-up

In a state that the other species locus is displayed, by clicking the exon of human RASV in Main view, the link to Genome alignment sequence page exits in the appeared pop-up if the exon is mapped in the genome alignment.

⑤ Display of other species reference sequences

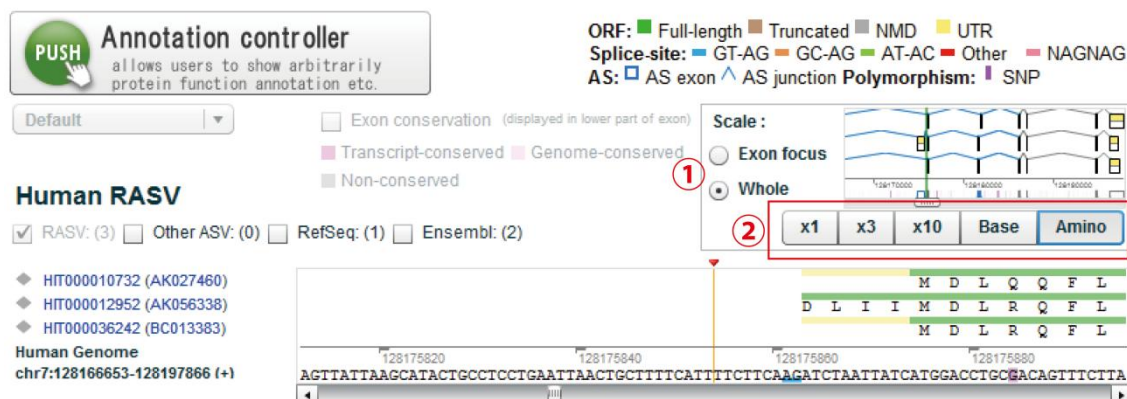
By checking the checkboxes, AS variants except for RASVs (Other ASVs), RefSeq and Ensembl transcripts of other species are displayed as reference sequences.

4. Scale view

AS locus structure map

HIX0007055: Calumenin precursor (Crocabin) (IEF SSP 9302).

Chromosome: 7 Position: 128166632..128198764 (32.1Kbp) Strand: + Number of RASVs: 3



① Selection of display form of RASV

Default is Exon focus which shows RASVs without constitutive introns. By changing to Whole, real mapping results of RASVs with all introns are shown.

② Zoom

There are x1 (default locus size), x3, x10, Base and Amino. By clicking Base, base sequences of RASV and genome are displayed. By clicking Amino, amino acid sequences which change codon of RASV into single character code of amino acid are displayed. In a state that the other species locus is displayed, the function of Base and Amino is unavailable.

5. Main view

AS locus structure map Download Wide view

HIX0007055: Calumenin precursor (Crocabin) (IEF SSP 9302).
Chromosome: 7 Position: 128166632..128198764 (32.1Kbp) Strand: + Number of RASVs: 3

Annotation controller
 allows users to show arbitrarily protein function annotation etc.

Default ☐ Exon conservation (displayed in lower part of exon) ☐ Transcript-conserved ☐ Genome-conserved ☐ Non-conserved

Human RASV ①

☒ RASV: (3) ☐ Other ASV: (0) ☒ RefSeq: (1) ☒ Ensembl: (2) ③

②

HIT000010732 (AK027460)
 HIT000012952 (AK056338)
 HIT000036242 (BC013383)
 NM_001219
 ENST00000249364
 ENST00000342367

Human Genome
 chr7:128166653-128198764 (+) ④

ORF: ☒ Full-length ☐ Truncated ☐ NMD ☐ UTR
 Splice-site: ☒ GT-AG ☐ GC-AG ☐ AT-AC ☐ Other ☐ NAGNAG
 AS: ☒ AS exon ☐ AS junction Polymorphism: ☐ SNP

Scale: ☒ Exon focus ☐ Whole

x1 x3 x10 Base Amino

GATGAGTTCTGAGCTACGGAGGAACCCCTCATTTCCTCAAAGTAATTTATTTTACAGCTTCTGGTTTCACATGAAATTGTTG
 GATGAGTTCTGAGCTACGGAGGAACCCCTCATTTCCTCAAAGTAATTTATTTTACAGCTTCTGGTTTCACATGAAATTGTTG
 GATGAGTTCTGAGCTACGGAGGAACCCCTCATTTCCTCAAAGTAATTTATTTTACAGCTTCTGGTTTCACATGAAATTGTTG
 GATGAGTTCTGAGCTACGGAGGAACCCCTCATTTCCTCAAAGTAATTTATTTTACAGCTTCTGGTTTCACATGAAATTGTTG
 GATGAGTTCTGAGCTACGGAGGAACCCCTCATTTCCTCAAAGTAATTTATTTTACAGCTTCTGGTTTCACATGAAATTGTTG
 GATGAGTTCTGAGCTACGGAGGAACCCCTCATTTCCTCAAAGTAATTTATTTTACAGCTTCTGGTTTCACATGAAATTGTTG

SNP
 A/G (rs1043550)

① Display of reference sequences

By clicking the checkbox, AS variants except for RASVs (Other ASVs), RefSeq and Ensembl transcripts are displayed as reference sequences.

② Transcript ID

HIT ID is linked to H-InvDB, accession number is linked to DDBJ, RefSeq and Ensembl are linked to each pages.

③ Display of transcripts

The structure and annotation information of RASVs and reference sequences are

displayed. By clicking the exon, a pop-up that AS pattern is described is appeared.

④ Display of genome

In human genome, SNPs can be observed. In a state that the genome is base sequences, a pop-up is appeared by clicking a SNP and it is possible to jump to dbSNP from it.