

Visualisation of genomic alignments in the RD-Connect GPAP

The RD-Connect GPAP enables visualisation of genomic alignments of samples from the Solve-RD project.

There are two ways of accessing the data.

- 1) from the RD-Connect GPAP results section
- 2) from the GPAP integrated IGV browser

1) from the RD-Connect GPAP results section

Select the variant which alignments you would like to visualise from the results section. In the middle section of the platform, go to the “samples” tab and click on the IGV position. You will be redirected to the GPAP IGV browser at this specific region.

Select your variant of interest

Click on the IGV position link

The screenshot shows the RD-Connect GPAP results section. At the top, there are tabs for 'Variants' and 'Cohorts (beta)'. Below this is a filter bar with 'PRESET FILTERS', 'RESET', 'SHARE', and 'RUN QUERY' buttons. The variant type is set to 'high moderate' and the population to 'gnomad_af'. A table of variants is displayed with columns for 'RD-Connect ID', 'Participant ID', 'GT', 'GQ', 'DP', and 'AAF'. The first row shows 'E002118' for 'RUMC_solve_rd_thaca_1587' with 'GT: 0/1', 'GQ: 99', 'DP: 116', and 'AAF: 0.56'. A blue box highlights the 'IGV Position' and 'IGV Region' links in the first row. Below the table, there are tabs for 'Phenotype', 'Analysis status', 'Variants (643)', 'Samples ()', and 'Exomiser'. A detailed variant table is shown at the bottom with columns for 'Chr', 'Position', 'dbSNP', 'Ref', 'Alt', 'Candidate', 'GT', 'INDEL', 'Gene', 'Effect Impact', 'ClinVar', 'CADD', 'SIFT', 'PP2', 'MT', '1000GP AF', 'gnomAD AF', and 'Internal Freq'.

RD-Connect ID	Participant ID	GT	GQ	DP	AAF
E002118	RUMC_solve_rd_thaca_1587	0/1	99	116	0.56

Chr	Position	dbSNP	Ref	Alt	Candidate	GT	INDEL	Gene	Effect Impact	ClinVar	CADD	SIFT	PP2	MT	1000GP AF	gnomAD AF	Internal Freq
1	3329213	rs201904226	G	A	0	TAG	G/A	PRDM16	MODERATE	B/LB	NA	T	D	N	NA	0.002039	0.002755

2) the GPAP integrated IGV browser

Go to: <https://platform.rd-connect.eu/igvgpap>

Enter the sample and genomic coordinates you would like to visualise and click on “search & see”.

The IGV-Browser (igv.js) hosted by the Genome-Phenome Analysis Platform (GPAP) accesses alignments served by the [European Genome-phenome Archive \(EGA\)](#) through its htsgset server.

Please, select an Experiment ID and a genomic location to visualise it in the IGB Browser.

Enter Experiment ID

Chromosome:

Enter Locus:

[Search & See](#)

IGV browser

You can edit the setting of the browser by opening the configuration menu on the top right of the screen (e.g . color by read strand, show soft clips, etc.)

The screenshot displays the IGV browser interface. At the top, the genome is identified as hg19, chromosome 1, with a specific region (chr1:3,329,194-3,329,224) selected, spanning 32 bp. The interface includes a search bar, navigation buttons (Cursor Guide, Center Line, Track Labels, Save SVG), and zoom controls. The main view shows a reference sequence (C C G T G C C A G C C A A A C G G C G G C G G C G G) and a track of sequencing reads. A configuration menu is open on the right side, highlighted with a red box, showing options for track name, height, color, and visibility. The 'Color by' section is expanded, showing options for read strand, first-of-pair strand, pair orientation, insert size (TLEN), tag, Show all bases, View as pairs, Show soft clips, Set visibility window, and Remove track.

IGV hg19 chr1 chr1:3,329,194-3,329,224 32 bp

Cursor Guide Center Line Track Labels Save SVG

3,329,195 bp 3,329,200 bp 3,329,205 bp 3,329,210 bp 3,329,215 bp 3,329,220 bp 3,329,225 bp

100 E002118

0

Refseq Genes PRDM16 PRDM16

Configuration menu options:

- Set track name
- Set track height
- Set track color
- Color by
 - read strand
 - first-of-pair strand
 - pair orientation
 - insert size (TLEN)
 - tag
- Show all bases
- View as pairs
- Show soft clips
- Set visibility window
- Remove track