

ggoncplot: an R package for visualising somatic mutation data from cancer patient cohorts

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Summary

The ggoncplot R package generates interactive oncoplots (also called oncoprints) to visualize mutational patterns across patient cancer cohorts (Figure 1). Oncoplots reveal patterns of gene co-mutation and include marginal plots that indicate co-occurrence of gene mutations and tumour features. It is useful to relate gene mutation patterns seen in an oncoplot to patterns seen in other plot types, including gene expression t-SNE plots or methylation UMAPs. There are, however, no existing oncoplot-generating R packages that support dynamic data linkage between different plots. To address this gap and enable rapid exploration of a variety of data types we constructed the ggoncplot package for the production of oncoplots that are easily integrated with custom visualisations and that support synchronised data-selections across plots (Figure 2). ggoncplot is available on GitHub at <https://github.com/selkamand/ggoncplot>.

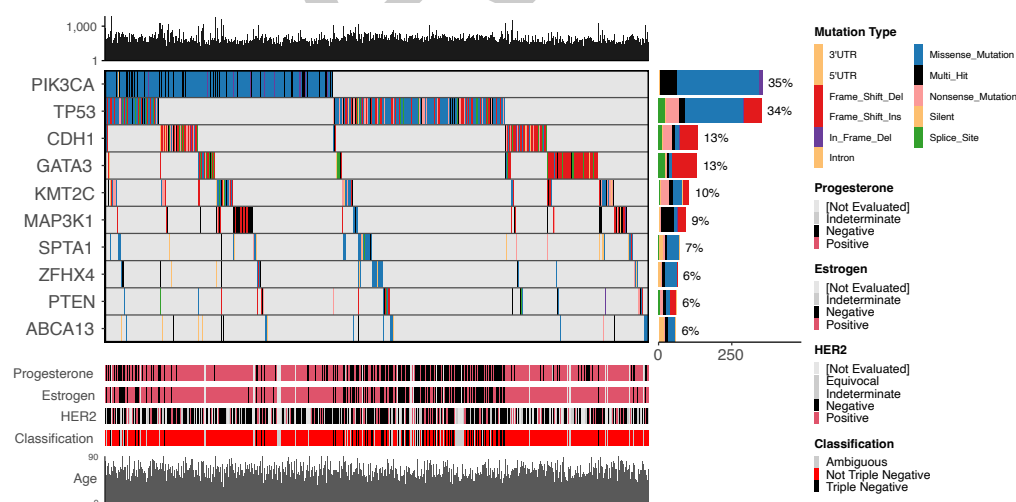


Figure 1: ggoncplot output visualising mutational trends in the TCGA breast carcinoma cohort. Individual patient samples are plotted on the x-axis, ordered by ggoncplot. The plot indicates (y-axis, sorted by genes mutation frequency) that PIK3CA is the most frequently mutated gene, followed by TP53. Marginal plots indicate the total number of mutations per sample (top), and the number of samples showing mutations in each gene, coloured by mutation type (right). A range of clinical features, including progesterone and estrogen receptor status are shown on the marginal plot at the bottom.

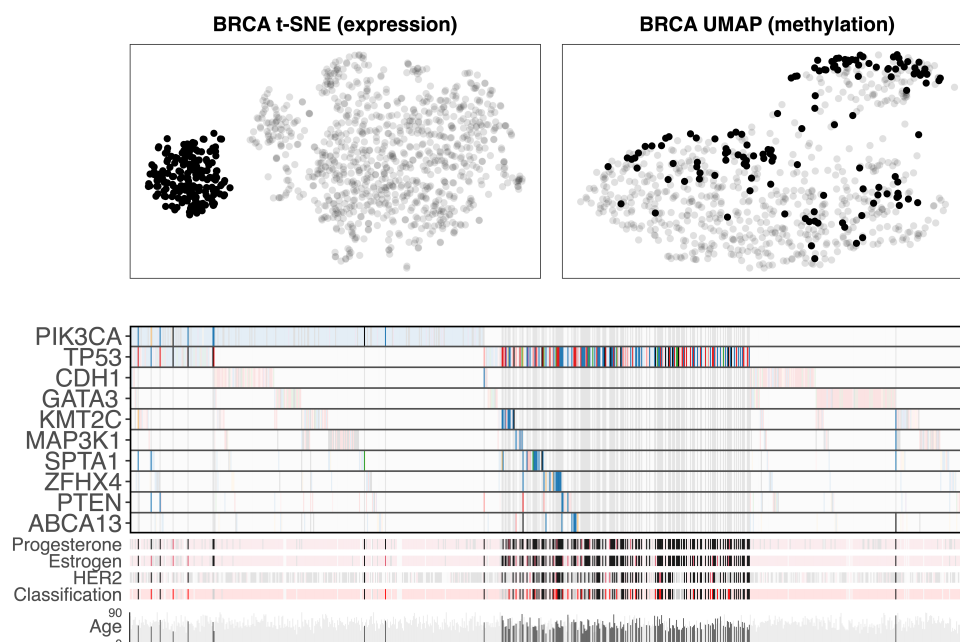


Figure 2: Example of the ggoncplot shown in Figure 1, where the oncoplot has been dynamically cross-linked to a gene expression t-SNE plot (top left) and a methylation UMAP (top right). Here, the lasso tool (see Figure 3) was used to select a cluster of gene expression data points (i.e., individual samples) in the t-SNE plot. Selected samples were automatically highlighted on the UMAP and oncoplot. This reveals that samples which cluster on the left of the t-SNE plot also cluster in the oncoplot, chiefly containing mutations in TP53 but wild type PIK3CA. The plots of progesterone, estrogen, HER2 status and triple negative classification show that the samples selected in the t-SNE are all virtually all triple negative breast cancers. In contrast to the oncoplot, the methylation UMAP shows no strong clustering, in line with knowledge of methylation patterns in triple negative breast cancer.

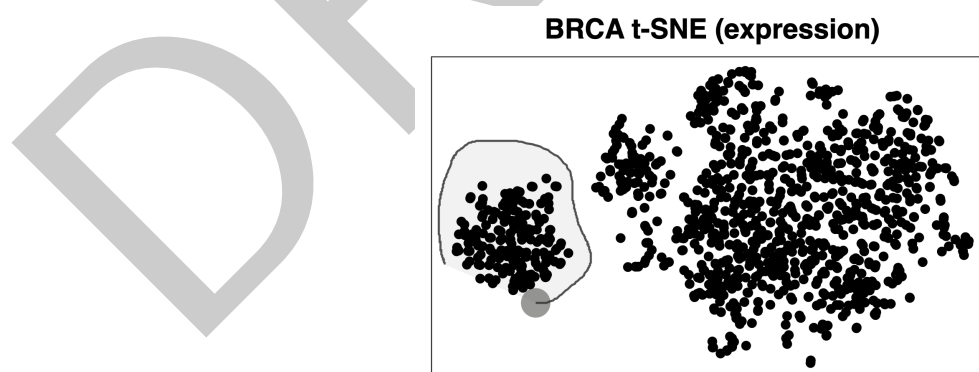


Figure 3: Example image of the t-SNE plot where the lasso tool is being used to manually delineate a data point cluster on the left side.

17 Statement of Need

18 Oncoplots are highly effectively for visualising cohort-level mutation but are challenging to
19 generate with the major R plotting systems (base, lattice, or ggplot2) due to their algorithmic
20 and graphical complexity. Simplifying the process would make oncoplots more accessible to

researchers. Packages like ComplexHeatmap (Gu, 2022), maftools (Mayakonda et al., 2018), and genVisR (Skidmore et al., 2016) all make static oncoplots easier to create, but there is still a significant unmet need for an easy method of creating oncoplots with the following features:

- Interactive plots: Customizable tooltips, cross-selection of samples across different plots, and auto-copying of sample identifiers on click.
- Support for tidy datasets: Compatibility with tidy, tabular mutation-level formats (MAF files or relational databases), typical of cancer cohort datasets.
- Auto colouring: Automatic selection of color palettes for datasets where consequence annotations are aligned with standard variant effect dictionaries (PAVE, SO, or MAF).
- Versatility: The ability to visualize entities other than gene mutations, including non-coding features (e.g., enhancers) and non-genomic entities (e.g., microbial presence in microbiome datasets).

We developed ggoncplot as the first R package that addresses all these challenges simultaneously (Figure 4). Examples of all key features are available in the ggoncplot manual.

Property	complexheatmap	maftools	GenVisR	ggoncplot
Sample sorting algorithm	memo sort	heirarchical sort	heirarchical sort	heirarchical sort
Plotting framework	BaseR	BaseR	ggplot2	ggplot2
Automatic rendering of clinical annotations as bar or tile plots based on datatype	No	No	No	Yes
Works on tabular, tidy, long-form input data as would be stored in large databases	No	Yes	Yes	Yes
Interactive	Yes ¹	No	No	Yes
Customisable tooltips	No	No	No	Yes
Allows any mutation dictionary to be used	Yes	No	Yes	Yes
Automatic colour palette selection when mutation impact dictionary conforms to known ontologies	No	Yes (MAF only)	No	Yes (MAF, SO, or PAVE)
Approach for resolving genes with multiple mutations	Different Visualisation on Plot ²	Flags as Multi-Hit	Picks more severe consequence or leaves to user ³	Flags as Multi-Hit
Supports a mutation level dataset as input	No ⁴	Yes	Yes	Yes
Native support for faceting by pathway	No	Yes	No	Yes
Supports marginal plots describing TMB, gene mutation recurrence, and clinical annotations	Yes	Yes	Yes	Yes

Figure 4: Comparison of R packages for creating oncoplots. ¹Requires the shiny and interactiveComplexHeatmap packages. ²Requires the user to first summarise mutations at the gene level and format as a sample by gene matrix with mutations separated by semicolons (wide format). ³For MAF inputs the most severe consequence is chosen, however for non-MAF datasets users must manually define the mutation impact hierarchy. ⁴Non-unique mutation types are treated as one observation, however if different mutation types affect one gene, the individual mutations can be plotted with different shapes or sizes in a user-configured manner.

Acknowledgements

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