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GenViz Module 3: Introduction to GenVisR

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Genomic Data Visualization and Interpretation

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Learning objectives of the course

- Module 1: Introduction to genomic data visualization and interpretation
- Module 2: Using R for genomic data visualization and interpretation
- **Module 3: Introduction to GenVisR**
- Module 4: Expression profiling, visualization, and interpretation
- Module 5: Variant annotation and interpretation
- Module 6: Q & A, discussion, integrated assignments, and working with your own data
- Tutorials
 - Provide working examples of data visualization and interpretation
 - Self contained, self explanatory, portable

Learning objectives of module 3

- Introduction to GenVisR
 - Creation and interpretation of GenVisR plots
 - waterfall()
 - TvTi()
 - genCov()
 - cnFreq()
 - cnSpec()
 - lohSpec()
 - covBars()
 - Adding plot layers to GenVisR
 - Arranging viewports with gridExtra
 - Aligning viewports with gtable

What is GenVisR?

- Provides a toolkit for visualizing Genomic data
- Implemented in R
- Emphasis on producing “publication quality” graphics with a minimal amount of user input.
- Part of bioconductor
 - <https://www.bioconductor.org/packages/3.3/bioc/html/GenVisR.html>
- Active development on github
 - <https://github.com/griffithlab/GenVisR>
- Functions focused in three areas “Small variants”, “Copy number alterations”, and “Data quality”

Why GenVisR?

- The package is built upon ggplot2 and will allow us to leverage information we've learned in previous modules.
- The package is intended to be flexible, supporting multiple file types, species, etc.
- The package is relatively popular; In the top 20% of bioconductor downloads.
- The package is regularly updated with improvements, bug fixes, etc.
- The package is maintained by the Griffith lab.

GenVisR::waterfall()

- What does it do?
 - Visualize the types of mutations within a cohort
 - Visualize the mutation burden in a data set
 - Visualize the proportion of samples with a mutated gene
 - Visualize clinical data
- Why do we care?
 - Determine mutually exclusive or co-occurring genomic events
 - Recognize patterns within clinical data
 - See if the mutation burden conforms to expectations

GenVisR::waterfall()

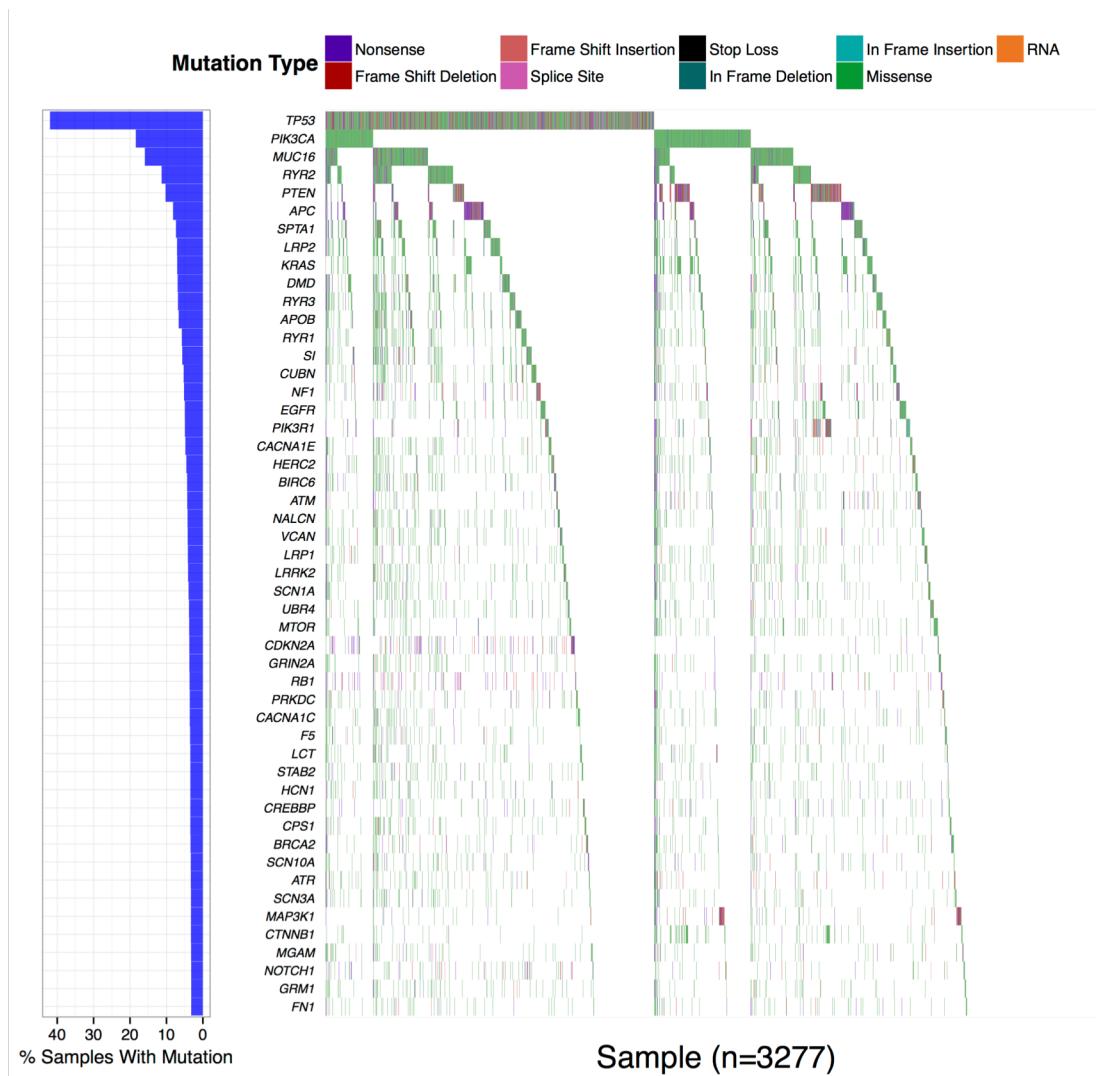
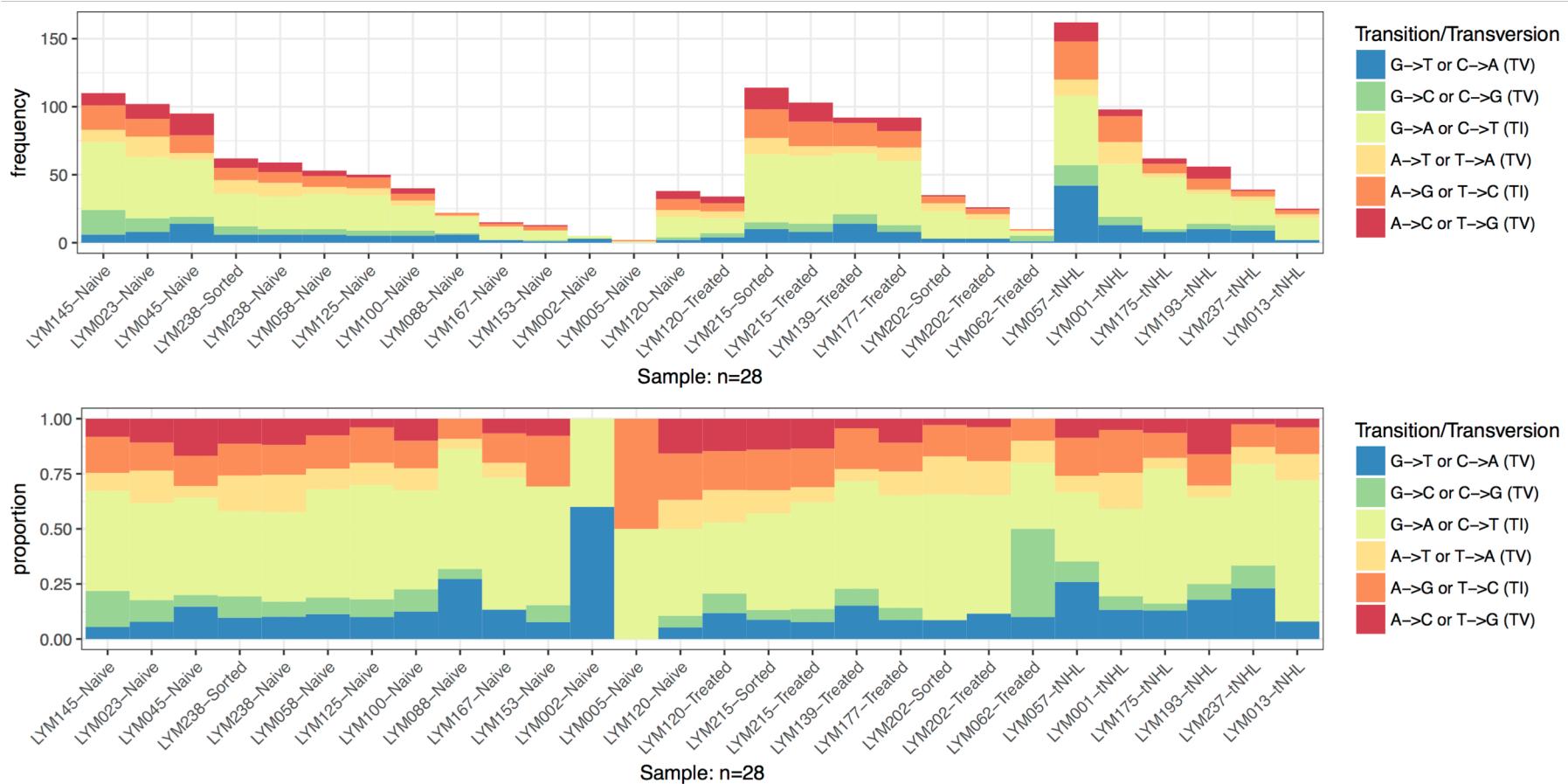


Fig. 2: “DGIdb 2.0: mining clinically relevant drug-gene interactions”

GenVisR::TvTi()

- What does it do?
 - Visualize Transition and Transversion proportions
 - Visualize Transition and Transversion frequencies
 - Compare these proportions with expectations
- Why do we care?
 - Visualize mutation profile patterns
 - Ex. Smoking tends to increase G -> T/C -> A transversions due to oxidative damage
 - Practical application verify smoking status of patients with lung carcinomas

GenVisR::TvTi()

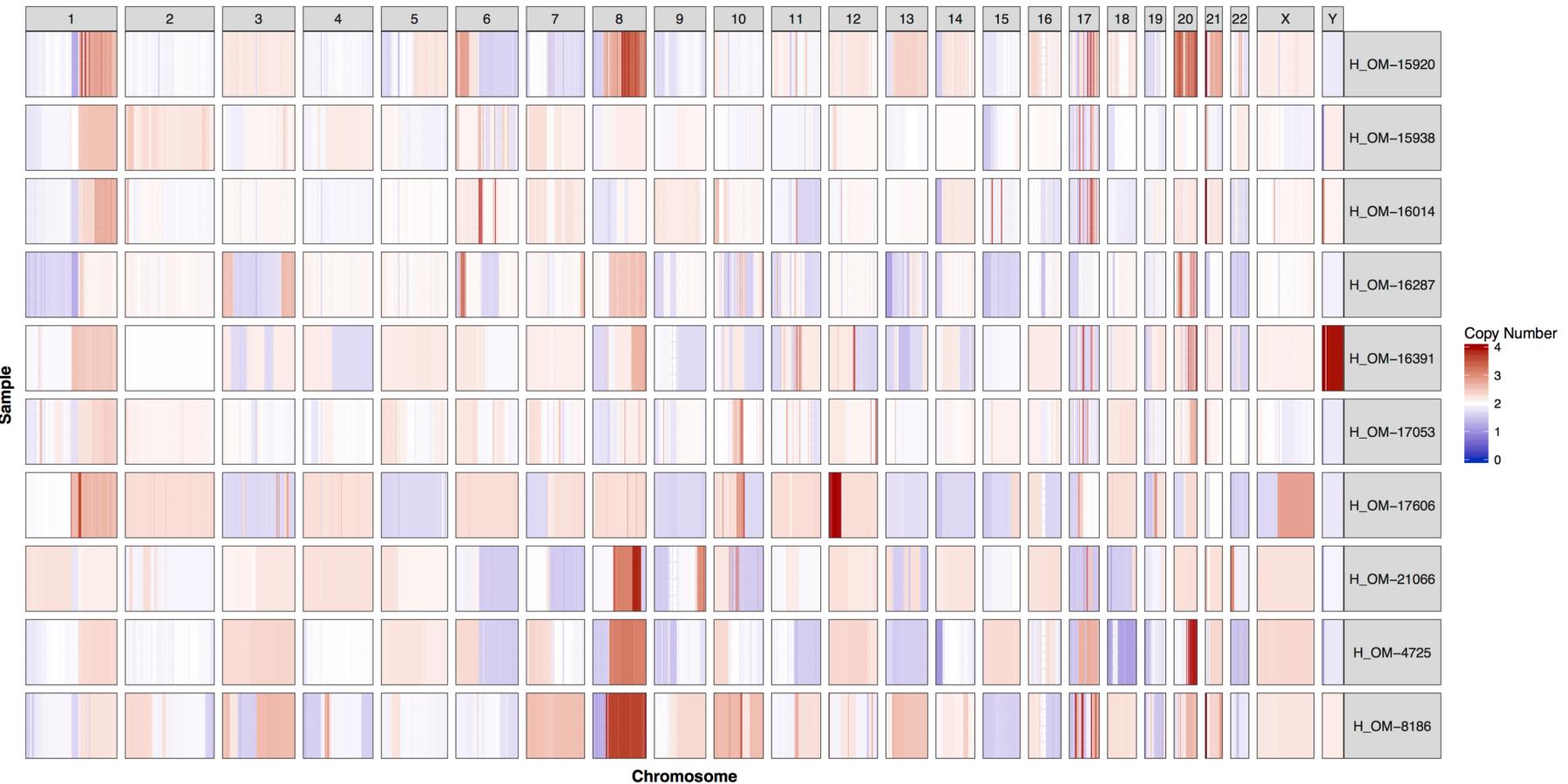


This research was originally published in Blood. Krysiak et al. Blood 2017 129:473-483 by Krysiak et al. licensed under © the American Society of Hematology

GenVisR::cnSpec()

- What does it do?
 - Display recurrent copy number alterations
- Why do we care?
 - Recognize patterns of amplifications and deletions while maintaining sample information

GenVisR::cnSpec()

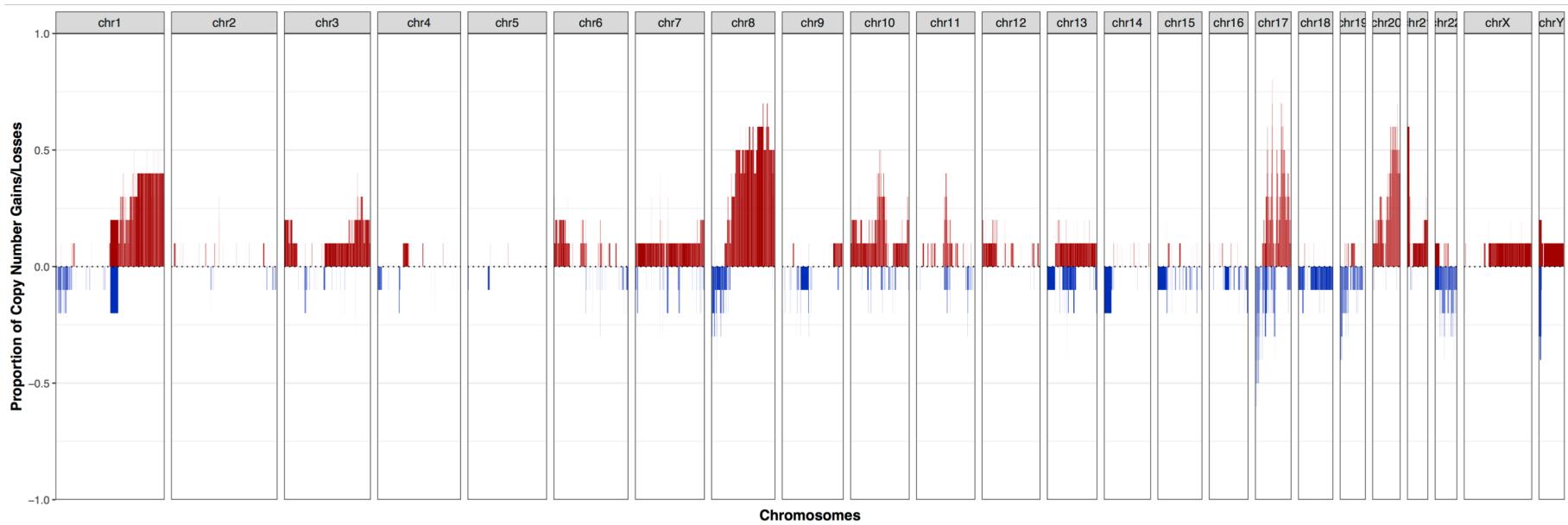


Data from: "Genomic characterization of HER2-positive breast cancer and response to neoadjuvant trastuzumab and chemotherapy-results from the ACOSOG Z1041 (Alliance) trial."

GenVisR::cnFreq()

- What does it do?
 - Another view displayed from cnSpec
 - Summarize copy number alterations across a cohort
 - Plots segmented copy number alterations for a cohort
- Why do we care?
 - Easily view recurrently amplified/deleted regions

GenVisR::cnFreq()

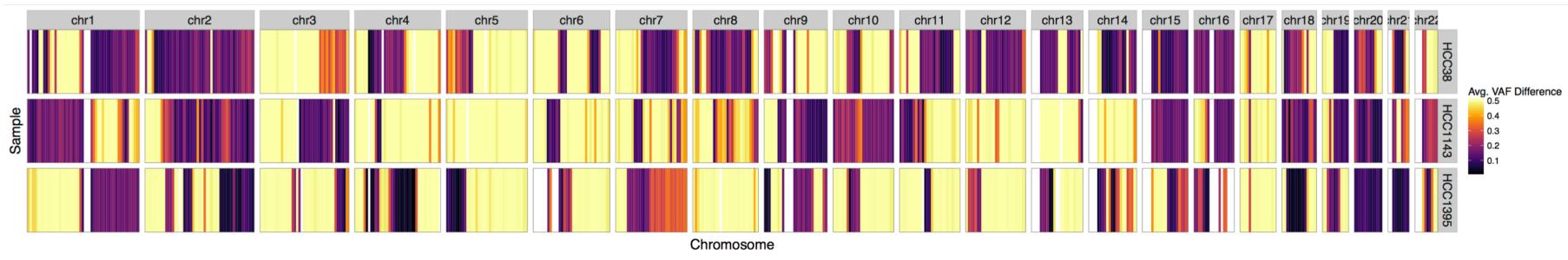


Data from: "Genomic characterization of HER2-positive breast cancer and response to neoadjuvant trastuzumab and chemotherapy-results from the ACOSOG Z1041 (Alliance) trial."

GenVisR::lohSpec()

- What does it do?
 - Visualize loss of heterozygosity (LOH) for multiple samples
- Why we care?
 - Copy-neutral LOH could be missed with a plot produced with functions such as cnSpec however these events are potentially interesting
 - lohSpec allows these events to be viewed and summarized for multiple samples allowing viewing of recurrent LOH patterns

GenVisR::lohSpec() for 3 breast cancer cell lines



GenVisR::genCov()

- What does it do?
 - Visualize coverage in a region of interest
- Why do we care?
 - Infer the impact of a structural deletion
 - Verify gene knockouts

GenVisR::genCov()

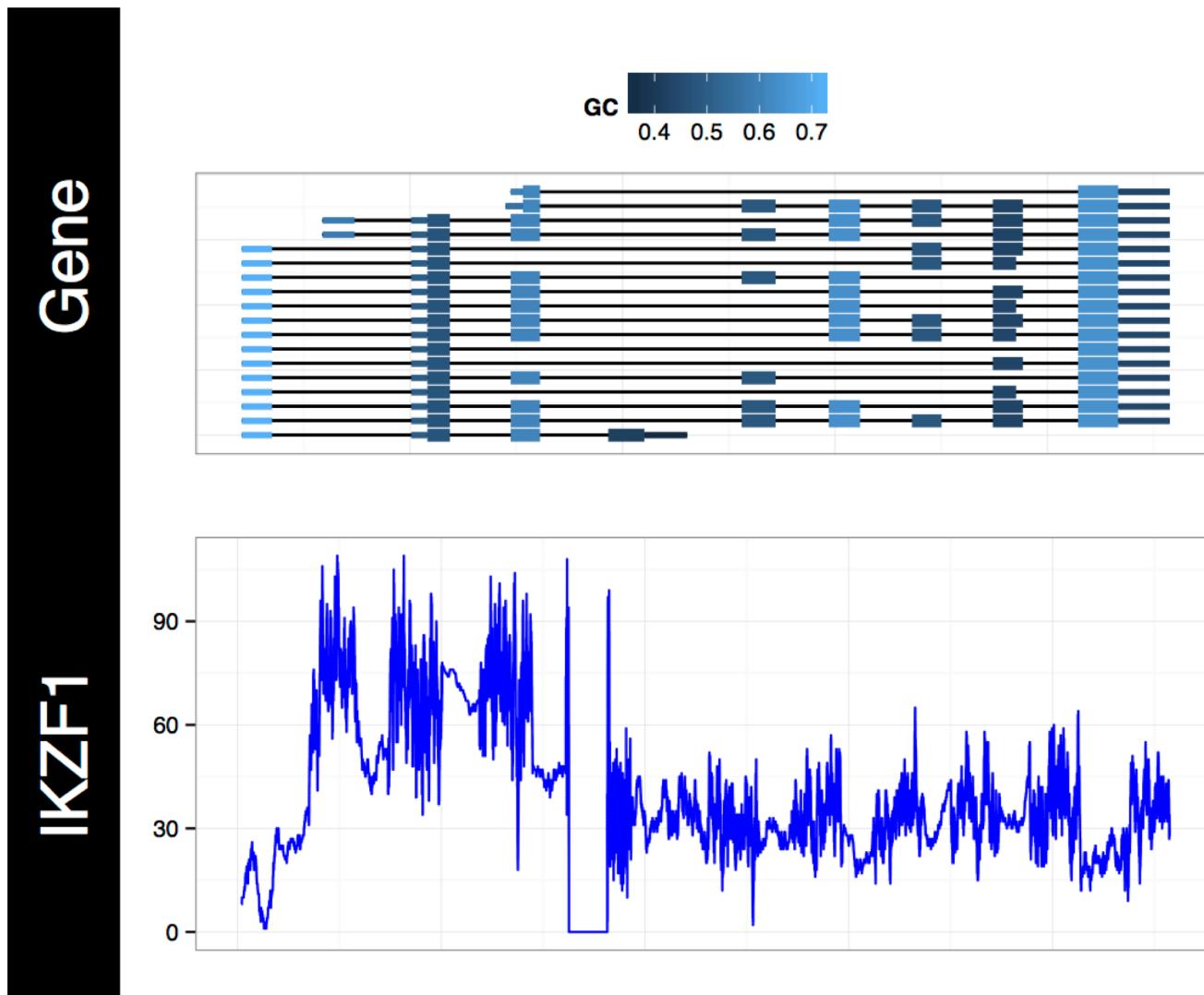
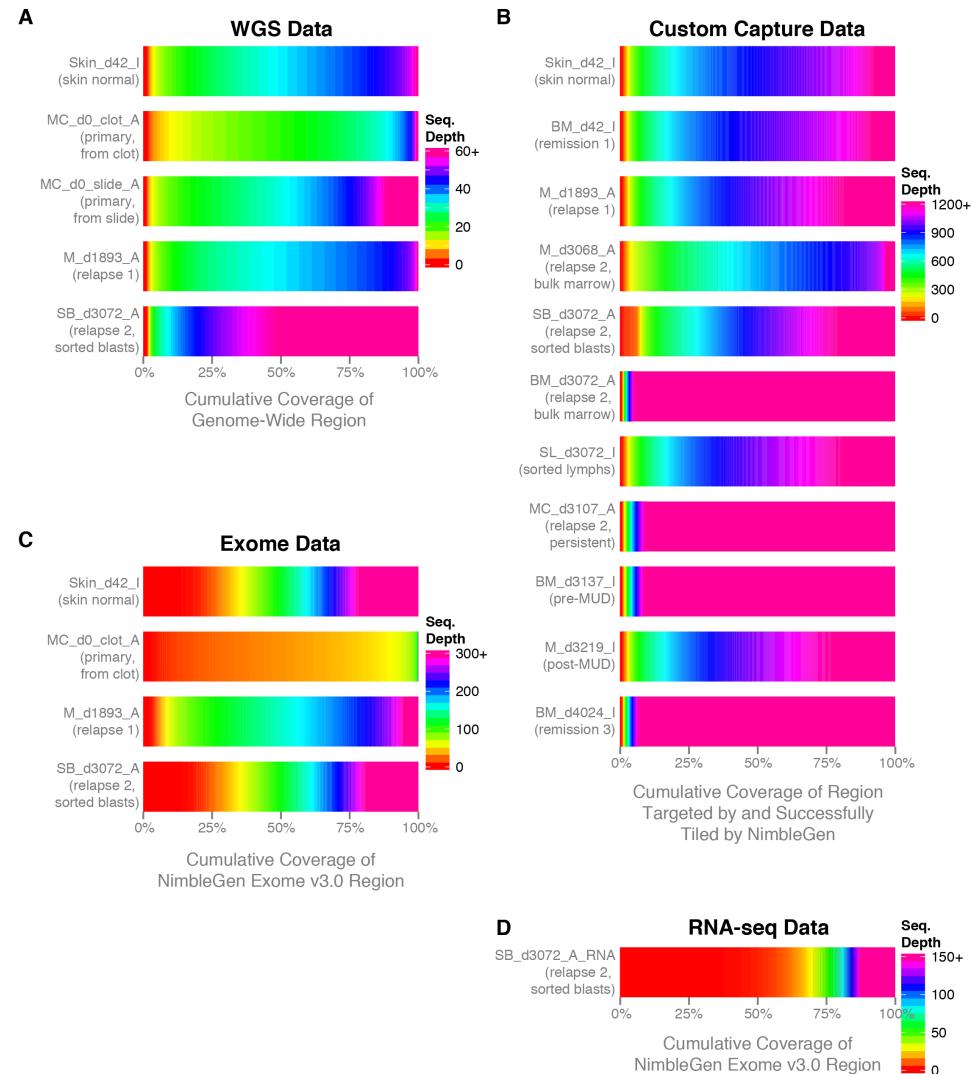


Fig. 2: “Comprehensive genomic analysis reveals FLT3 activation and a therapeutic strategy for a patient with relapsed adult B-lymphoblastic leukemia”

GenVisR::covBars()

- What does it do?
 - Visualize sequencing coverage achieved across samples
- Why do we care?
 - Immediately identify if a sample has failed sequencing or has not achieved the expected sequencing depth

GenVisR::covBars()



Supp. Fig. 3: “Comprehensive genomic analysis reveals FLT3 activation and a therapeutic strategy for a patient with relapsed adult B-lymphoblastic leukemia”