

## **Title**

Visualizing genomic instability in tumors for rapid and robust identification of HRD patients

## **Category**

Processing Pipelines And Methods

## **Challenge**

Development of visualization methods/pipeline/GUI to highlight genomic scar features that identify homologous repair deficient (HRD) samples vs normal samples.

HRD can be assessed by quantifying the total amount of allelic imbalance, single-base substitution signatures, and indel signatures in tumor samples. We are curious about the following: (1) How do these different metrics compare to each other within HRD and normal samples? (2) Can we develop visualizations to quickly contrast deficient vs intact tumors? (3) Are the locations of genomic scars on the genome random or distributed in certain regions?

Can we create an intuitive image/overlay/visualization that highlights differences in genomic scars that enables rapid and robust discrimination between HRD and normal patient samples?

Inputs for each sample could consist of WGS/WES data, variant call information, and mutational signature information (COSMIC) readily available on TCGA or St. Jude Cloud – Genomics Platform, etc.

## **Benefit**

The community needs a visualization to assist experimental biologist or clinicians to quickly assess HR-deficiency or genomic instability in tumor patients. A direct benefit of this work will be identification of driving sources of genomic instability which give rise to genomic scars or lesions. These visualizations can be added to existing annotations on St. Jude pediatric portals.

## **Helpful Tools, Packages, or Software**

Genomic Scars: ScarHRD (R package), Sequenza (unix tool), SigProfiler (Python suite), MutationalPatterns (R package), Signal (<https://signal.mutationalsignatures.com/>) Visualization: R Shiny app, Perl Circos module, MatLab Plot, etc.

Examples of contrasts: Myriad Genetics Slide 8

(<https://investor.myriad.com/static-files/8ef72264-ca8c-4b01-b0e9-35fa83c63889>) & HRDetect Circos Plots

(<https://www.nature.com/articles/nm.4292/figures/1>)

## **Test Data**

Cancer genomic datasets: TCGA, COSMIC, St. Jude Cloud