

Tapestri CNV Analysis - Instruction Manual v1.0

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1. Introduction

Using the Mission Bio Tapestri Platform, reagents and the `tapestri.cnv` R package, we explore Single Nucleotide Variants (SNVs) and Copy Number Variations (CNVs), including loss of heterozygosity (LOH), which plays a large role in cancer evolution and contributes to cancer heterogeneity. More details can be found in the [Application Note](#).

The `tapestri.cnv` R package is an exploratory data analysis tool to discover gene-level and/or chromosome-level amplifications and deletions, including LOH. This manual provides instructions on how to download, install and use the package.

If you have any feedback or questions regarding the content of this document and the scripts provided contact Mission Bio Support at support@missionbio.com.

2. Prerequisites

This guide is intended for users with access to the Tapestri Instrument and the Tapestri Pipeline software.

Hardware requirements

To use the R package, the hardware requirements are:

- Minimum of 8 GB of RAM (16GB highly recommended)

Software dependencies

The following software/packages need to be installed in order to use these R scripts:

- R (v3.6.0 or later)

3. Installation

Install R

3.1. Install [R](#) version 3.6 or later (recommended: Install [R Studio](#)). Open R or R Studio and execute the below commands to Install all required packages.

```
#Install `devtools` first and then install the other packages. `devtools`
will attempt to automatically install prerequisites for these packages.

#NOTE: Install each package independently, as the install script may prompt
to update previously downloaded packages.

> install.packages(c("devtools", "hdf5r", "digest"),
repos='http://cran.us.r-project.org')
> devtools::install_github("mojaveazure/loomR")
> devtools::install_github("jokergoo/ComplexHeatmap")

> if (!requireNamespace("BiocManager", quietly = TRUE))
  install.packages("BiocManager")
> BiocManager::install("karyoploteR")
> BiocManager::install("annotatr")
> BiocManager::install("TxDb.Hsapiens.UCSC.hg19.knownGene")
> BiocManager::install("org.Hs.eg.db")
```

Install Tapestri CNV R package

3.2. Download Tapestri CNV R package [zip file](#) & then install as a local package by executing the below commands.

```
#type in the path to the zip file before installation
> devtools::install_local(path = "tapestri.cnv_1.0.0.tar.gz",
repos='http://cran.us.r-project.org', upgrade="never")
```

```
#view package contents  
> help(package="tapestri.cnv")
```

4. Download demo datasets

Two demo datasets are available to test the Tapestri CNV R package.

- 4-cell line mix that was processed with the Tapestri Single-Cell DNA AML Panel.
- 2-cell line mix (KG-1/RAJI 50:50) that was processed with the Tapestri Single-Cell DNA Myeloid Panel.

Instructions on how to process these datasets are included in the R package (in the “inst” folder). The datasets are available from the [datasets](#) menu in the Mission Bio customer portal. Each of the above-mentioned CNV datasets contains the 4 files needed to run the R package.

If you would like to run data from your own panels/runs, download the following files from your Tapestri runs:

File	Filename	Where to find
Amplicon bed file	amplicons. bed *	Panel zip file used for data processing on Tapestri Pipeline
Barcode Distribution file	Example. barcode.cell.distribution.tsv <i>May be 1, 2, or 8 files.</i>	Tapestri Pipeline account
VCF header file	example. vcf_header.txt	Tapestri Pipeline account
Loom file	example. loom	Tapestri Pipeline account

* For your own datasets, please access the panel.zip file you use to process your data on the Tapestri Pipeline. Unzip the file and access the **.bed** file. If you cannot find the file, please contact support@missionbio.com for assistance.

Once downloaded follow the instructions provided in the **README.md** file that is included with the R package.