Pipeline Overview

The DNase-seq pipeline inputs gzipped DNA-sequencing reads in the form of fastqs and a genome index. These fastqs come from technical replicates¹ that are processed with bwa to generate unfiltered and filtered alignments (bam format). The filtered alignments, combined with a DNase mappability reference (https://www.encodeproject.org/references/ENCSR792JJZ/) depending on assembly, provides additional outputs:

- Merging one or more technical replicate bams into a biological replicate level unfiltered bam file; subsequently, filtering and UMI deduplication will produce a filtered bam file.
- Hotspot calling. The filtered bam files will produce hotspots and peak files (bed and bigBed format), and signal files (bigWig format)
 - inputs: filtered_biorep.bam; outputs: hotspots.bed/bb, peaks.bed/bb, signal.bw
- IDR of 2 replicate peak sets. inputs: peaks.bed; outputs: idr.bed/bb, qc

The pipeline does not have any restrictions towards its inputs, however it is recommended to have at least two biological replicates² and the read length be 36nt.

For single-end library preparation: <u>View pipeline</u>
For paired-end library preparation: <u>View pipeline</u>

Genomic References Used in this Pipeline

Unmodified Genome References and Chromosome Sizes: encodeproject.org/ENCSR425FOI

References

Lazarovici A, Zhou T, Shafer A, Machado AC, Riley TR, Sandstrom R, Sabo PJ, Lu Y, Rohs R, Stamatoyannopoulos JA, Bussemaker HJ. Proceedings of the National Academy of Sciences. 2013 Apr 16;110(16):6376-81.

Sabo PJ, Hawrylycz M, Wallace JC, Humbert R, Yu M, Shafer A, Kawamoto J, Hall R, Mack J, Dorschner MO *et al*. <u>Discovery of functional noncoding elements by digital analysis of chromatin structure</u>. *Proc Natl Acad Sci U S A*. 2004 Nov 30;101(48):16837-42.

Sabo PJ, Kuehn MS, Thurman R, Johnson BE, Johnson EM, Cao H, Yu M, Rosenzweig E, Goldy J, Haydock A *et al.* Genome-scale mapping of DNase I sensitivity in vivo using tiling DNA microarrays. *Nat Methods*. 2006 Jul;3(7):511-8.

¹ ENCODE definition of "technical replicates" can be found at (www.encodeproject.org/data-standards/terms/#replication)

² ENCODE definition of "biological replicates" can be found at (www.encodeproject.org/data-standards/terms/#replication)