Citation: Shelton. J.M., Coleman M.C., Herndon. N et al (2015) Tools and pipelines for BioNano data: molecule assembly pipeline and FASTA super scaffolding tool. *BMC Genomics.* **16**(734) [Accessed on 21/02/2019] Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4587741/>

Source: <https://bionanogenomics.com/wp-content/uploads/2018/04/30073-Bionano-Solve-Theory-of-Operation-Hybrid-Scaffold.pdf>

Tools:

Bionano softwares primarily used for hybrid scaffolding and analysing structural variation in genome.

Bionano Irys hardware is used for optical mapping with long-DNA sequencing reads as input and a long genome map as output.

hybrid-scaffold = long genome map + sequencing assembly.

Technical procedures:

Uses of a long genome map:

* validate order of sequencing fragments.
* identify chimeric joins in sequence assembly
* evaluate the size of gap between adjacent sequences.

How to construct hybrid scaffold:

* long-read sequencer produces long DNA reads from sample.
* Irys used for optical mapping. After which a long-genome map is produced from long DNA reads.
* hybrid-scaffold = long genome map/ bionano assembly + sequencing assembly.
* Assume the assembly has good contiguity and quality, >100kbp

Purpose of a hybrid scaffold:

* A representative study of chromosomal structures.
* Improve N50 of assembly.

How Irys works (optical map + hybrid scaffolding automated):

1) generate in silico maps for sequence data;

2) align sequences against Bionano genome maps to identify and resolve potential conflicts (chimeric joints) in either data set;

3) merge the non-conflicting maps into hybrid scaffolds;

4) generate an alignment between the sequence maps and the hybrid scaffolds;

5) generate AGP and FASTA files for the scaffolds.

Chimeric join leads to junction conflicts:

short-reads unable to span across long DNA repeat.

After alignment the junctions for alignment with bionano assembly will be different to alignment with sequencing assembly.

Terms of questions:

contiguity, N50, scaffold, chimeric join/junction conflict between assemblies,

These terms are relevant in lectures:

[ w2:l5, w5:l12-15 ]