

Human genome sequencing on Oxford Nanopore PromethION

Biomina Lunch Talk 2018.03.27

Introduction

- PhD student UA/VIB/IWT
- Oxford Nanopore sequencing
 - MinION
 - PromethION
- Illumina sequencing
 - RNA-sequencing
 - Exome sequencing
 - Targeted resequencing
- Python
 - R



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<https://gigabaseorgigabyte.wordpress.com>



<https://github.com/wdecoaster>



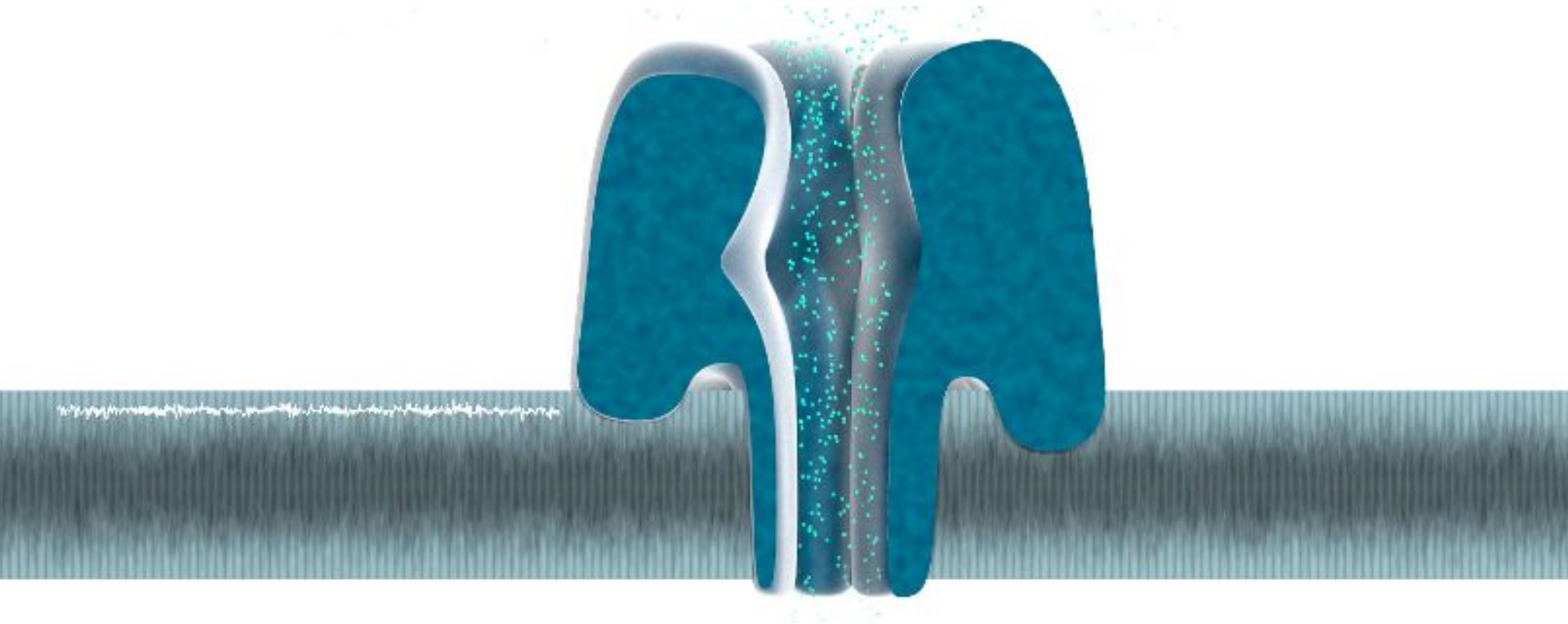
BioStars
— BIOINFORMATICS EXPLAINED —



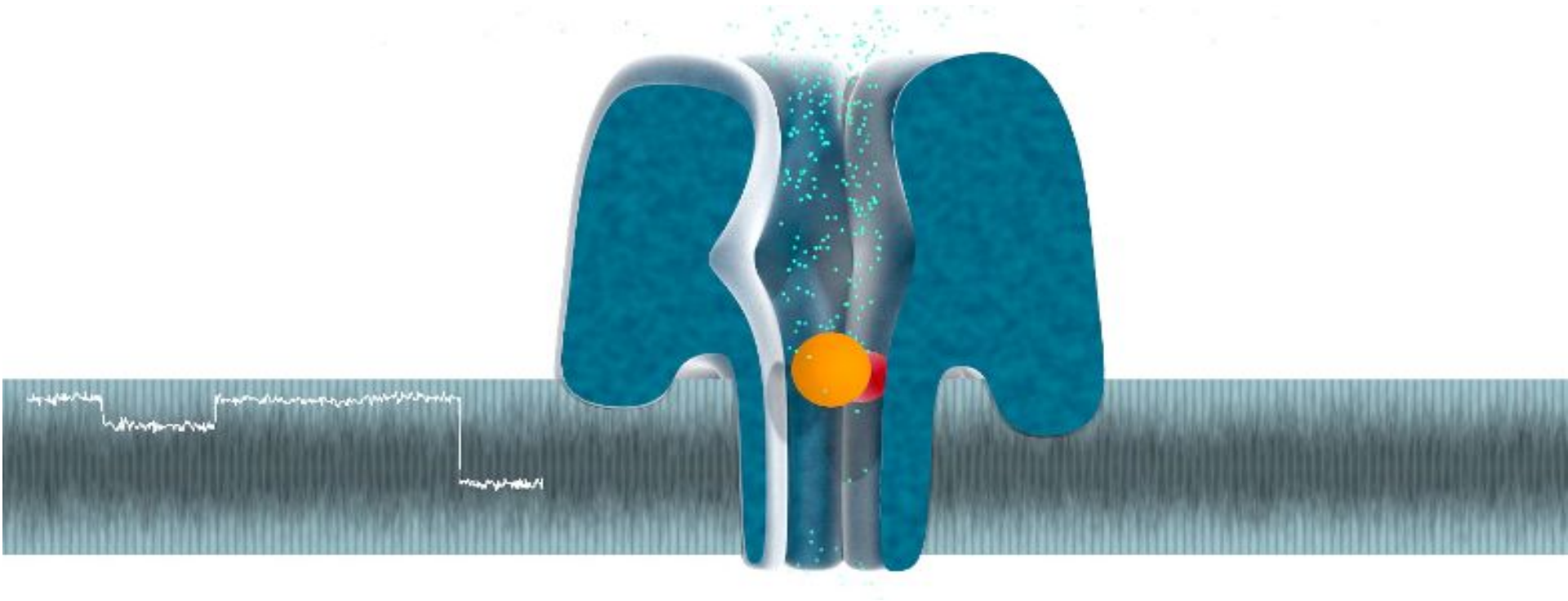
Nanopore sequencing

A minimal introduction

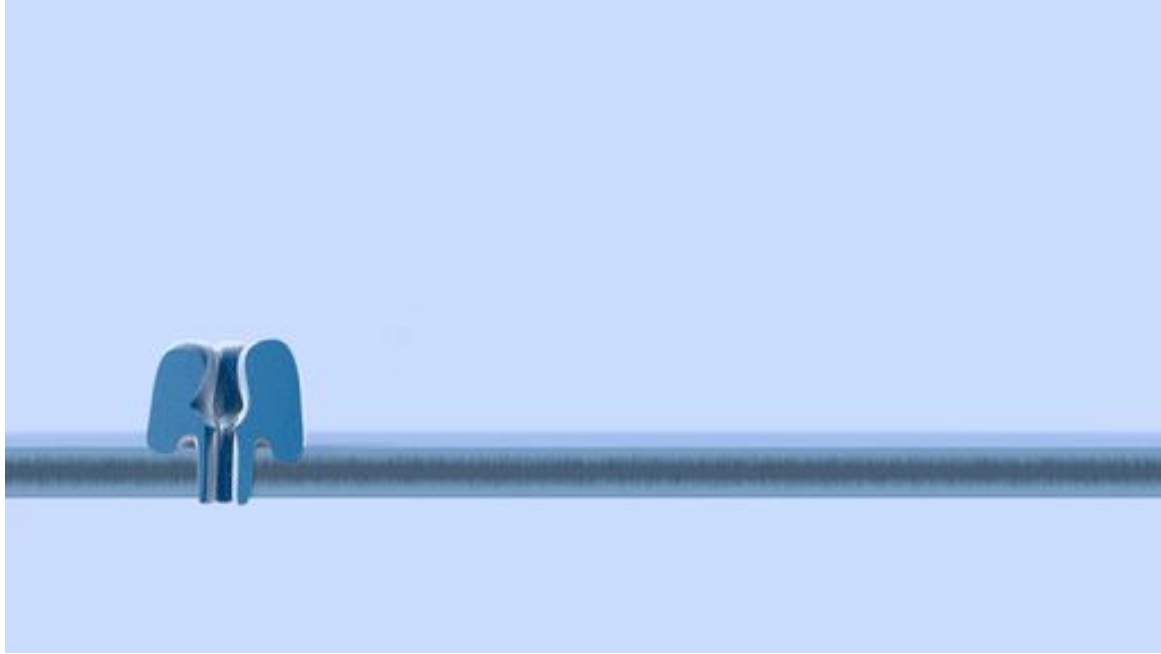
Technology: tiny pores in a membrane



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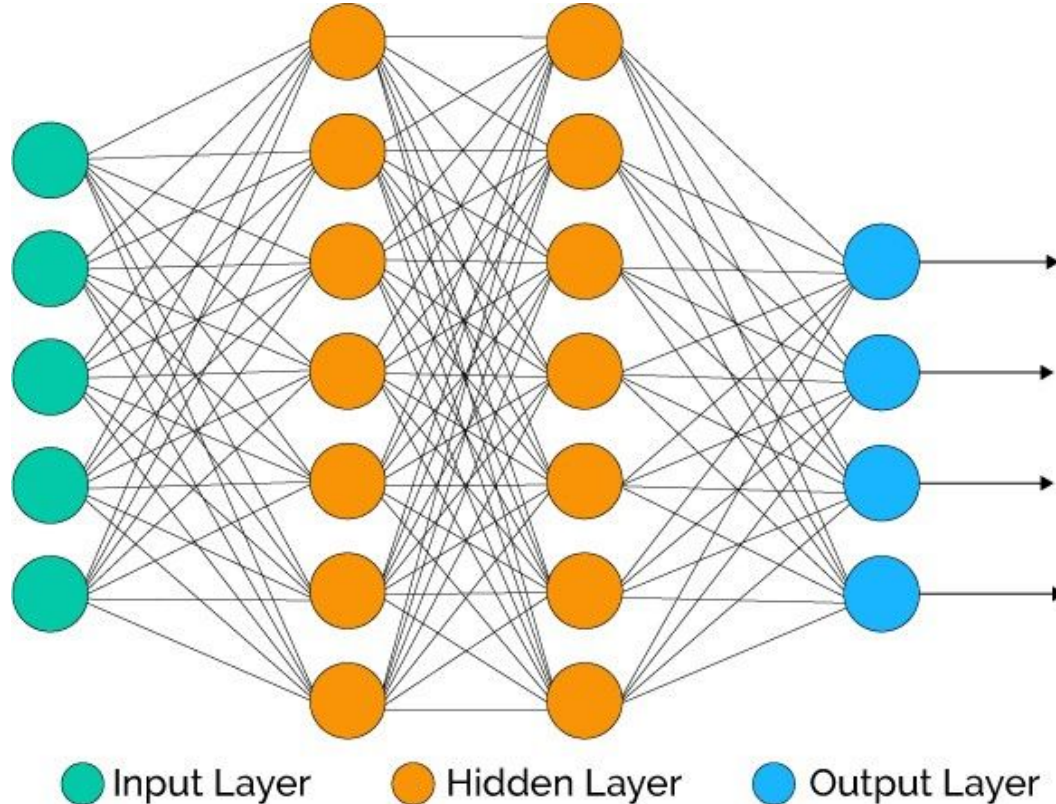


Technology: DNA sequencing



Technology: Recurrent Neural Network

Machine
learning
algorithm



Our Oxford Nanopore sequencing platforms

MinION



PromethION

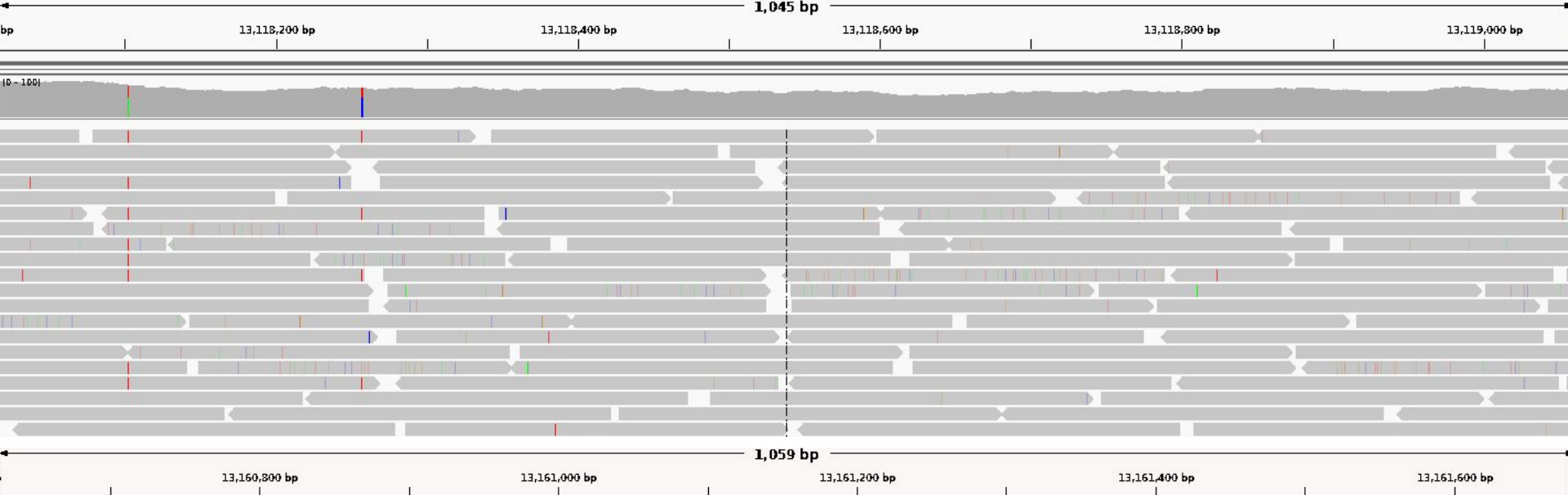


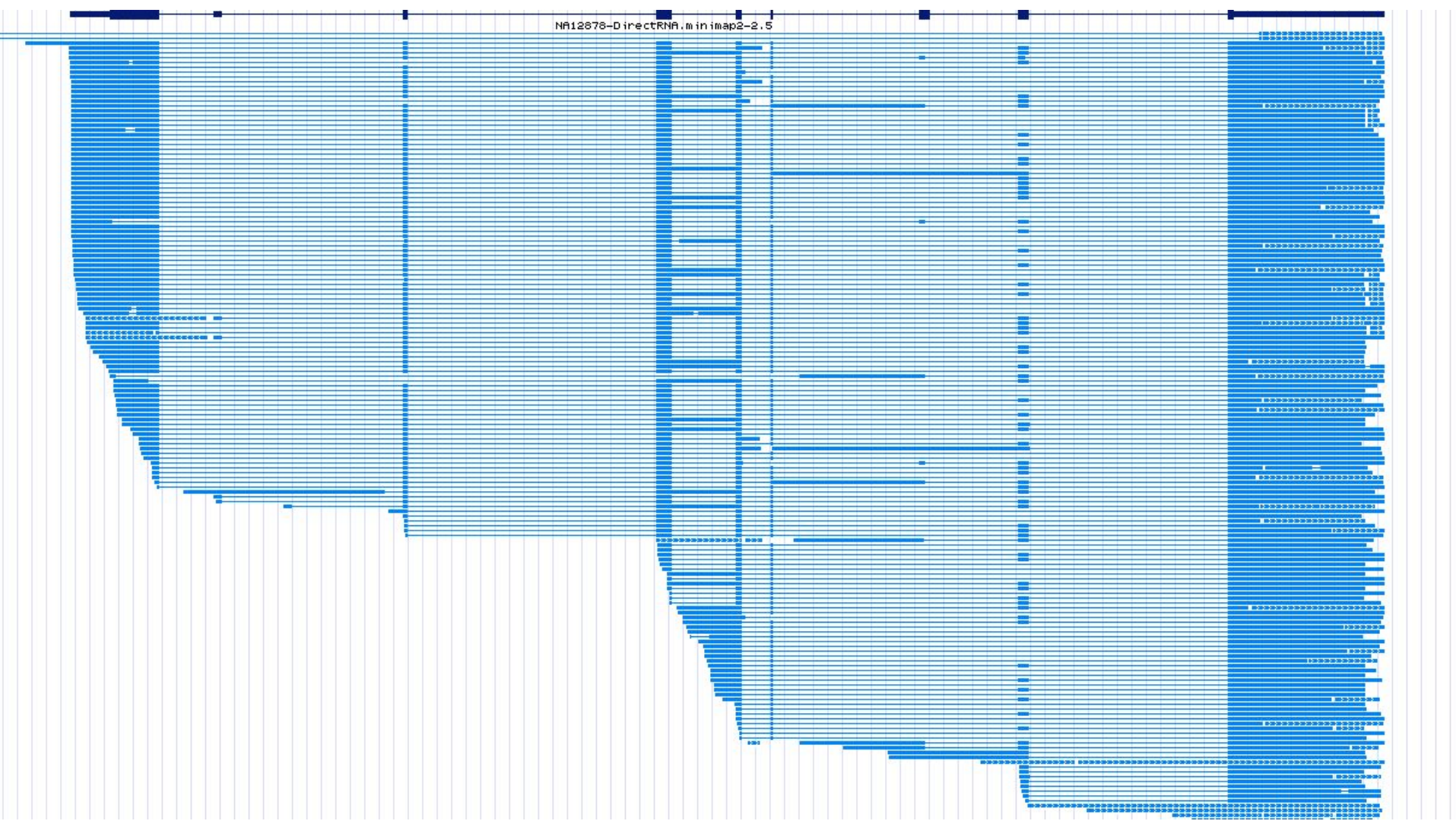
Characteristics

- Raw read accuracy: ~88%
- Long reads
 - Read length only limited by input DNA
 - 10kb is normal or short
 - Longest read: >1Megabase

Applications

- DNA
 - genome assembly
 - structural variant identification
 - phasing of variants
- RNA/cDNA
 - full transcripts
 - alternative splicing
- Native sequencing
 - Nucleotide modifications
- Fast
 - in field identification/diagnostics





NanoPack

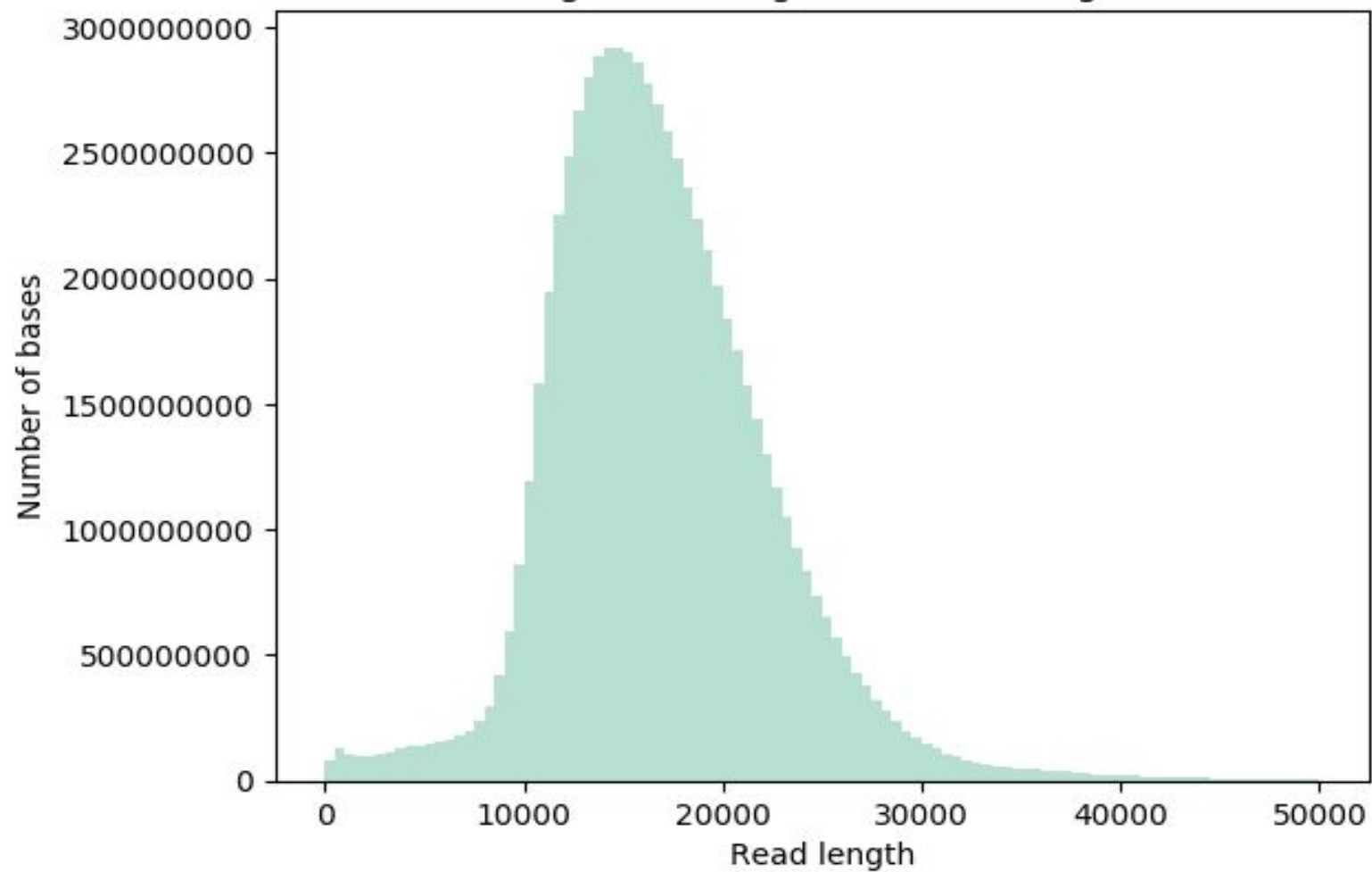
Python scripts for visualizing and processing Oxford Nanopore sequencing data

- NanoPlot `pip install nanopack`
 - NanoComp
 - NanoStat
 - NanoLyse
 - NanoFilt `pip install NanoPlot`
 - NanoQC `conda install nanoplot`
-
- Web service: <http://nanoplot.bioinf.be/>
 - Command line tools: <https://github.com/wdecoster/nanopack>

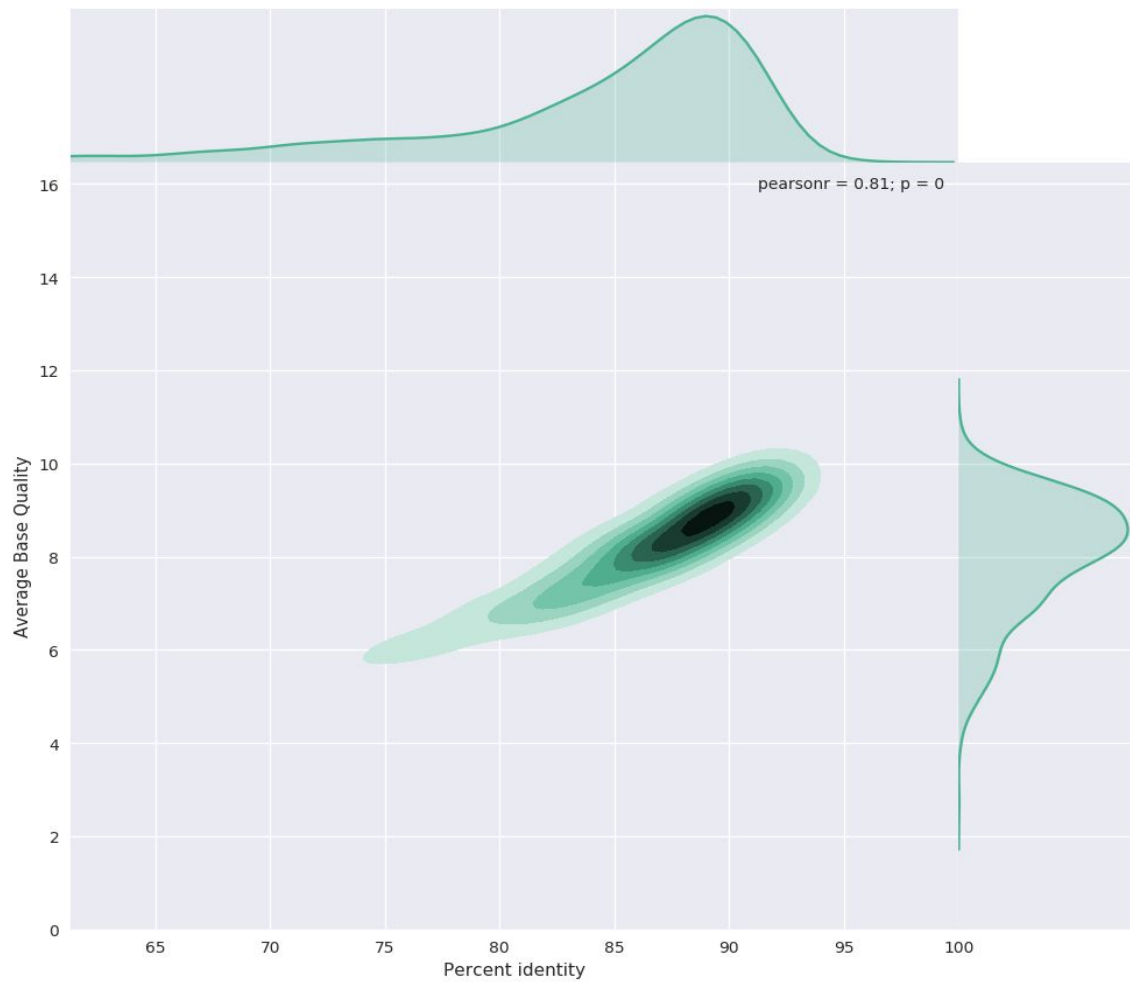
Wouter De Coster, Svernn D'Hert, Darrin T Schultz, Marc Cruts, Christine Van Broeckhoven;

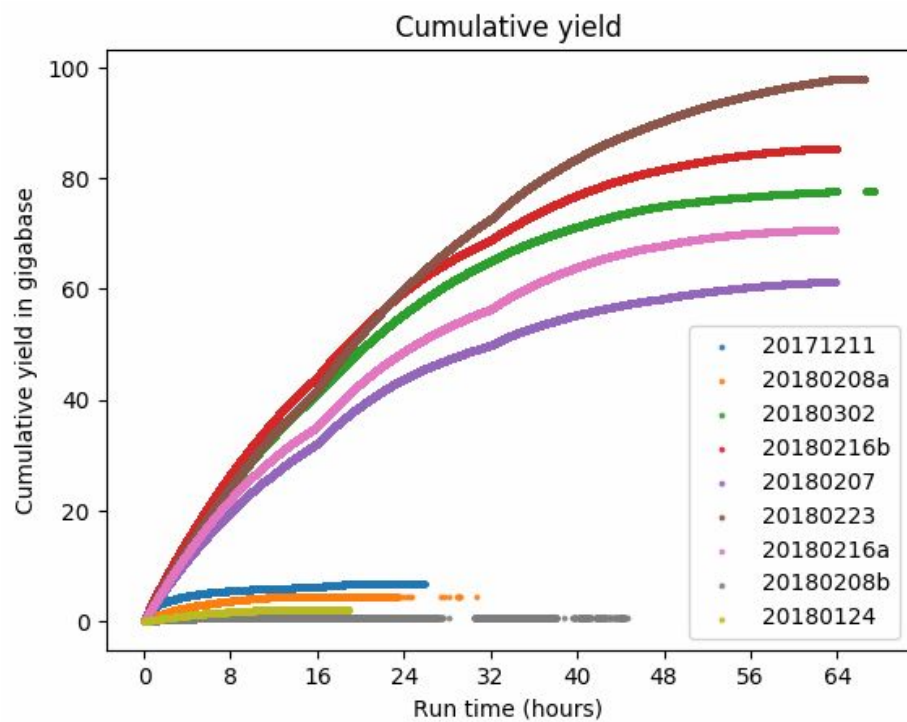
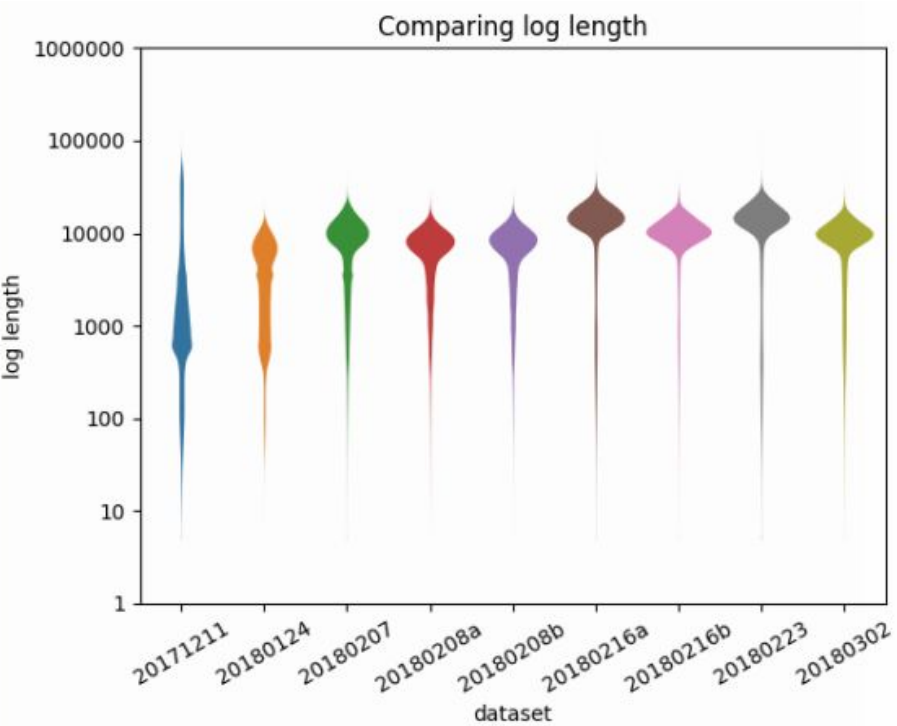
NanoPack: visualizing and processing long read sequencing data, *Bioinformatics*

Weighted Histogram of read lengths

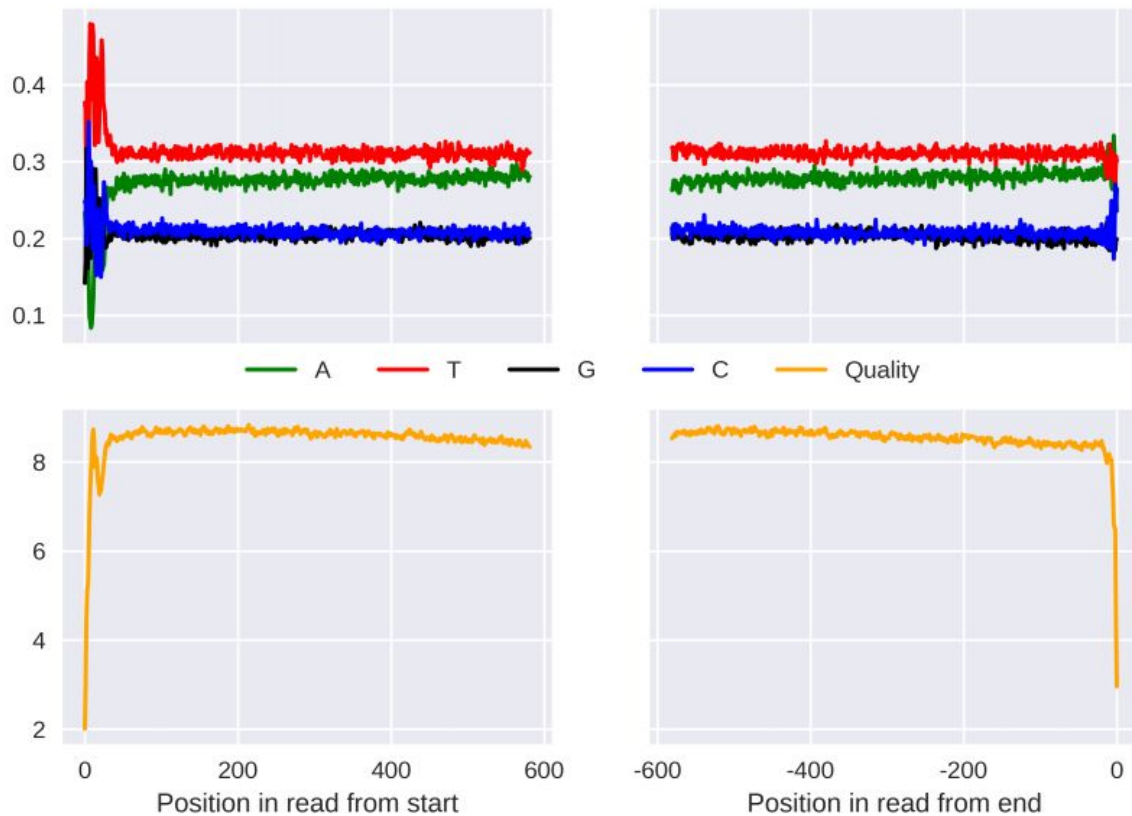


Percent identity vs Average Base Quality plot

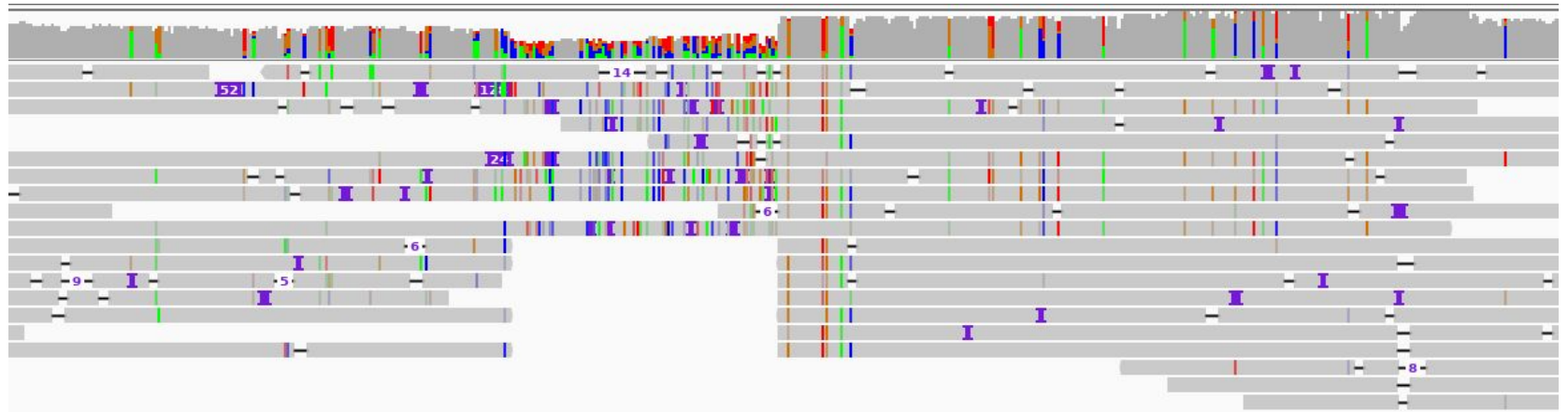
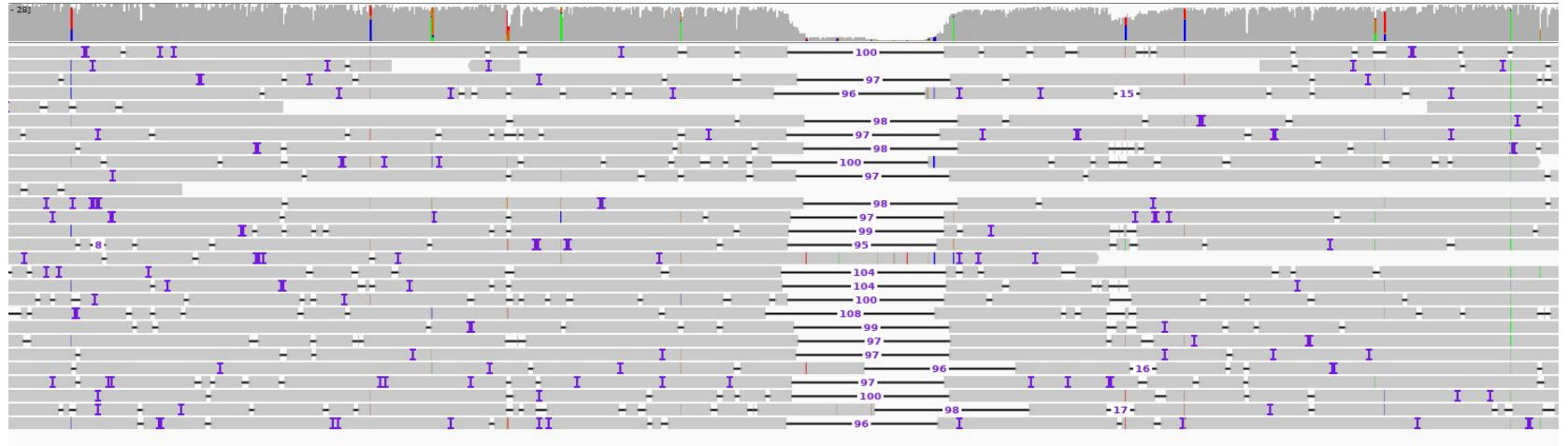




NanoQC

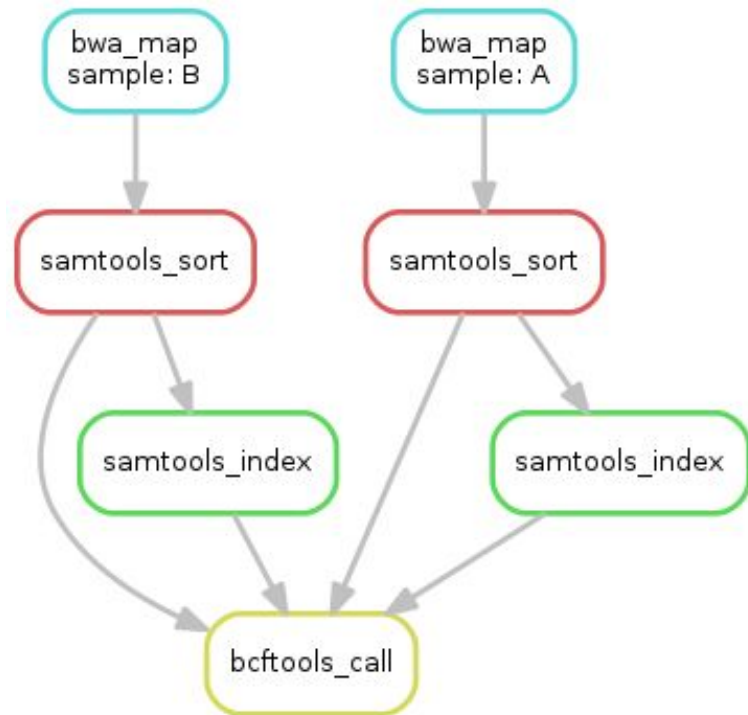


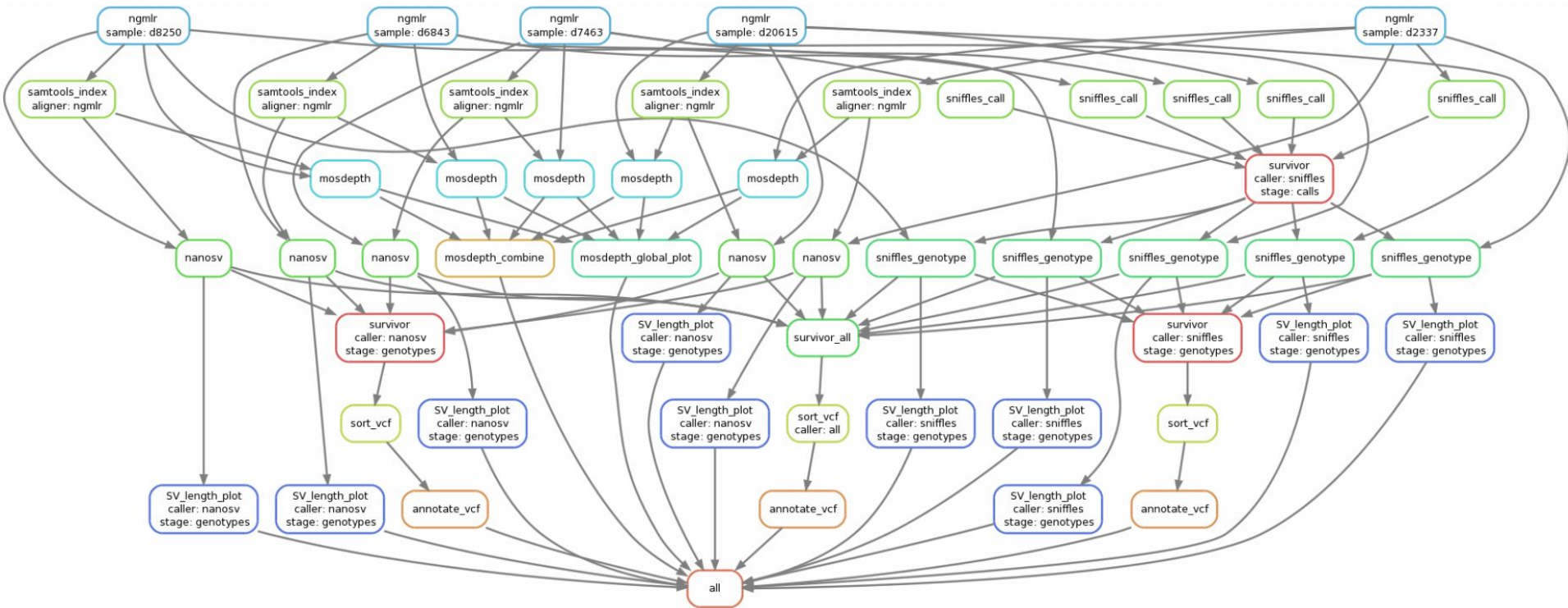
Structural variation



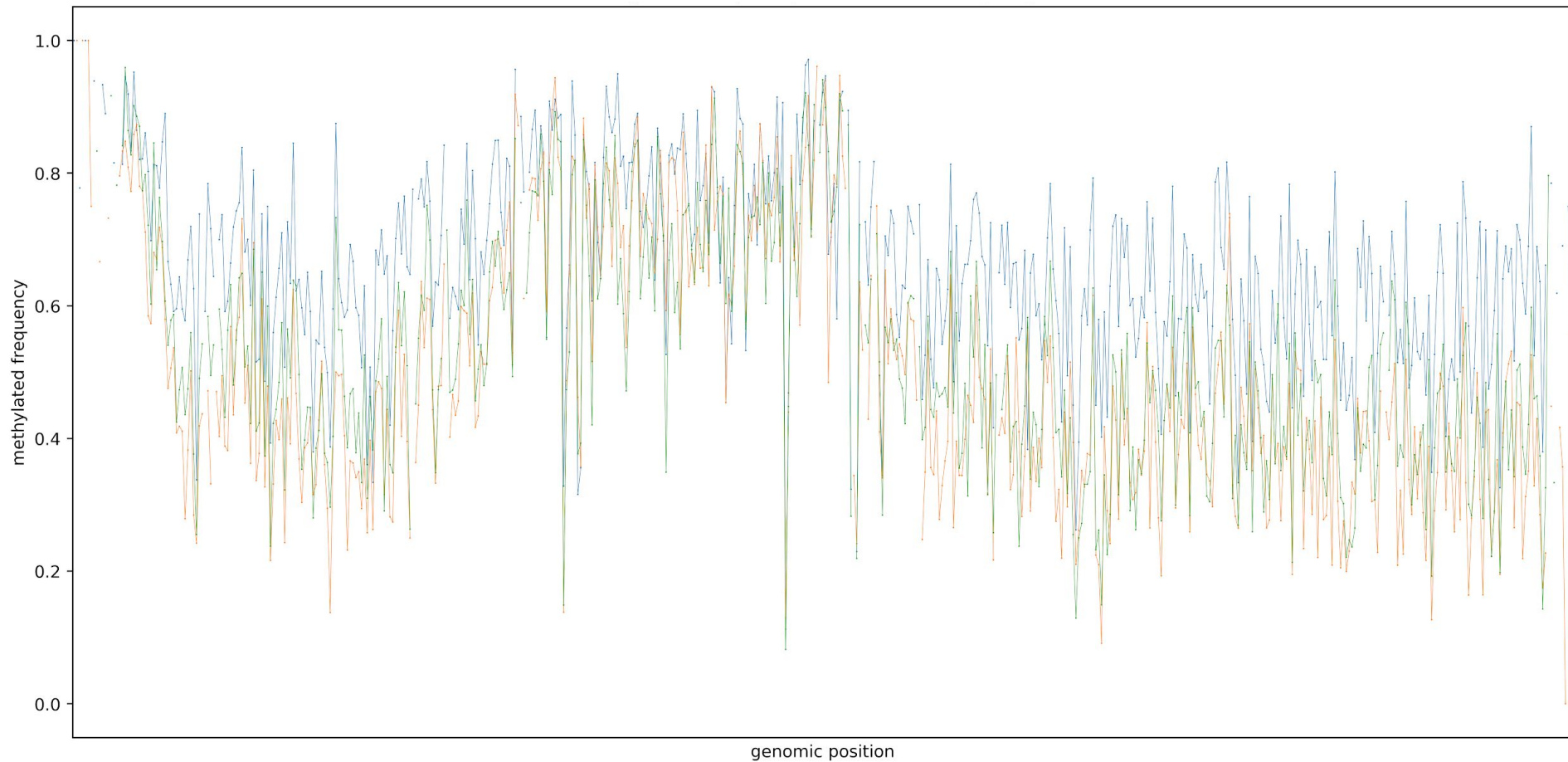
Structural variation

- Snakemake workflow
 - NGMLR + Sniffles
 - NanoSV
 - Mosdepth
- <https://github.com/wdecoster/nano-snakemake>

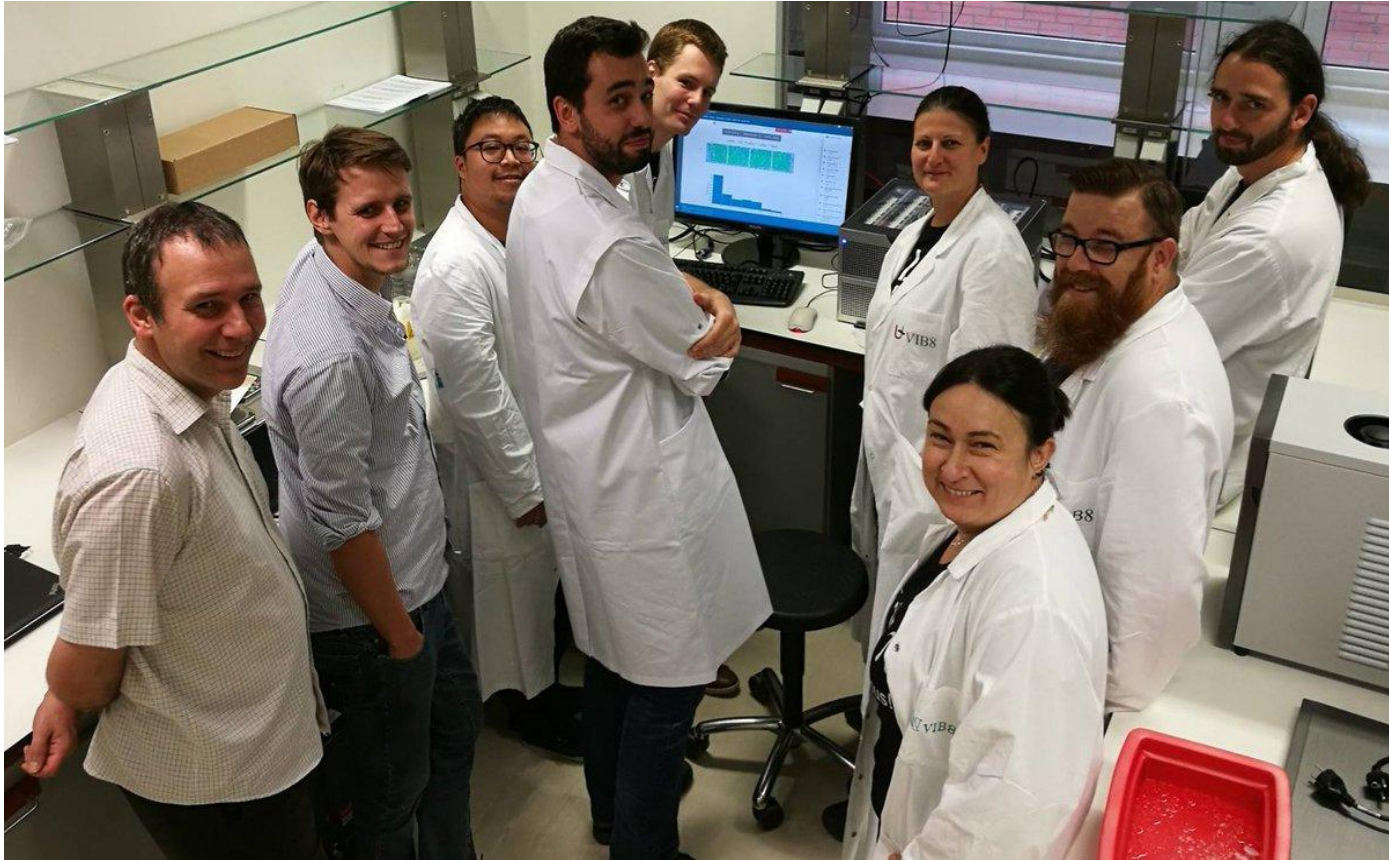




Nucleotide modifications



Acknowledgments



Acknowledgments



VIB
GENOMICS
CORE

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BIOCONDA

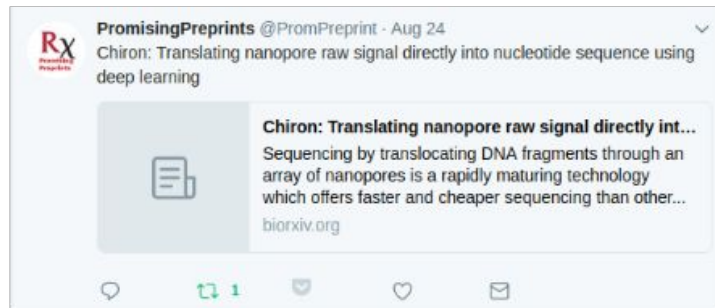
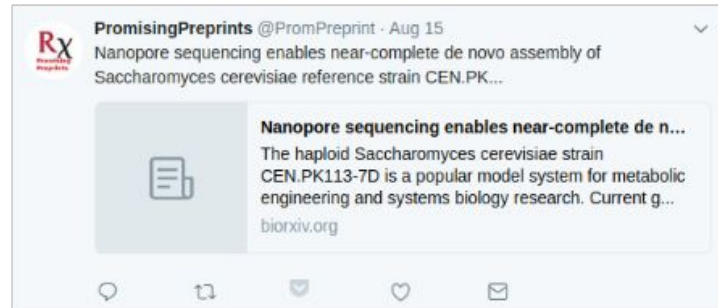
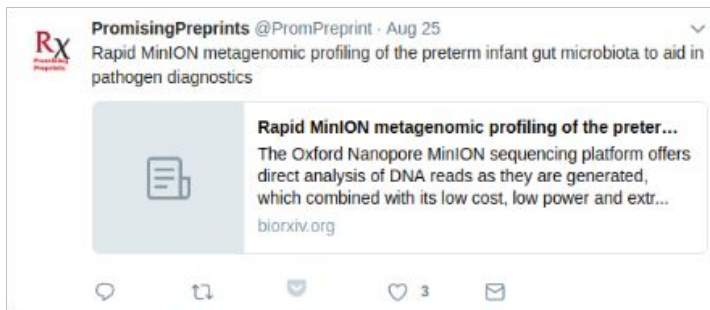


stackoverflow



Twitter bot Promising Preprints

- @PromPreprint
- Top 10% altmetric score for bioRxiv





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<https://gigabaseorgigabyte.wordpress.com>



<https://github.com/wdecoster>