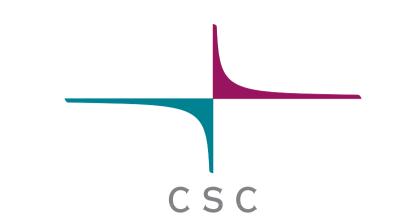
## Scripting for large-scale sequencing based on Hadoop



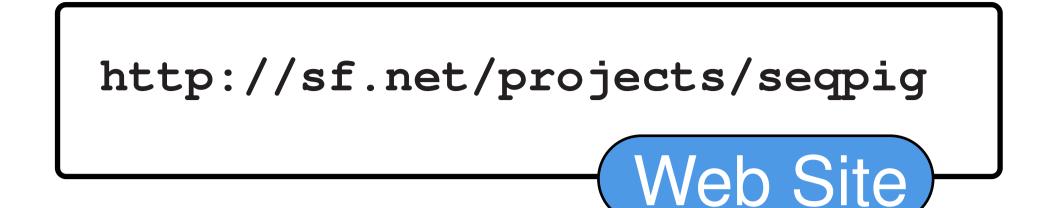
André Schumacher and Keijo Heljanko firstname.lastname@aalto.fi



Luca Pireddu and Gianluigi Zanetti firstname.lastname@crs4.it



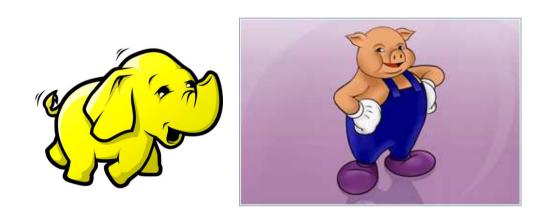
Aleksi Kallio and Eija Korpelainen firstname.lastname@csc.fi



• SeqPig is a tool that facilitates using the Hadoop distributed computing framework to analyse and manipulate NGS data

Data sizes are increasing faster than processing power and disk read speed
—SeqPig on Hadoop can help scale the throughput of analysis workflows
An add-on to Apache Pig (http://pig.apache.org); scripts are written in *Pig Latin*Scripts are automatically run as parallel computing jobs
Ideal for computing statistics, filtering, projecting and reformatting data
Supports interactive analysis through its powerful command-line interface
Parallel scripting environment opens up exciting possibilities for scalable analysis pipelines with a short development cycle





- Hadoop is the open-source reference implementation of MapReduce
  Widely used in data-intensive industries
- -e.g., Yahoo!, Facebook, Twitter, LinkedIn, etc.
- MapReduce allows the scalable processing of large amounts of data
- Automatically handles splitting/moving data and hardware failures

SeqPig extends Pig with a number of features for processing sequencing data:

- data input and output components
  - import and export functions for: Fastq, Qseq, SAM and BAM
- extract fields and transform HTS data
  - so you can handle sequencing data like native Pig data (e.g., process SAM flags)
- a collection of scripts and custom functions for frequent tasks
- implement speed-optimized or simplified task-specific functions
- Pig is a scripting language and execution engine built on top of Hadoop
- Simplifies the use of Hadoop through its concise SQL-like logic

## Hadoop and Pig

• SeqPig has been tested on Amazon's Elastic MapReduce service

- Users may rent computing time on the cloud to run their SeqPig scripts
- One can also share data in S3 storage buckets with other cloudenabled software



StatisticSpeedupRead length distribution28.8Avg. read quality distribution28.8Avg. base quality distribution28.0GC-content distribution8.3Per-cycle base and base quality28.0All of the above6.9

Data size: 61.42 GB; 16-slave Hadoop cluster vs 1 node for FastQC

Example: speedup over FastQC

Convert qseq into fastq: reads = LOAD 'in.qseq' usingQseqUDFLoader(); STORE reads INTO 'out.fastq' using FastqUDFStorer();

Compute a histogram of GC content: reads\_by\_bases = FOREACH reads GENERATE UnalignedReadSplit(sequence, quality); read\_gc = FOREACH reads\_by\_bases { only\_gc = FILTER \$0 BY readbase == 'G' OR readbase == 'C'; GENERATE COUNT(only\_gc) as count; } read\_gc\_counts = FOREACH (GROUP read\_gc BY count) GENERATE group as gc\_count, COUNT\_STAR(\$1) as count; STORE read\_gc\_counts INTO 'GC\_count\_histogram.txt'; Compute a histogram of read lenghts: read\_len = FOREACH reads GENERATE STRLEN(sequence); read\_len\_counts = FOREACH (GROUP read\_len BY \$0) GENERATE group AS len, COUNT\_STAR(\$1) as count; STORE read\_len\_counts INTO 'read\_len\_histogram.txt';

Compute a histogram of base composition and base qualities:

read\_seq\_qul = FOREACH reads GENERATE sequence, quality; base\_qual\_counts = FOREACH (GROUP read\_seq\_qual ALL) GENERATE BaseCounts(\$1.\$0), BaseQualCounts(\$1.\$1); formatted\_base\_qual\_counts = FOREACH base\_qual\_counts GENERATE FormatBaseCounts(\$0), FormatBaseQualCounts(\$1);



Seal

• Niemenmaa M, Kallio A, Schumacher A, Klemel P, Korpelainen E, and Heljanko K. (2012) Hadoop-BAM: directly manipulating next generation sequencing data in the cloud. Bioinformatics 28(6):876-877. http://hadoop-bam.sf.net

• Pireddu,L., Leo,S. and Zanetti,G. (2011). SEAL: a Distributed Short Read Mapping and Duplicate Removal Tool. Bioinformatics. http://biodoop-seal.sf.net

• Andrews S., et al. FastQC website: http://www.bioinformatics.babraham.ac.uk/projects/fastqc

