Distributed Genome Preprocessing

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Abstract

- Genome preprocessing – analyzing and manipulating short read data in order to improve downstream assembly
- Hadoop – A distributed computing framework that scales to an arbitrary number of nodes
- Counting *k*-mers, substrings of reads, in order to determine read reliability
- Remove reads with low *k*-mer counts from data
- Testing shows that our approach is scalable and effective

Preprocessing

- Short read data straight from a sequencer is unreliable

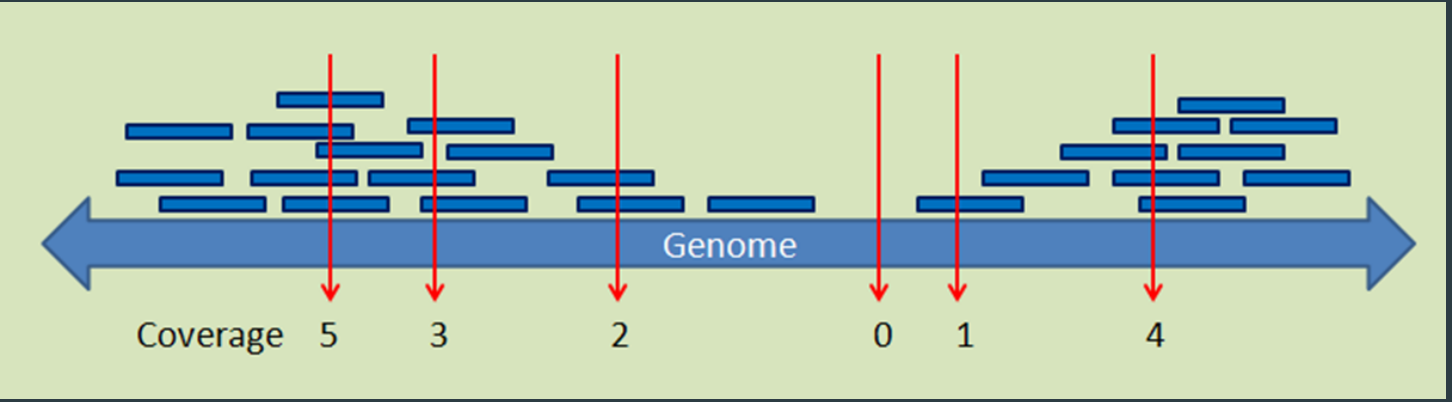
```
@SRR067577.10004404/2
CCATAGATGCCAGAATCTATCCCTGCCCTCGGCGTGAGACCTCTGCTGGGGAACTGGTGTTTGCCGTATCAGACCACAGCAAGGTGTCGTAATGGAGTCTC
+
IIIIIIIIIGIIIGIHHIIGIIIHIHIIGDGEBGBIIIIHIHHGIHBHE<EDIEDDD=DBA@DD@FBEBDDDBEBEDB@FDB>B4?08?;?;>?>B@9D##
@SRR067577.10029246/1
CTGAATCCCTTTTTCCATGGATAGCTTTCTGGTAGACCATAAATGAAAGCATGGCCACAGCATCTTGGCAGCCAGCACAGACCCATGGAACTCTTGGCCAG
+
HHGHGHHHHGHHHDHGHHEB<FBEG<BGGBDDAGFHHHBHHHHGBDD3GDGEBF@HHHBHGDDF<D>DGBDFFEFFBEEHCDF8E38?=>DADA#######
```

Examples of read data. Each read is four lines long. The lines are as follows: Identifier, Read, Optional Description, Quality Score

- Two major approaches: trimming, error correction
- Goal is to improve average quality of reads used

Methods

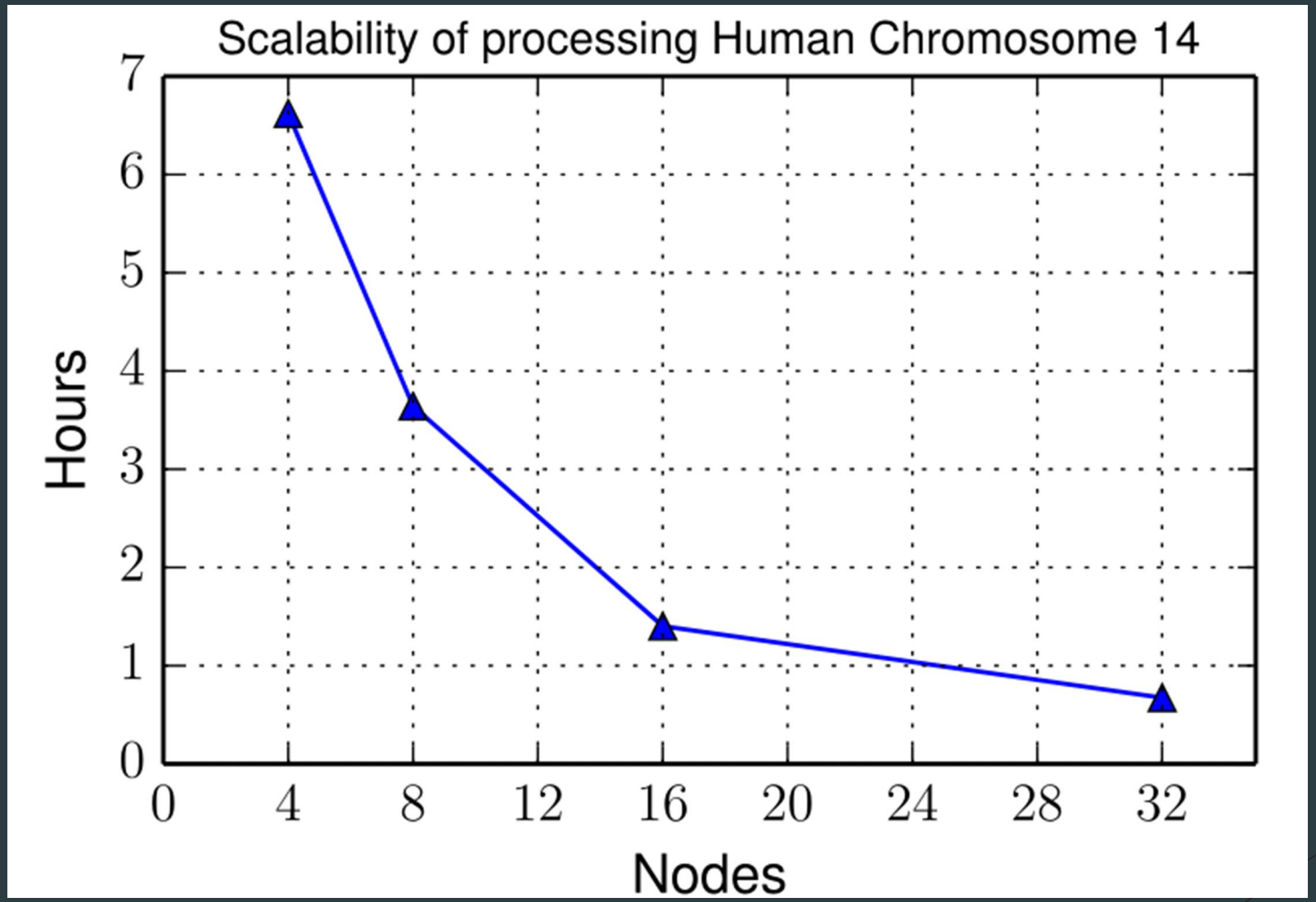
- Count *k*-mers and filter based on *k*-mer frequency



- Four steps:
  1. Count *k*-mers
  2. Reorganize by read identifiers
  3. Filter reads based on their *k*-mer coverage
  4. Apply the filter to the original read data

Results

- Tested scalability by timing process on human chromosome 14 data set and doubling nodes used: 4, 8, 16, 32
- Tested accuracy by using Velvet assembler to determine assembly quality before and after preprocessing.


Scalability of processing Human Chromosome 14
Hours
Nodes
0
1
2
3
4
5
6
7
0
4
8
12
16
20
24
28
32

Assembly of Read Data		
	Unfiltered	Filtered
Nodes in Graph	4419219	2792604
N50	249	599
Max Length	6760	9524

Other Work

- Implemented other trimmers:
  - Average quality filter
  - Sliding window filter
  - *Q*-mer counter
- Started writing simple Spark scripts for *k*-mer analysis

References

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