ABUMPIMP 2024: The 2nd Minisymposium on Applications and Benefits of UPMEM commercial Massively Parallel Processing-In-Memory Platform
Full day minisymposium at EURO-PAR 2024

Compression of Human Genomic Data

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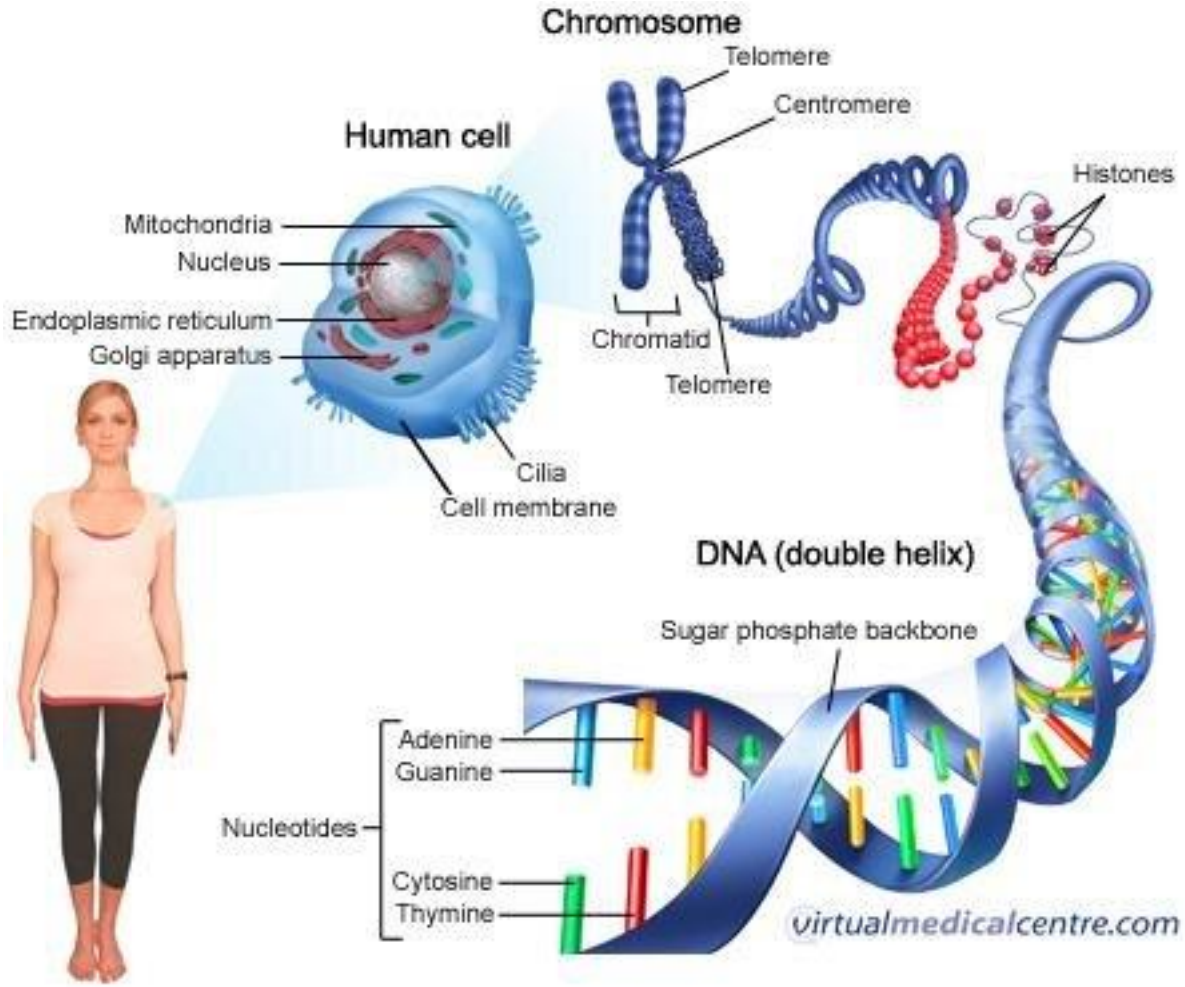
Human Genome

Human Genome

- 23 pairs of chromosomes / cell
- 3.2 x 10^9 bp

Each genome is different

- Many small variations
  - SNPs (Single Nucleotide Polymorphism)
  - Micro-satellites
  - …
- A few Large variations



Sequencing

DNA extraction

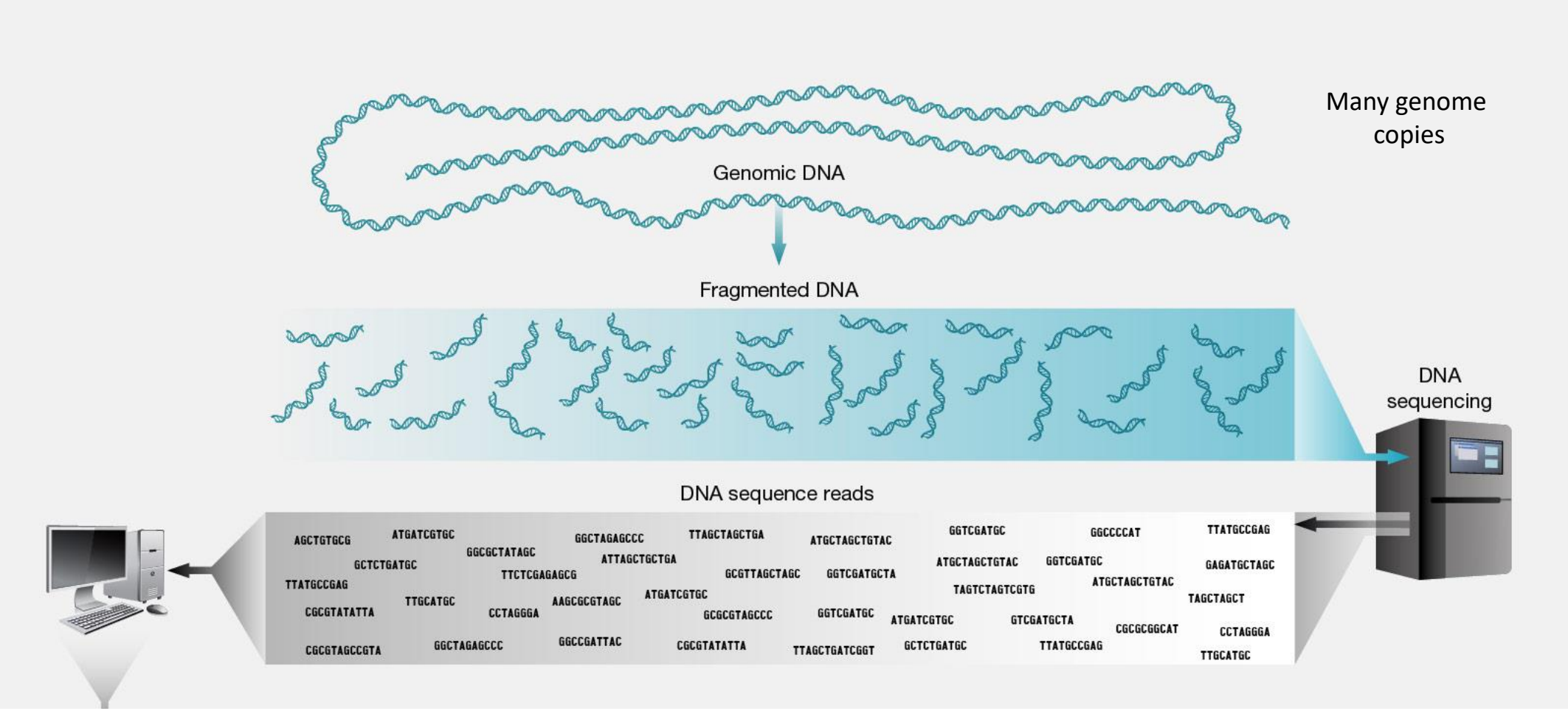
Illumina sequencer

Raw data

```
ATGGATAGG
ACCAGGATT
AGAGGACCA
GAGGAGAGT
ATAGTACCA
TTTATTTTA
```

300 – 500 GBytes

Sequencing principle



Genomic Data

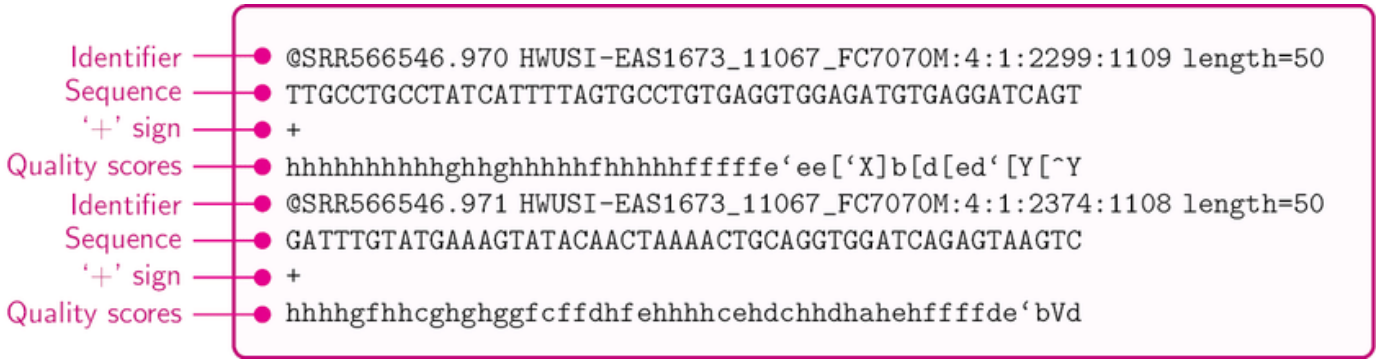
Short reads

- Length: ~300 bp
- Error rate: ~0.1%
- Technology: Illumina

Long reads

- Length: 10Kbp to 100Kbp
- Error rate: 1%-3%
- Technology: PacBio, ONT

FASTA / FASTQ format → text file

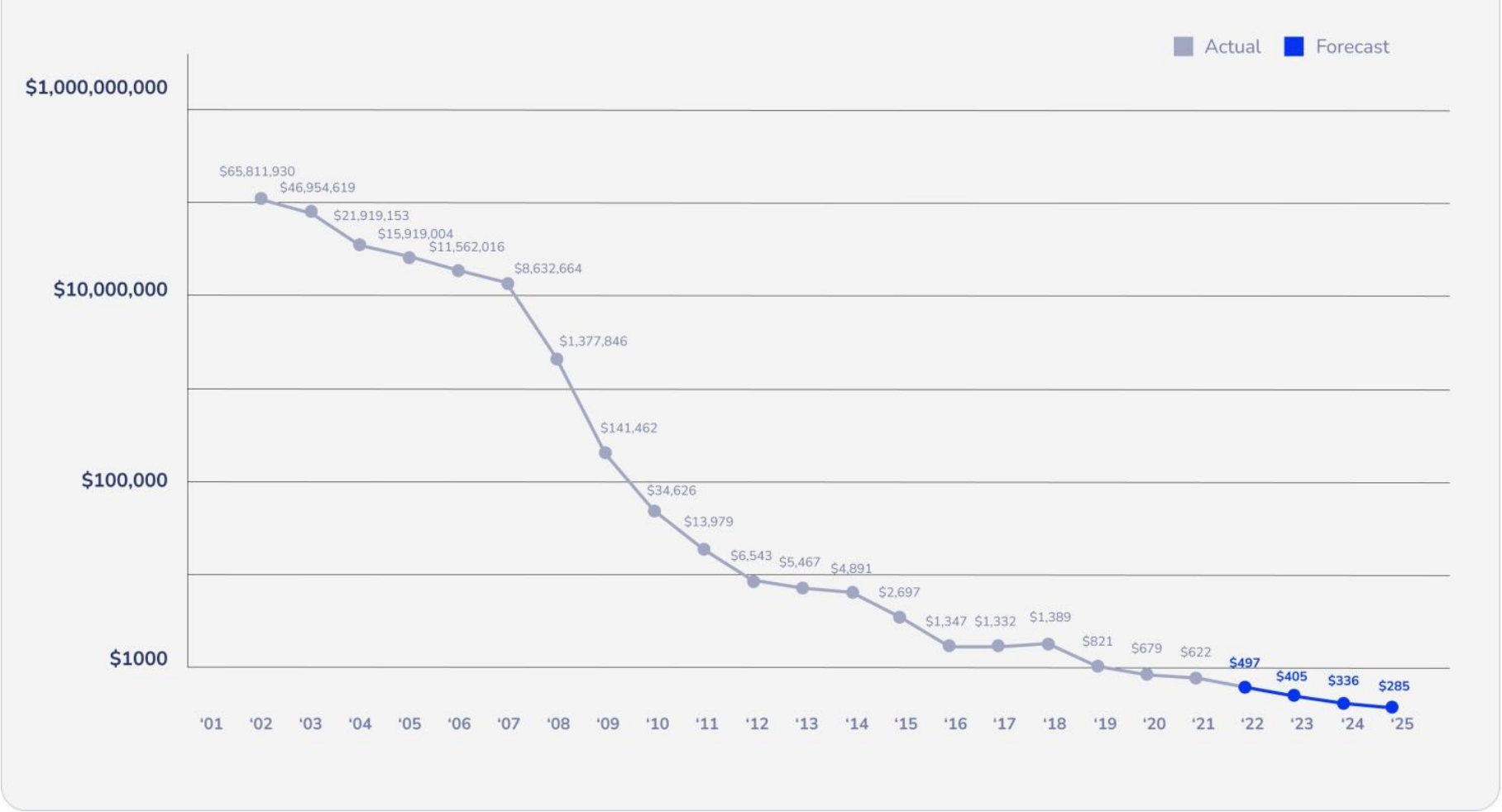


FASTQ

Human Genome

- Coverage: 30X to 50X
- 10^9 Reads: 2x150 bp
- 300 GB to 500 GB

Human genome sequencing cost



https://www.linkedin.com/pulse/cost-sequencing-human-genome-fall-over-50-2025-3billionpairs/

Genomic data compression

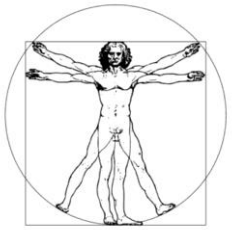
Lossless

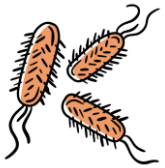
Without reference

- Standard tools (gzip,bzip2,…)
- Genomic dedicated tools

With reference

- Genozip
  - currently the best tool (fast, good ratio)
  - used as reference





Compression with reference

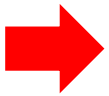
Human Genome Reference: 3.2 x 10^9 nt

TTGAGACCAGATTAGAGACCAGGGGAGAGACCCAGGGAGTTA**CATACCATGACCAGGGGATT**TAGGGATTCCGATATAGGACGCCATTACCAGTGACCAGGATAGCCAGGTTAGA…

CATACCATGACCAGGGGATTTAGGGAAT
GGTAGGACCAGGGATTGGGATTAGGAGA
TGGACCAGGATAGGACCAGATAACAGTA
GTCCACATACCATAGGACCACTTACCAT
...

Short reads

```
42 (S,27,A)
10988
1970 (D,13)
7892
...
```

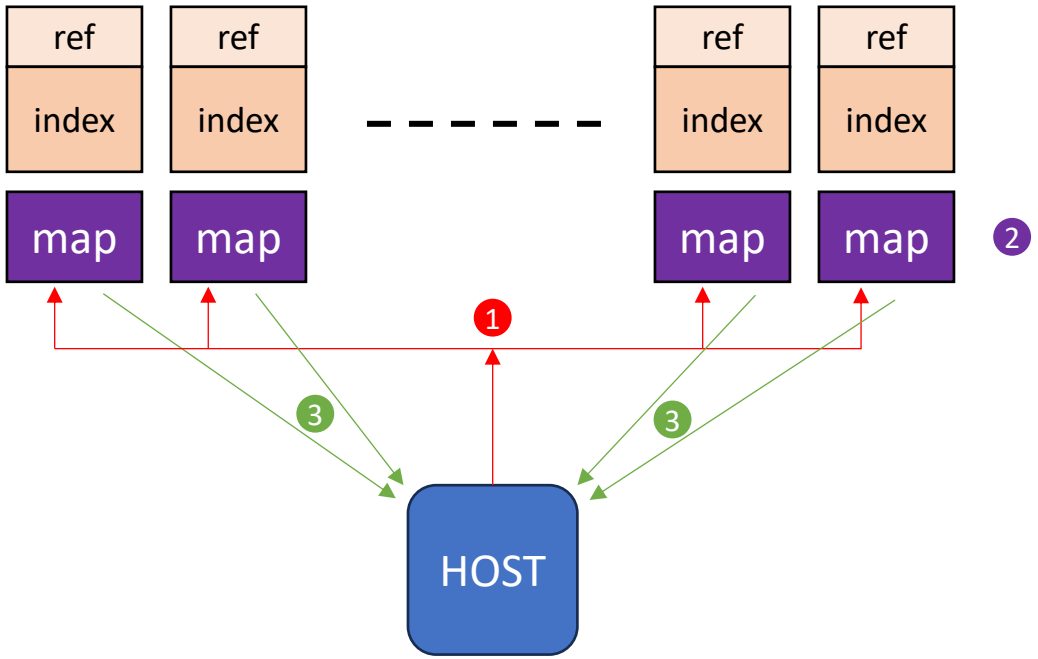


Mapping problem: find the best match as fast as possible

UPMEM PiM implementation

MRAM
Reference genome
+ index

DPU



The reference is distributed over the DPUs

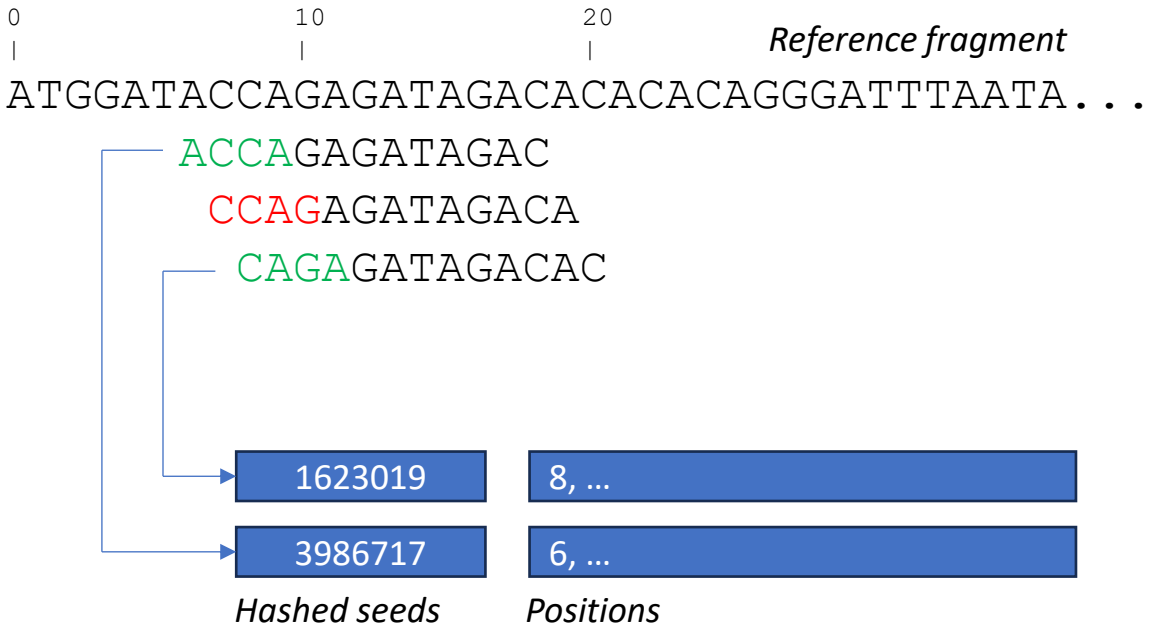
- Each DPU as a fraction of the Human Genome in its MRAM
- An index is built to speed-up the mapping

Broadcast reads to DPUs 1

Mapping on DPUs 2

DPUs sent back match (if any) 3

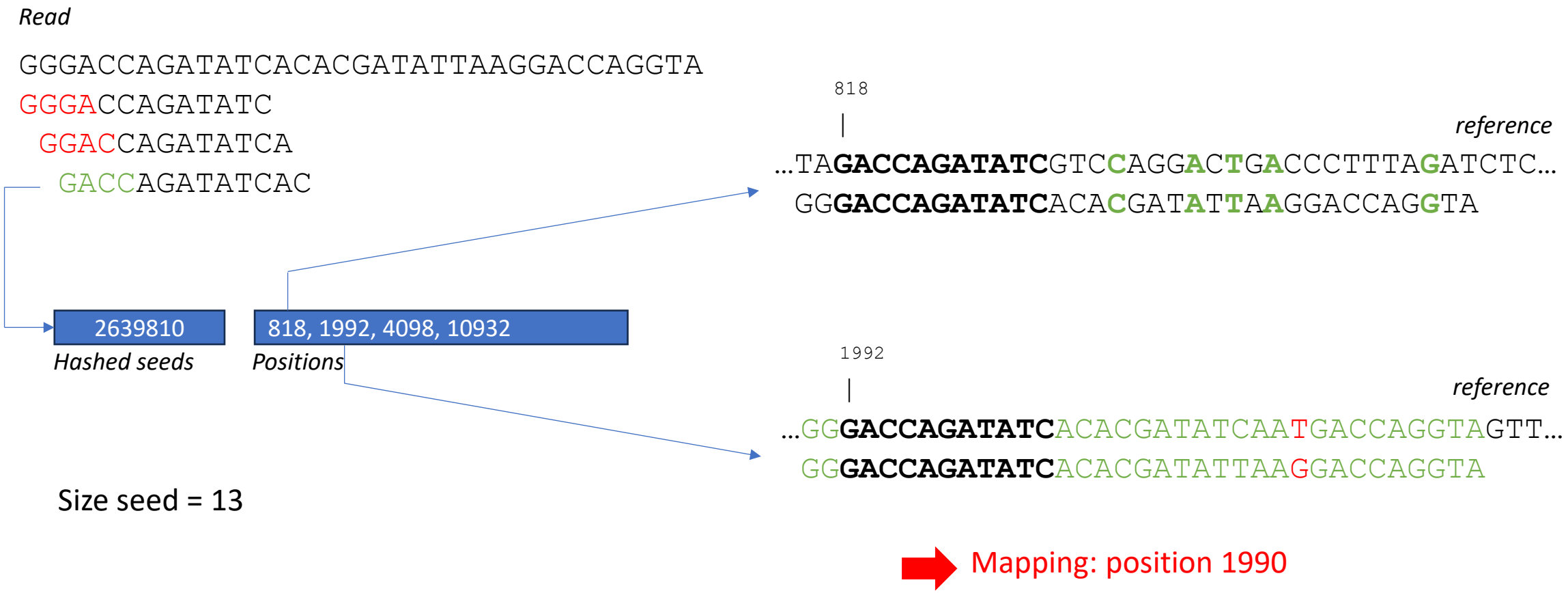
Reference index



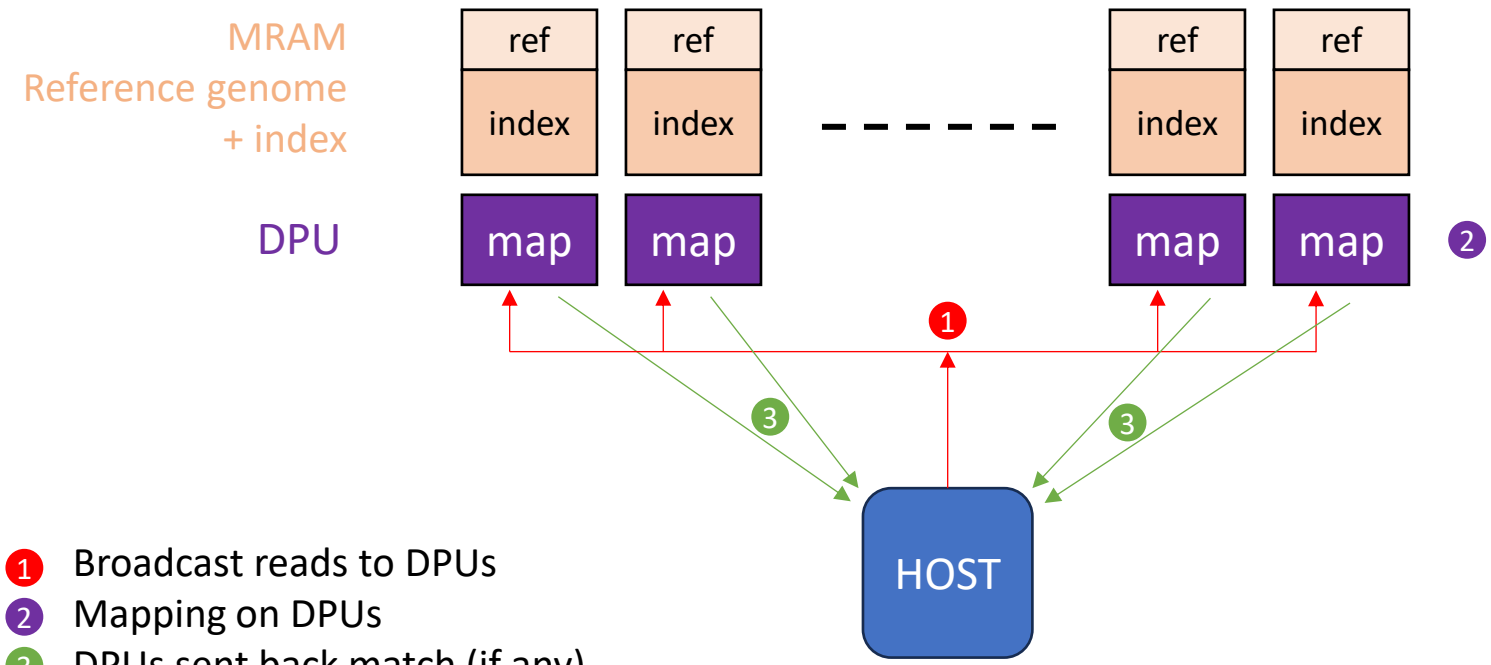
Each DPU indexes its reference fragment by iterating 13-mers (called seeds) and inserting their positions in a table

Low-interest seeds are filtered out

Mapping


Read
GGGACCAGATATCACACGATATTAAGGACCAGGTA
GGGACCAGATATC
GGACCAGATATCA
GACCAGATATCAC
2639810
Hashed seeds
818, 1992, 4098, 10932
Positions
818
reference
…TAGACCAGATATCGTCCAGGACTGACCCTTTAGATCTC…
GGGACCAGATATCACACGATATTAAGGACCAGGTA
1992
reference
…GGGACCAGATATCACACGATATCAATGACCAGGTAGTT…
GGGACCAGATATCACACGATATTAAGGACCAGGTA
Size seed = 13
Mapping: position 1990

Implementation limitation



1. Broadcast reads to DPUs
2. Mapping on DPUs
3. DPUs sent back match (if any)

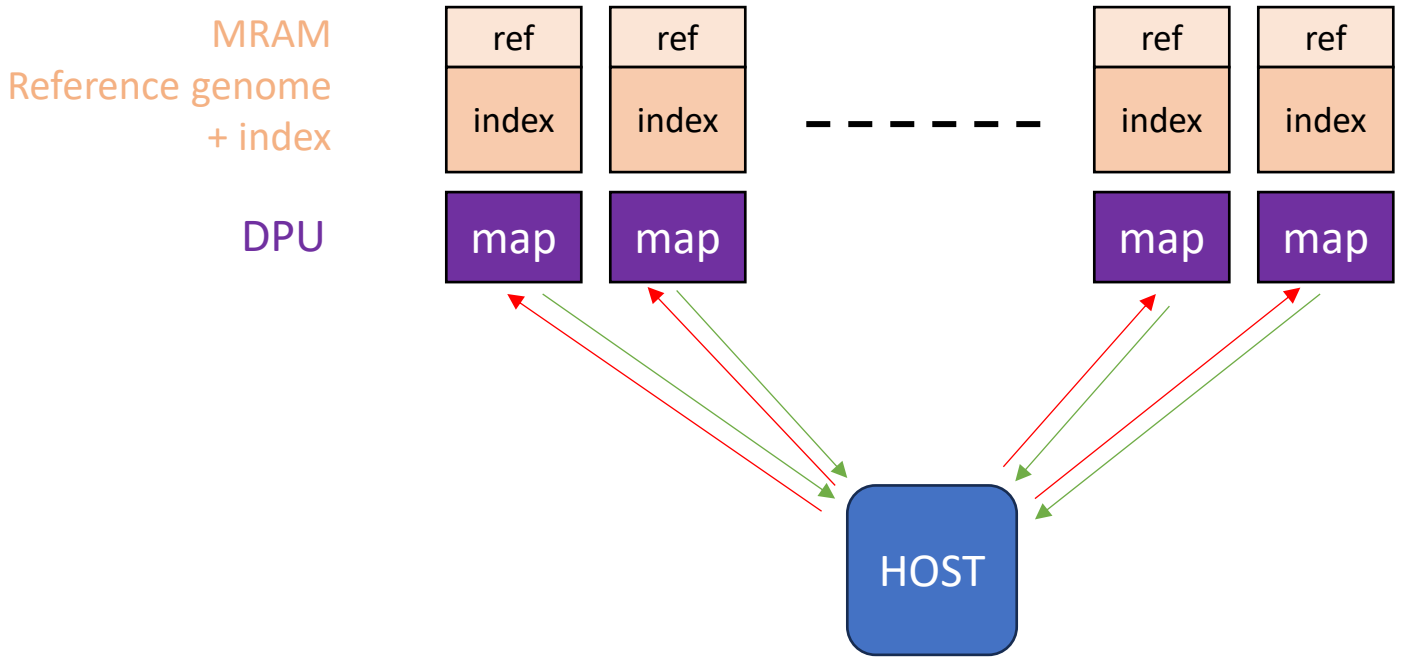
All reads send to all DPUs

- High data transfer
- High DPU workload

Only a few DPUs send back mapping positions

Read dispatcher

Instead of broadcasting all reads to all DPU, send reads selectively to some DPUs



Target reads to right DPUs 1

Mapping on DPUs 2

DPUs sent back match (if any) 3

Advantages
- Less data transfer
- Faster mapping

Problem
- Find good criteria to target the right DPUs

Bloom filter

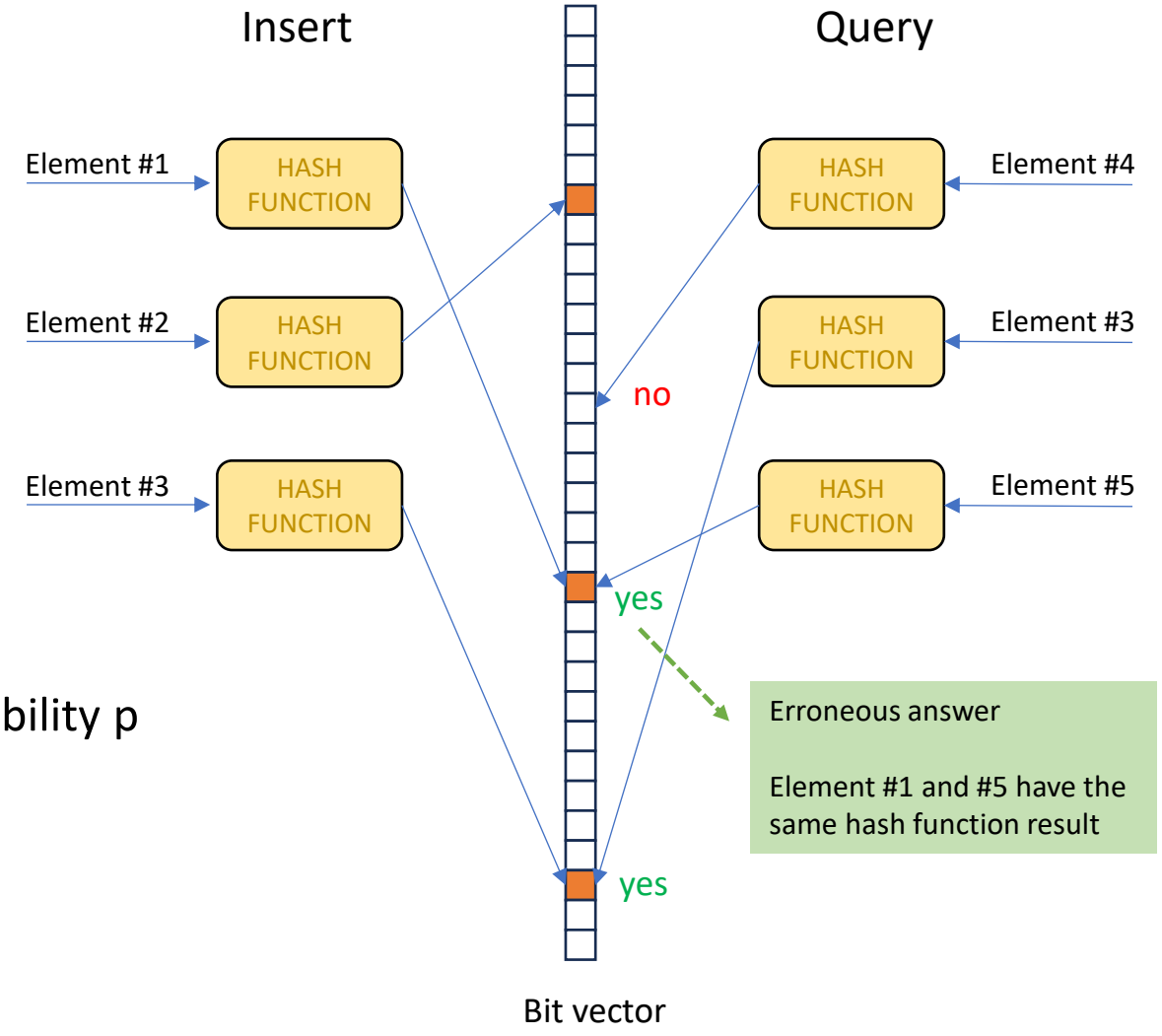
Probabilistic data structure

Two operations

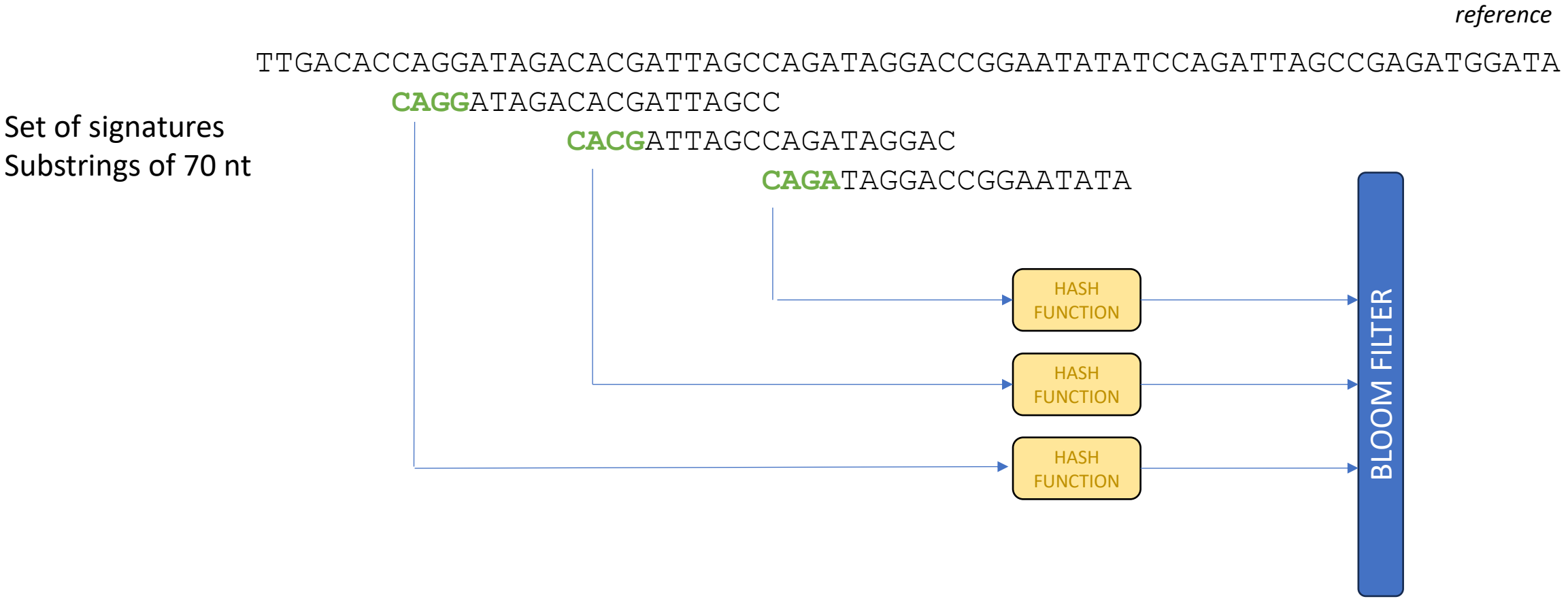
- Insert an element
- Query the presence of an element
  - no → the element is absent
  - yes → the element is present with a probability p

Compact data structure

- Vector of N bits
- Hash function returns a value in [0,N-1]



Bloom filter associated to a DPU



Bloom filter based dispatcher

The bloom filter is located on the host

Experiment

- Server:
  - Intel Xeon Silver 4215 @ 2.5 GHz
  - 256 GB of DDR4, 20 UPMEM DIMMS @ 350 MHz (160 GB, 2560 DPUs)
- Data sets

	Plain file size (GB)	Read count	Read size	Year
Genome GRCh38	3.2	1	3.1 Gbp	2013
Reads H1 (SRR14724532)*				
FASTQ	2x 121.1	356 M	151 bp	2021
FASTA	2x 66.3			
Reads H2 (SRR14724533)†				
FASTQ	2x 122.9	361 M	151 bp	2021
FASTA	2x 67.3			

* https://www.ebi.ac.uk/ena/browser/view/SRR14724532
† https://www.ebi.ac.uk/ena/browser/view/SRR14724533

reference

Sequencing data

Bloom filter dispatcher efficiency

8 ranks (512 DPUs)

- Reference: 6.25 x 10^6 nt / DPU (MRAM full)

Size of the Bloom filter: ~ 10 GBytes

On average, each read is send to ~2 DPUs → High efficiency

Host bottleneck

- DPUs are idle ~ 40% of the time

Compression ratio

Software	% of original size FASTQ	FASTA	
UPMEM PIM	5.52	5.05	
Genozip –e	4.08	18.46	
Genozip –e fast	5.27	18.49	*fast mode*
Genozip –e @16	4.09	18.56	*16 threads*
Genozip –e fast @16	5.32	18.61	

Compression time

Current implementation uses 8 ranks (512 DPUs)

Software	Compression time (s)	
	Elapsed	CPU
UPMEM PiM	🏆 188	🏆 2251
genozip -e	310	9546
genozip -e -fast	227	7000
genozip -e -@16	510	8151
genozip -e -@16 -fast	362	5680

Energy consumption

Software	Energy consumption (kJoules)			
	DRAM*	PKG*	DPUs†	**Total**
UPMEM PiM	9.12	20.37	7.16	🏆 36.65
genozip -e	15.38	41.66	N/A	57.04
genozip -e -fast	11.52	31.89	N/A	43.41
genozip -e -@16	25.26	61.70	N/A	86.96
genozip -e -@16 -fast	18.52	44.60	N/A	63.12

* Measured with Intel® RAPL
† Estimated with execution time

Conclusion

Sequencing the human genome is becoming inexpensive

- Use in personalized medicine
- Medical data that needs to be kept for several decades
- Efficient storage needed (space / time / energy)

PiM implementation of genomic data compression

- relieves server load
- reduces power consumption

Many other genomic algorithms benefit from PiM accelerators

- BioPiM project: https://biopim.eu/
- GenoPiM project: https://genopim.irisa.fr/

Thank you for your attention

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