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Massively Parallel Mapping of Next Generation Sequence Reads Using



Motivation

• DNA sequence alignment problem is a character-level comparison of DNA sequences obtained from one or more samples against a database of reference genome sequence of the same or a similar species.

• Huge computational burden due to the comparison of >1 billion short (100 characters, or base pairs) "reads" against a very long (3 billion base pairs) reference genome. • Requires 30-50 CPU days for mapping & alignment • >1 million whole human genomes by the end of 2017! • Clinical sequencing in trials in the US • Genome sequencing as a routine test at hospitals • We need very fast, accurate, and low-cost analysis methods





Test genome



Reference Genome (HGP 3.2 billions)

Whole Genome Sequencing





Paired-end sequencing

Read mapping

th	
60	
en	
Jd	
G	





Reference length

		∞	∞	∞	∞	∞	∞	∞	∞
			8	8	8	8	8	8	8
			Α	8	8	8	8	8	8
		Α	В	С	8	8	8	8	8
8	Α	В	C			8	8	8	8
8	8	C					8	8	8
8	8	8			· · · · · · · · · · · · · · · · · · ·			8	8
8	8	8	8			·····			8
8	8	8	8	8			and a second second		

Edit distance



• Develop and implement a GPGPU-friendly algorithms to map DNA sequence reads to the reference genome



• Implementation using the CUDA (Compute Unified Device Architecture) platform, and testing using the NVIDIA Tesla K20 GPGPU processors.

• Take advantage of the embarrassingly parallel nature of the problem to concurrently align millions of read vs reference pairs





			-	Α	С	Α	С	Α	С	Т	Α	G	Τ	Α
		-	0	9	0	0	0	0						
		Α	0	1		-1	-1	1	0					
		G	0	0	0		-2	-2	0	-1				
		С	0	-1	1	0		-2	-1	-1	-2			
		Α	0	1	0	2	1	-0	-1	-2	0	-1		
		С		0	2	1	3	2		0	-1	-1	-2	
		Α			1	3	2	4	3	2	1	0	-1	-1
		С				2	4	3	5	4	\rightarrow	2	1	0
		Α					3	5	4	4	5	-1	3	2
		Т						4	4	5	4	4		4
		Δ							3	4	6	5	4	6
		G								2	5	7	6	5



Reference length









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e, e



length Read



Reference length

	-	Α	С	Α	С	Α	С	Τ	Α	G	Τ	Α	
-	0	0	0	0	0	0							
Α	0	1	0	-1	-1	1	0						
G	0	0	0	-1	-2	-2	0	-1					
С	0	-1	1	0	-1	-2	-1	-1	-2				
Α	0	1	0	2	1	0	-1	-2	0	-1			
С		0	2	1	3	2	1	0	-1	-1	-2		
Α			1	3	2	4	3	2	1	0	-1	-1	1
С				2	4	3	5	4	3	2	1	0	
Α					3	5	4	4	5	4	3	2	
Т						4	4	5	4	4	5	4	
Α							3	4	6	5	4	6	
G								3	5	7	6	5	
											-		



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- architectures.

- mapping applications.

Contributions

simultaneous alignments.

characteristics of the GPGPU.

the maximum allowed error threshold set by the user.

reference genome size and error allowance. data to the GPU global memory in the initialization step. based post processing all together, except for I/O operations.

- Map a time-consuming application to massively parallel GPU
- Move the compute-intense parallel verification step to the GPGPUs. • Collect the reads in a buffer, then pass to the GPGPU for millions of
- Determine the number of alignments automatically by considering the
- Adjust the number of threads used per alignment dynamically based on
- Ability to be merged with any existing and future hash-table based read
- Ability to be used for various configurations like different read sizes,
- Reduce host to GPU transfer time significantly by placing all relevant
- Develop dynamic programming backtracking in GPU, bypassing CPU-





							10118											
		_	Α	С	Α	С	Α	С	T	Α	G	Τ	Α					
	-	0	0	0	0	0	0											
	Α	0	1	0	-1	-1	1	0										
	G	0	0	Ŷ	-1	-2	-2	0	-1									
	С	0	-1	1		-1	-2	-1	-1	-2								
	Α	0	1	0	2		0	-1	-2	0	-1							
	С		0	2	1	3		1	0	-1	-1	-2						
	Α			1	3	2	4		2	1	0	· -1	-1		Α	- C	Α	C /
	С				2	4	3	5		3	2	1 `\	. 0		Α (G C	Α	С
	Α					3	5	4	4		4	3	2					
	Τ						4	4	5	4		5	4.			4		1
	Α							3	4	6	5		6	· <u> </u>	_		-	-1
	G								3	5	7	6				1		0
I																		

Re	Read length + Reference length / /																					
0	0	1	0	0	-1	-1	-2	0	-1	-1	-2	-1	/1	0								
0	0	-1	-1	-2	-2	-2	-1	-1	-2	0	-1	1	0	2	1	3	2	4				
0	0	1	0	0	1	0	-1	1	0	2	1	3	2	4	3	5	4	4	5	4	6	5
0	0	0	-1	1	0	2	1	3	2	4	3	5	4	4	5	4	6	5	7	6		
0	0	1	0	2	1	3	2	4	3	5	4	4	3	4	3	5						



Reference length



ACTAGTA-AC-A-TAG





	500
	450
	400
	350
<u>d</u>	300
oeear	250
7	200
	150
	100
	50
	0

)	2,000,000	4,000,000	6,000,000	8
			Number o	of t







Thank You



