Deploying deep neural network with ONNX for efficient genome analysis with nanopore sequencing

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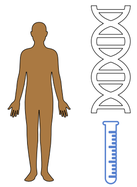
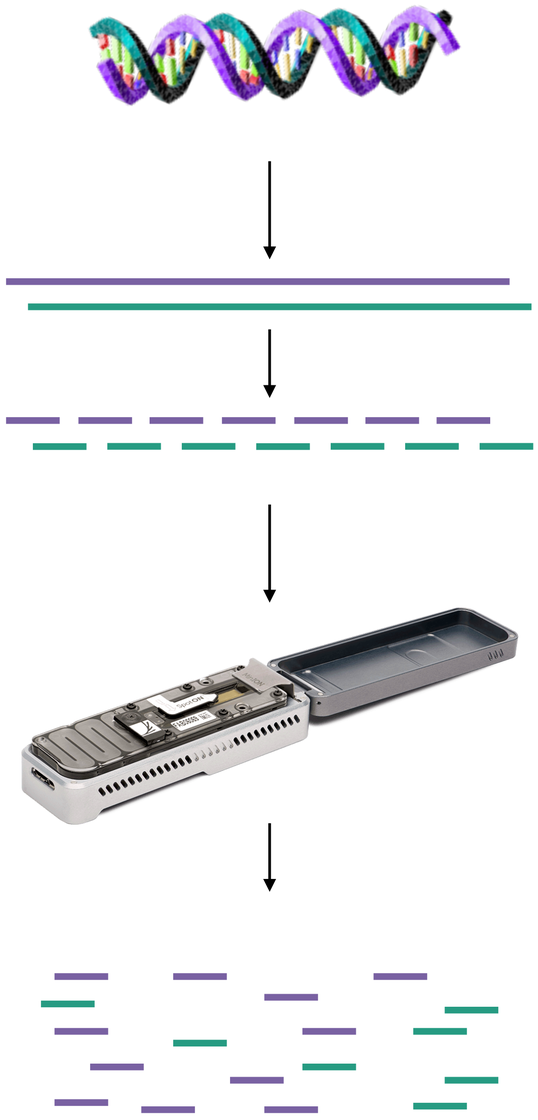
Overview of genome analysis



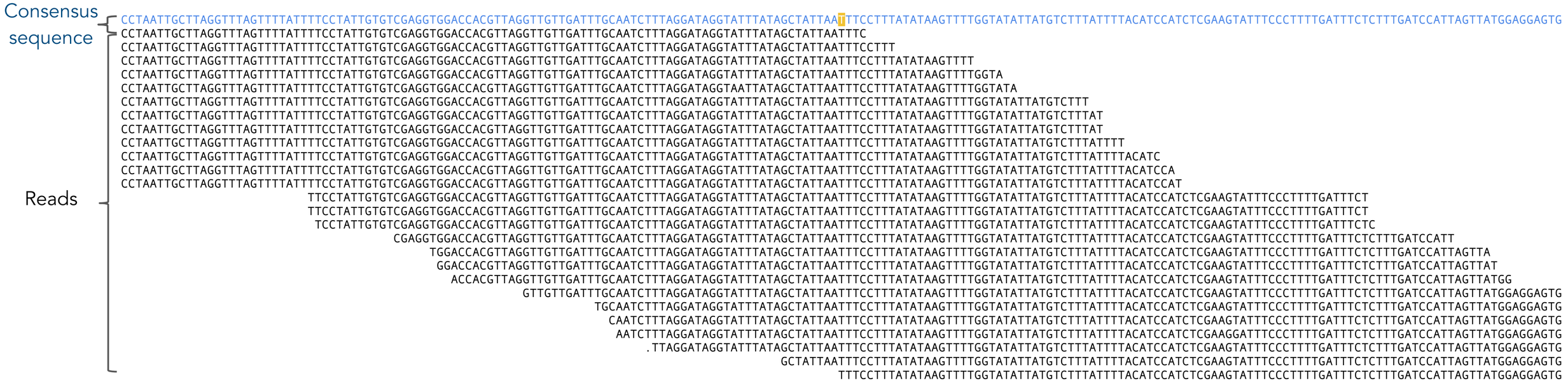
Sample collection

Library Preparation



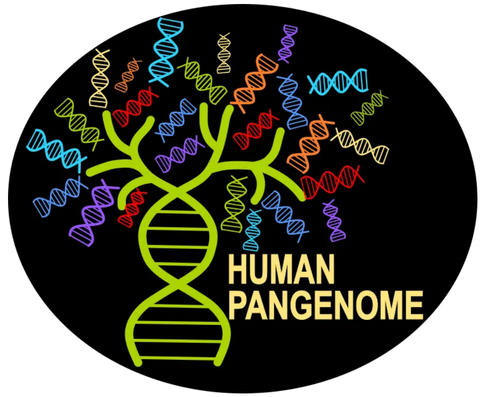
ACGTTACGTTATTCAGTTT

de novo assembly



Global effort

T2T
TELOMERE-TO-TELOMERE CONSORTIUM



Human Pangenome Reference Consortium

VGP
VERTEBRATE
GENOMES
PROJECT
A PROJECT OF THE G10K CONSORTIUM


Darwin
TREE
of
LIFE

Nanopore sequencing and the Shasta toolkit enable efficient *de novo* assembly of eleven human genomes.

Kishwar Shafin et al.

Nature Biotechnology, Accepted March 26, 2020

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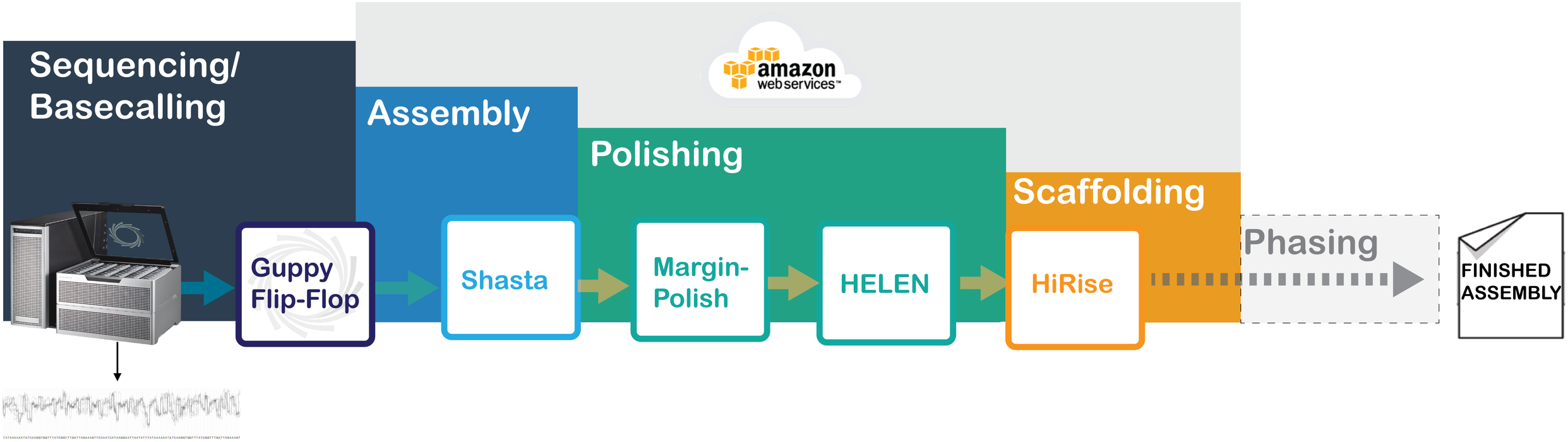

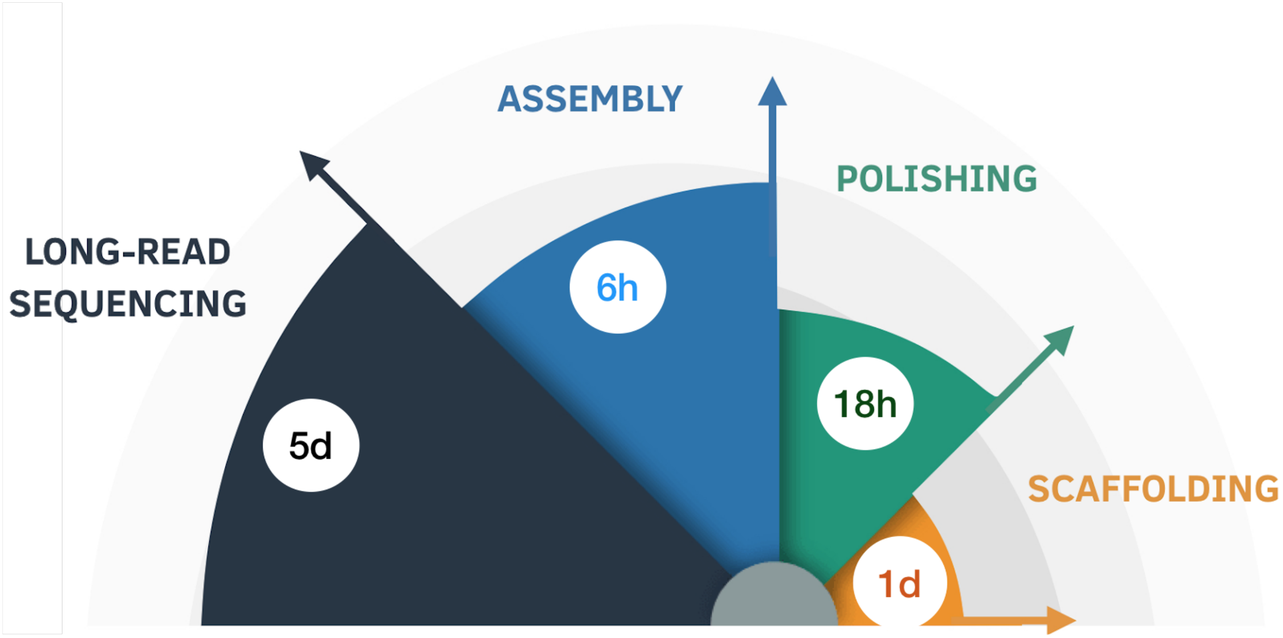

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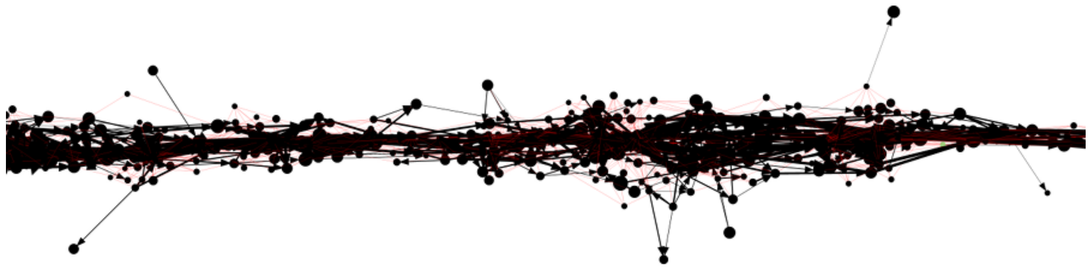
Shasta assembler

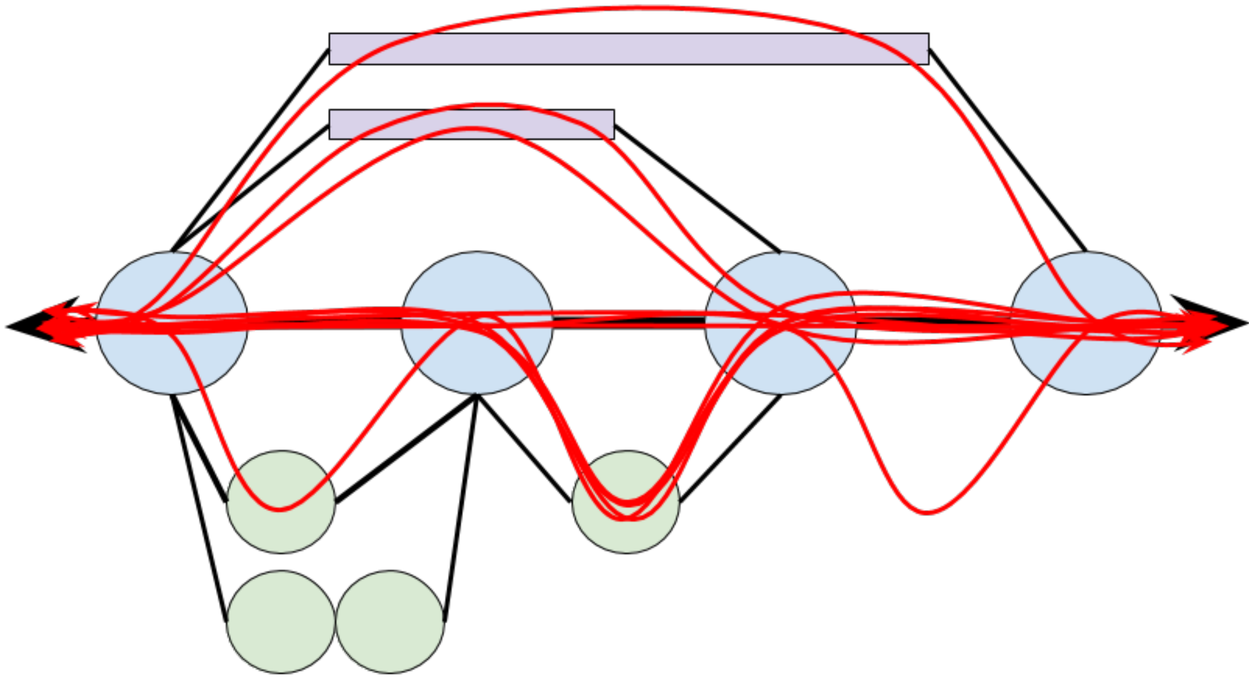
```
 560       570       580       590       600       610       620       630       640       650       660
.|....+....|....+....|....+....|....+....|....+....|....+....|....+....|....+....|....+....|....+....|....+.
1122111132111221311111131211111131211121111211111311112111221111111212111241522531412221112111111111311111
ATATCATCGCATGATCTGAGTACAGCTGTGACTATCACTCATATCAGACTACTGACATGTGATACTCATAGTGCTATACTACTAGTCAGTCTATGTGTATGTGTGATA
 TATCATCGCA  ATCTGAGTAC                          GACTACTGAC          TCATAGTGCT      CTAGTCAGTC ATGTGTATGT
        GCATGATCTG                                    TGACATGTGA    CATAGTGCTA           AGTCTATGTG
                 CTGAGTACAG                                                                    TGTGTATGTG
                  TGAGTACAGC                                                                    GTGTATGTGT
```



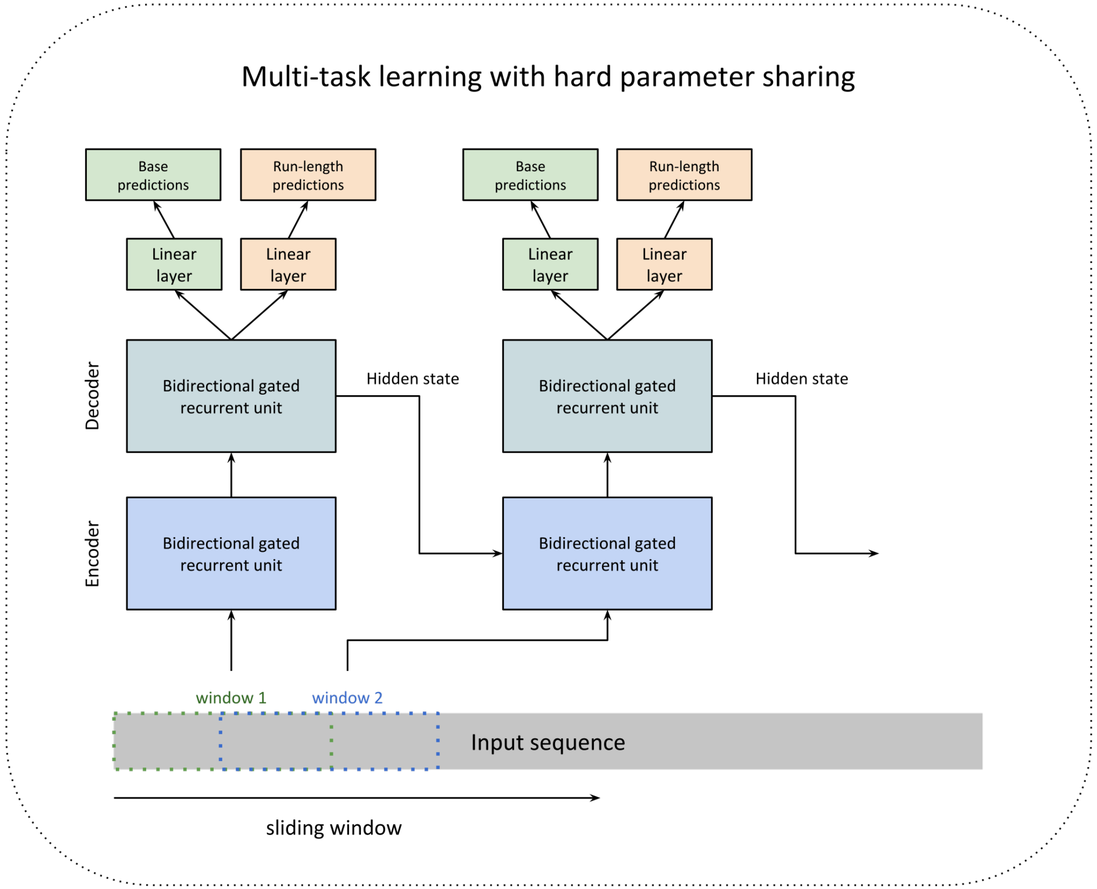
https://github.com/chanzuckerberg/shasta

Paolo Carnevali, CZI

Margin-Polish

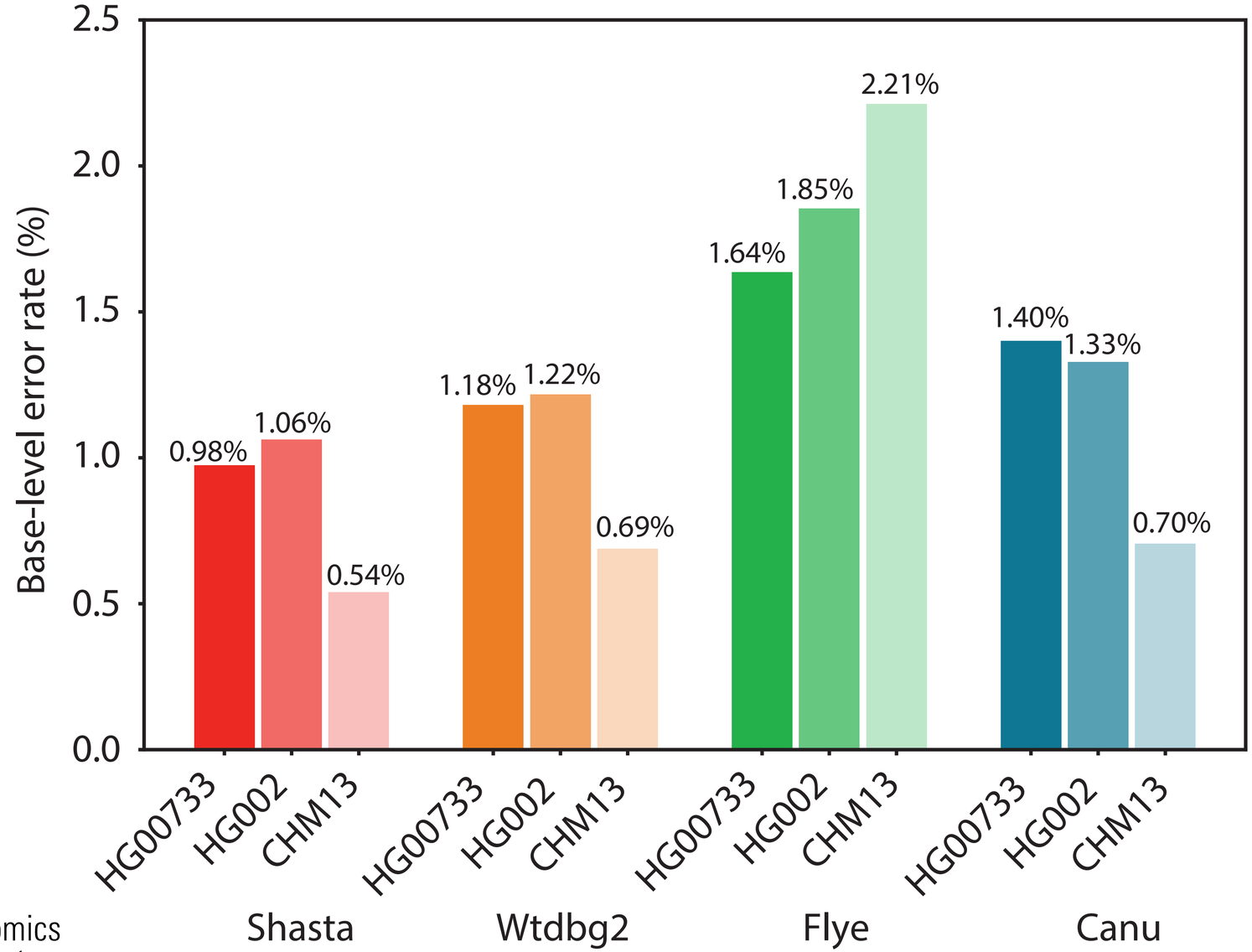


H.E.L.E.N.

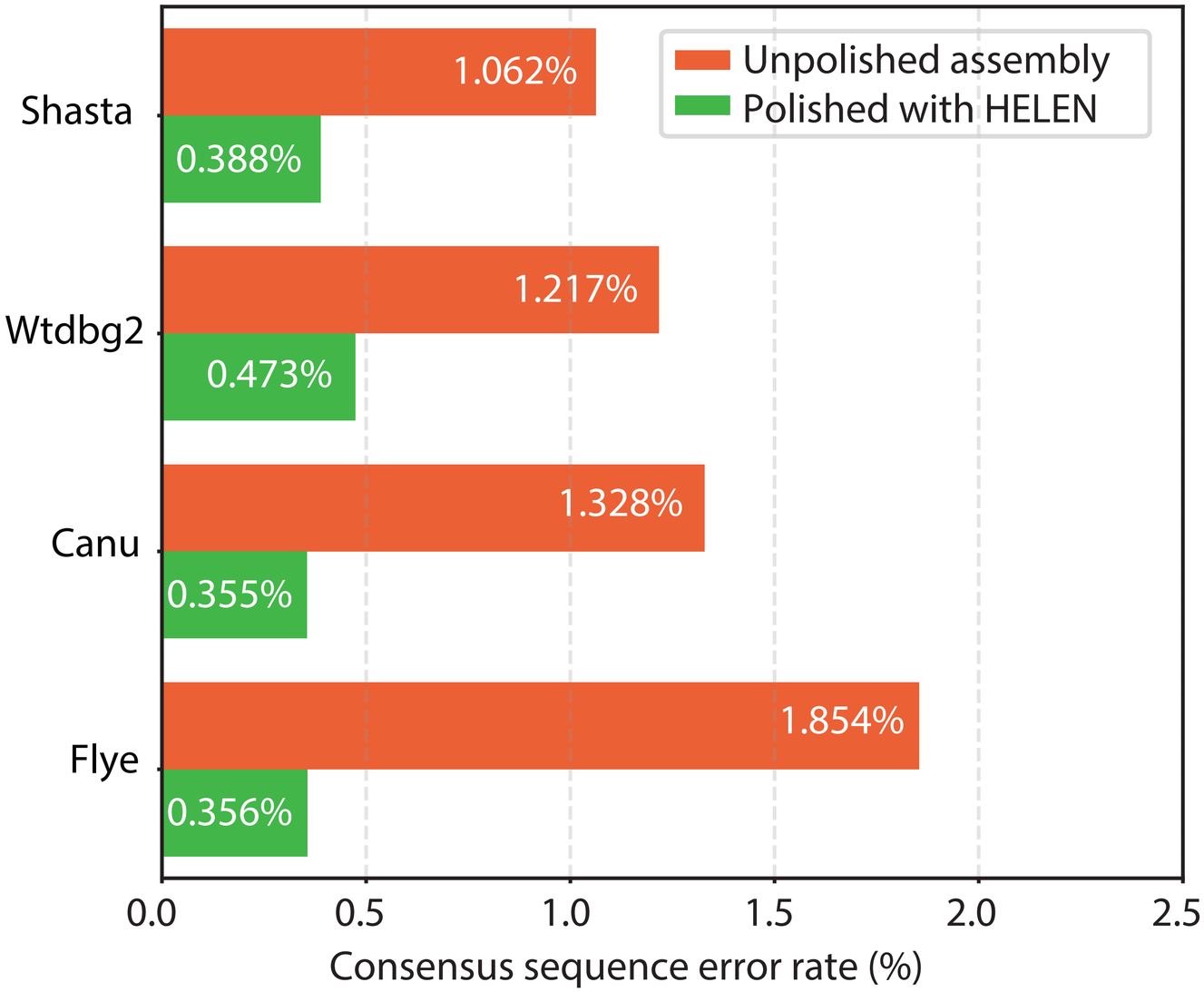


https://github.com/kishwarshafin/helen

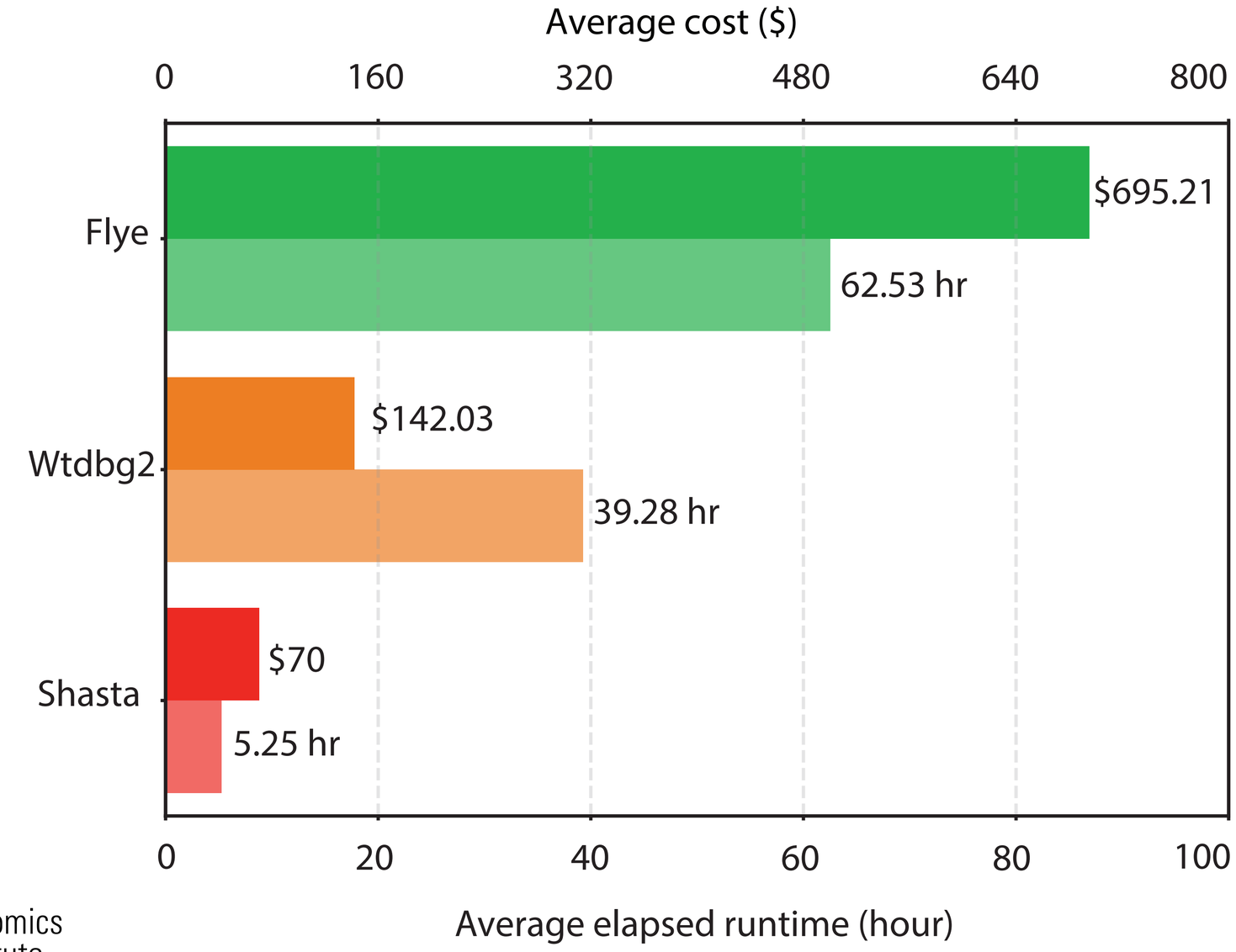
Base-level accuracy



Base-level accuracy improvement



Shasta run-time



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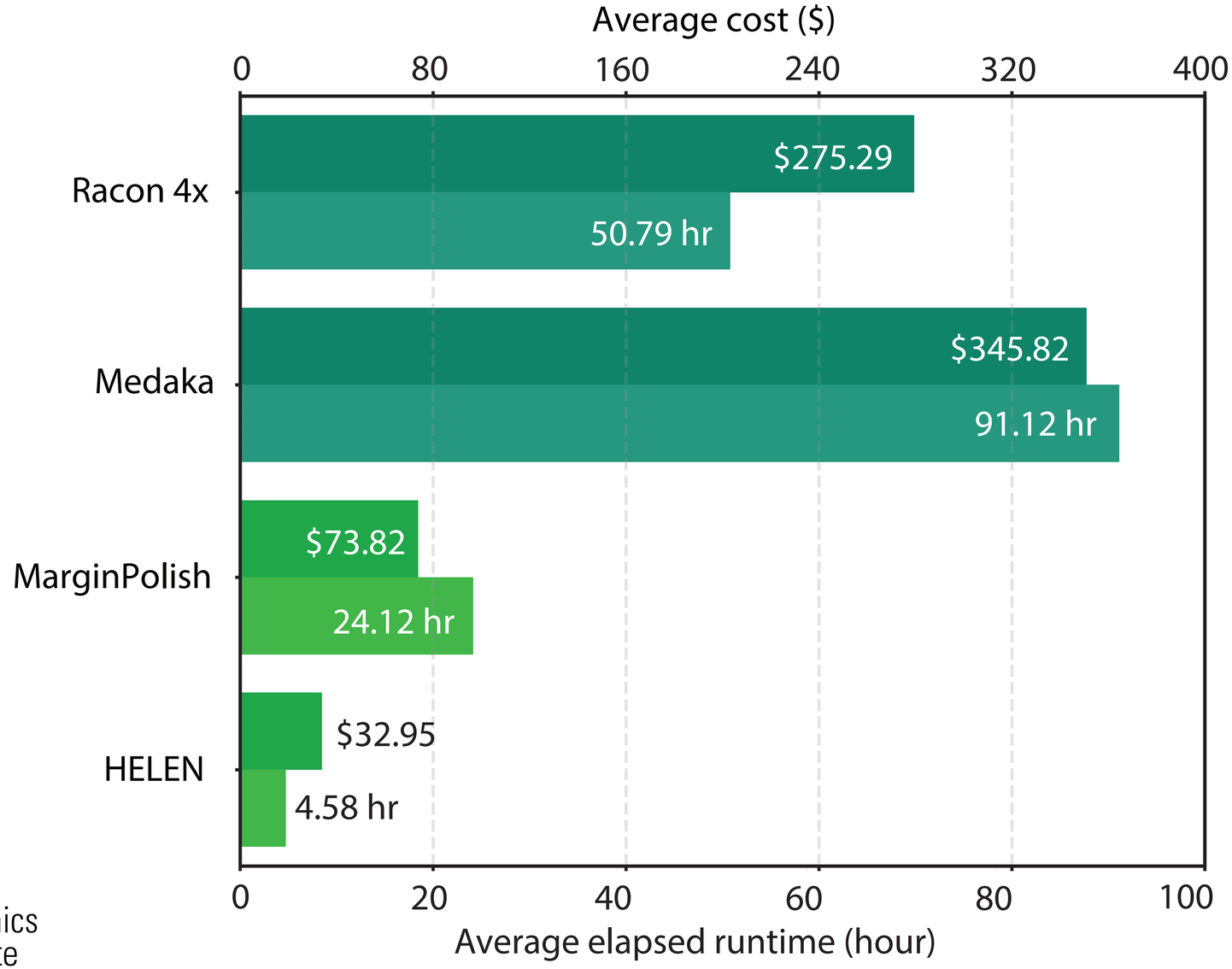
Run-time analysis



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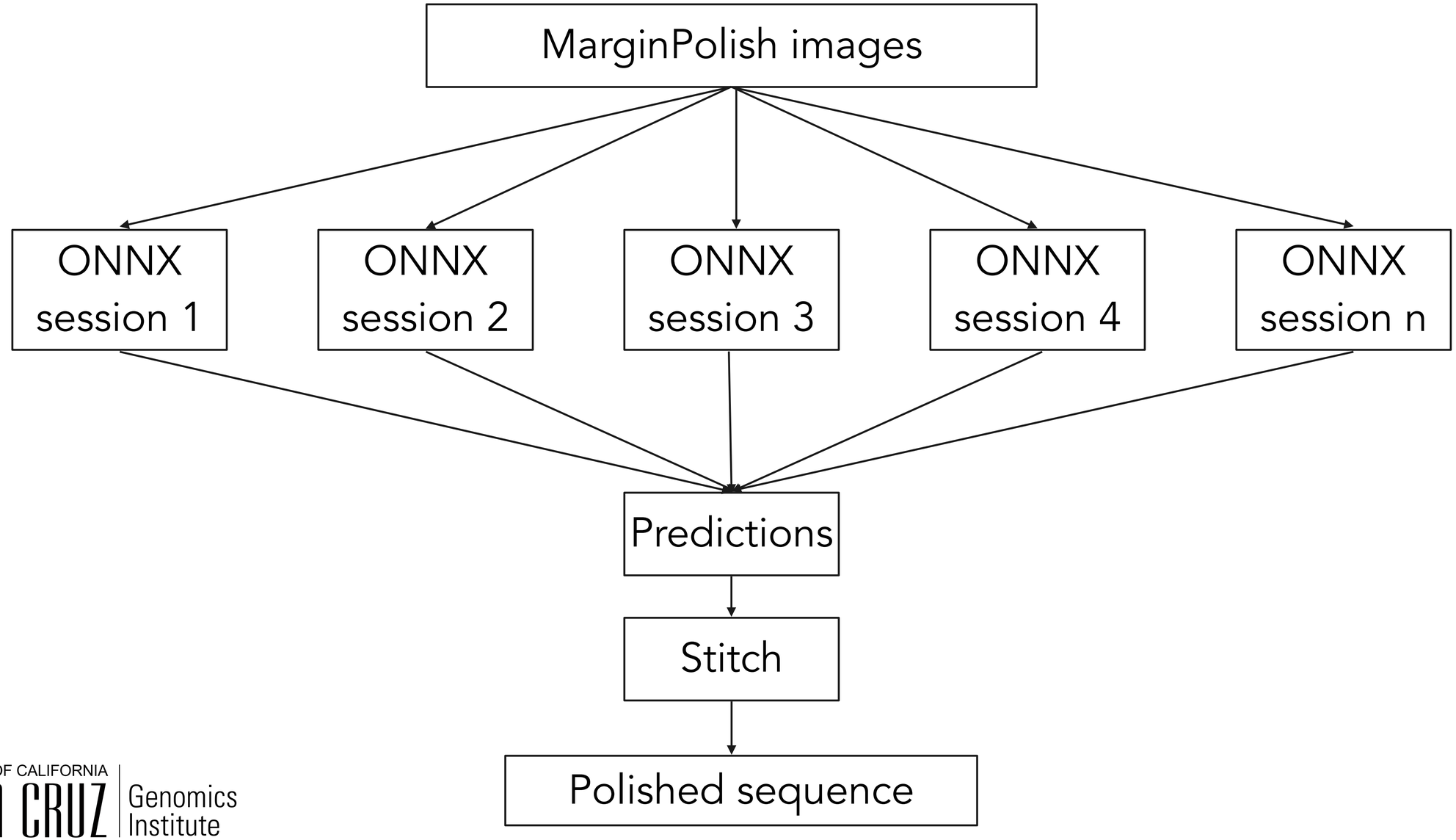
ONNX based HELEN CPU model deployment



Run-time improvement after introducing ONNX runtime

Subset	Expected genome size	Previous release (Wall-clock time)	Current release (Wall-clock time)
Human genome (HG00733)	3.2 Gb	40 hours	7 hours
Human genome (HG00733)	3.2 Gb	36 hours	6 hours
Human genome (CHM13)	3.2 Gb	42 hours	8 hours
Listeria monocytogenes (Microbial)	2.8 Mb	45 mins	6 mins
Bacillus subtilis (Microbial)	4.2 Mb	2 hours	12 mins
Salmonella entrica (Microbial)	5.1 Mb	3 hours	18 mins
Escherichia coli (Microbial)	4.6 Mb	2 hours	13 mins

Acknowledgements

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Google Health

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Sidharth Goel
Maria Nattestad
Howard Yang

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