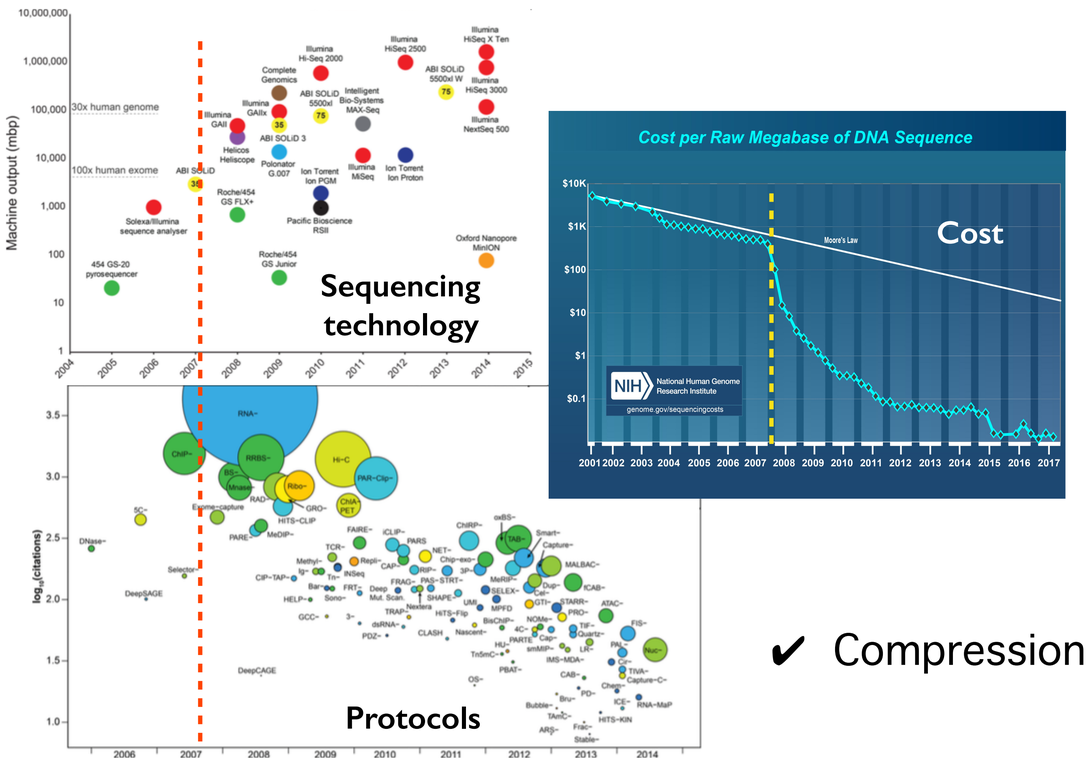
Transcriptome reconstruction with quality score distortion in reference-based alignment

Ana A. Hernandez-Lopez, Claudio Alberti and Marco Mattavelli
École Polytechnique Fédérale de Lausanne, SCI-STI-MM, Lausanne, Switzerland

Motivation

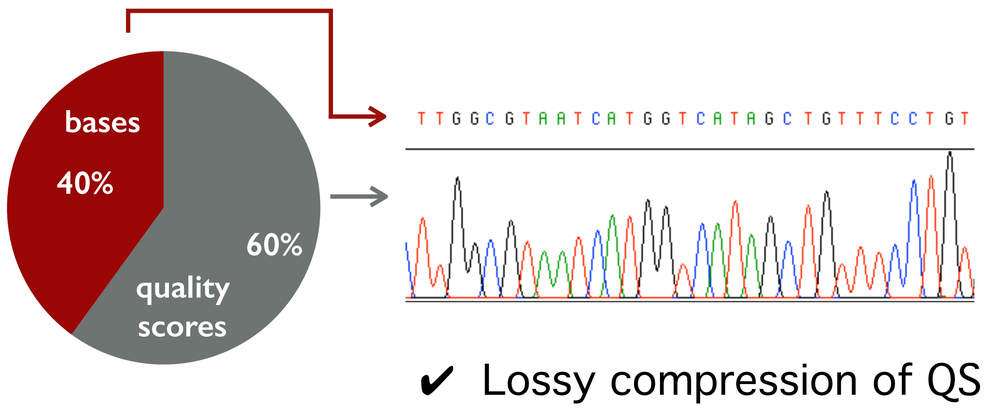
How to cope with the deluge of genomic data?



✔ Compression

Reuter A., et al. "High-throughput sequencing technologies". Molecular Cell, 2015

Is all data informative? Heed the quality scores (QS)



✔ Lossy compression of QS

What downstream applications can deal with QS distortion?

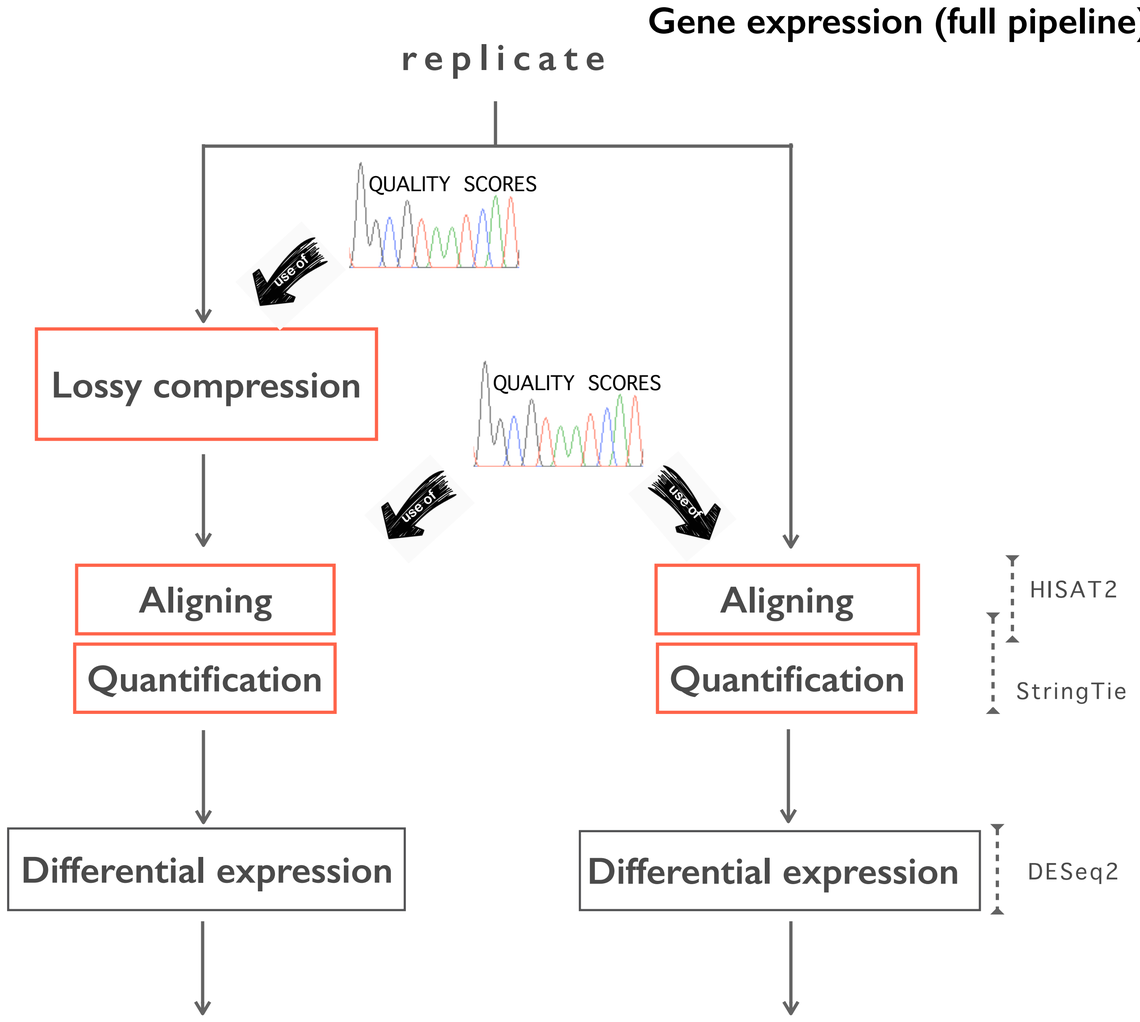
Objectives

- Application of lossy compression in a candidate application for differential gene expression (DGE)
- Evaluate impact of lossy QS on transcript reconstruction
- Measure the effect of lossy QS on the calling of significant genes

Methods

Transcript reconstruction

Gene expression (full pipeline)

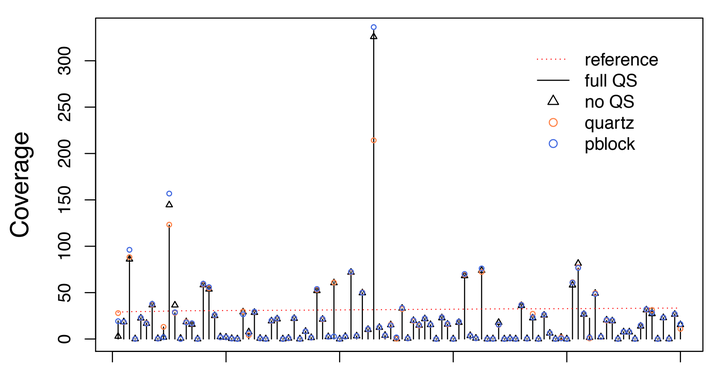


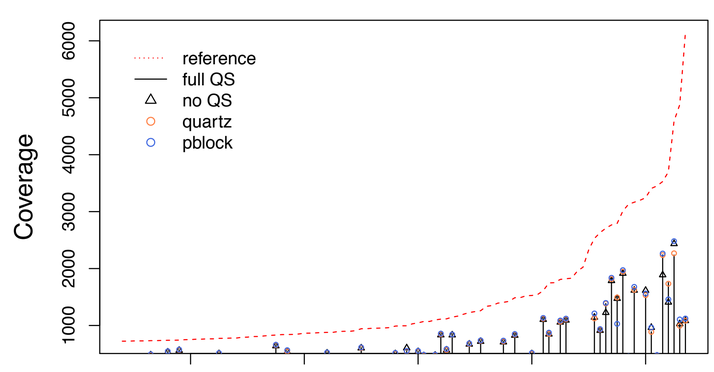
Results

Transcript reconstruction — simulated sample —

		1M	5M	10M	bits/QS
	full QS	77.77	78.28	79.63	3.16
	no QS	76.5	77	78.25	0
Lossy compression	Quartz	77.37	77.56	79.29	1.12
	P-Block	78.73	78.91	80.61	0.98

Alignment percentage





Gene expression — real samples, six replicates —

	cond		UQ2	UQ8	Quartz	P-/R-Block
yeast	1	3.075 [3.05, 3.10]	0.2 [0.2, 0.21]	0.735 [0.72, 0.75]	1.75 [1.66, 1.89]	1.015 [1.0, 1.04]
	2	3.08 [3.05, 3.09]	0.205 [0.2, 0.21]	0.735 [0.72, 0.75]	1.025 [1.75, 1.85]	1.015 [1.0, 1.03]
MCF-7	1	2.21 [1.49, 2.47]	0.16 [0.07, 0.19]	0.70 [0.35, 0.82]	0.57 [0.46, 0.61]	0.975 [0.52, 1.13]
	2	1.68 [1.59, 1.95]	0.09 [0.08, 0.12]	0.44 [0.4, 0.57]	0.49 [0.48, 0.55]	0.635 [0.58, 0.80]

Compression rate

	regulation	log2 fold change UQ2	UQ8	Quartz	P-/R-Block	gene
yeast	up	6.0629	6.0574	5.9761	6.0631	5.9764	YOR192C-A
		5.7313	5.8074	5.8105	5.8147	5.8108	YDR034C-C
		3.6137	3.5778	5.0871	5.2070	3.5193	YHR214C-C
		2.8025	2.7971	2.7996	2.8031	2.7980	YPL025C
		2.5757	2.5702	**2.6641**	2.5764	2.5716	YOR376W
		2.4249	2.3629	**2.5722**	2.3671	2.4629	YPR158C-C
	down	-8.0886	-8.0846	-8.0834	-8.0899	-8.0844	YOR192C-B
		-8.0082	-8.0026	-8.0032	-8.0103	-8.0080	YDR034C-D
		-6.2723	-6.3004	-6.1566	**-6.6860**	**-6.1452**	YER160C
		-3.4012	-2.8554	**-6.0406**	**-6.4943**	**-6.1324**	YHR214C-B
		-2.4985	-2.5184	-4.5319	**-4.8144**	**-3.0414**	YDR210W-A
		-1.8940	-1.8929	-2.4752	**-2.5104**	**-2.5042**	YKL078W

Ranked list of expressed genes

Conclusions

We find that differential gene expression (with a quality-aware assembler) is a promising application over which to use lossy quality score compression. This is supported by the observation that the calling of the most salient and discernible genes are affected by their compression.