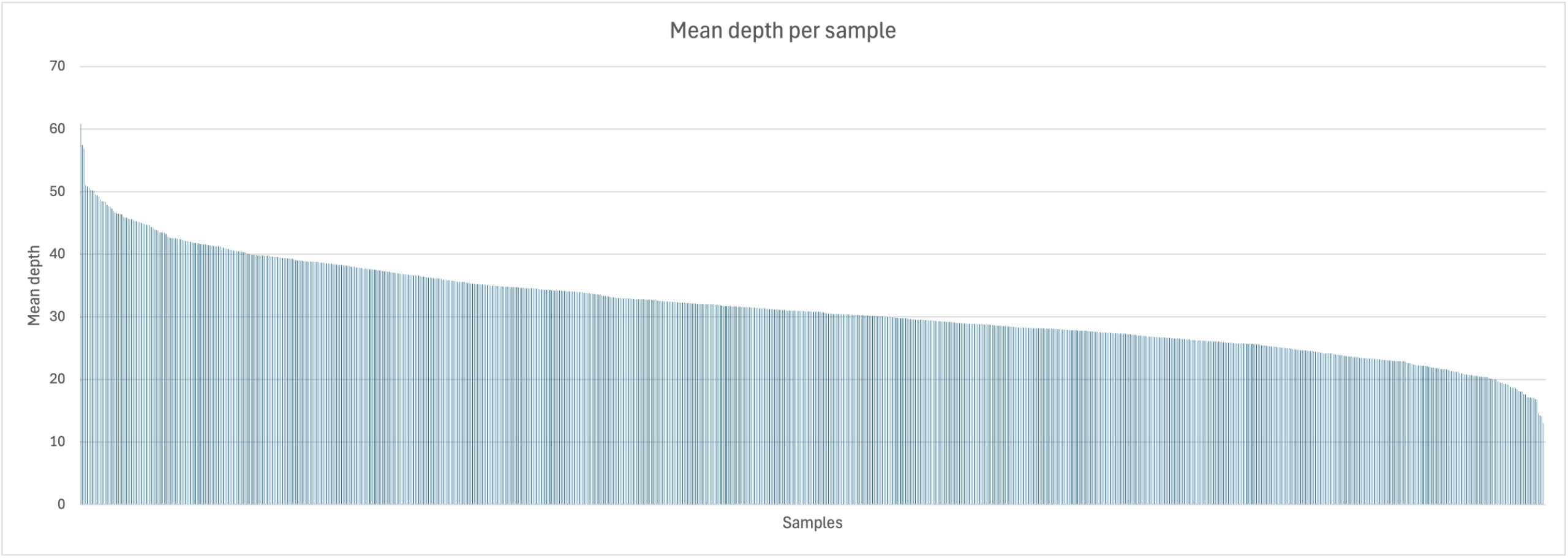
Sequence it all: The whole genome, not a capture

WGS Project #1: December-January 2025, v 1.7 chemistry
≈ 900 Human Samples: “same cost as a 4 MB capture”
Library creation: NEB PCR-free
Loading: 15 samples / wafer (≈ 60 wafers)



Sequence it all: The whole genome, not a capture

Genome-in-a-bottle benchmarking met expectations (≈35X)

SNPs					
True-pos-call	False-pos	False-neg	Precision	Sensitivity	F-measure
3,192,834	1,995	23,002	99.94%	99.28%	99.61%
Indels					
True-pos-call	False-pos	False-neg	Precision	Sensitivity	F-measure
317,896	1,284	10,143	99.60%	96.91%	98.23%
Overall					
True-pos-call	False-pos	False-neg	Precision	Sensitivity	F-measure
3,510,730	3,279	33,145	99.91%	99.06%	99.48%

Sequence it all: The whole genome, not a capture

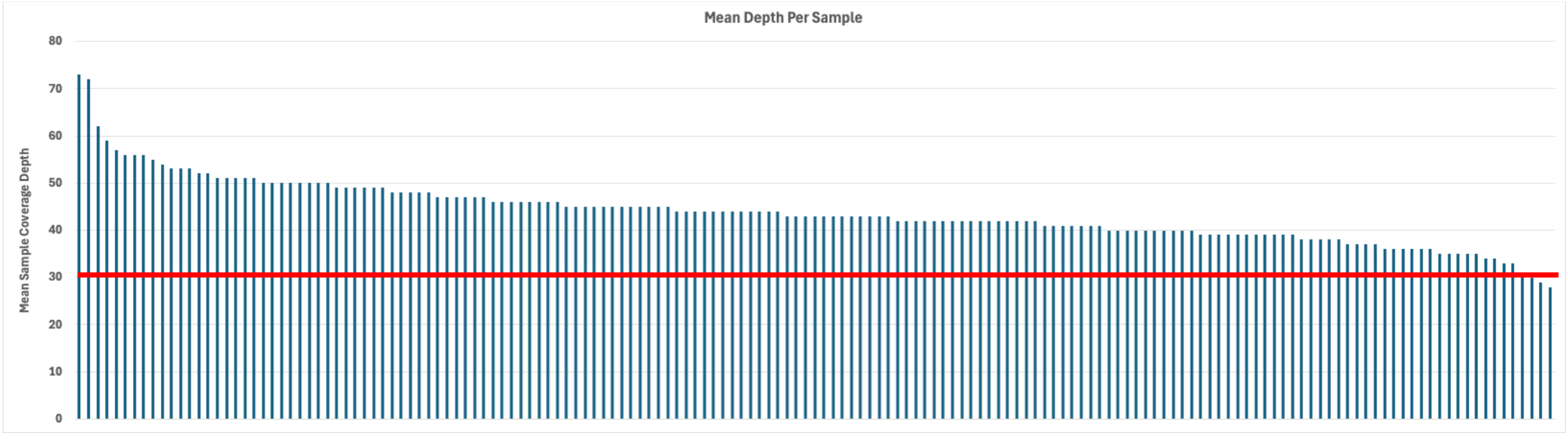
WGS Project #2: August-October 2025, v 1.8 chemistry
≈ 900 Human Samples: "same cost as genotyping array"
Library creation: NEB PCR-free
Loading: 24 samples / wafer (≈ 40 wafers)

Wafer	Reads PF (M)	Aligned Bases (TB)	Read Length (bp)
1	11,757	3.17	281
2	13,359	3.66	284
3	12,969	3.26	274
4	13,194	3.50	277
5	13,090	3.54	278
6	12,918	3.50	280
7	13,267	3.49	273
8	13,792	3.63	274
9	13,844	3.69	276
10	13,322	3.53	275

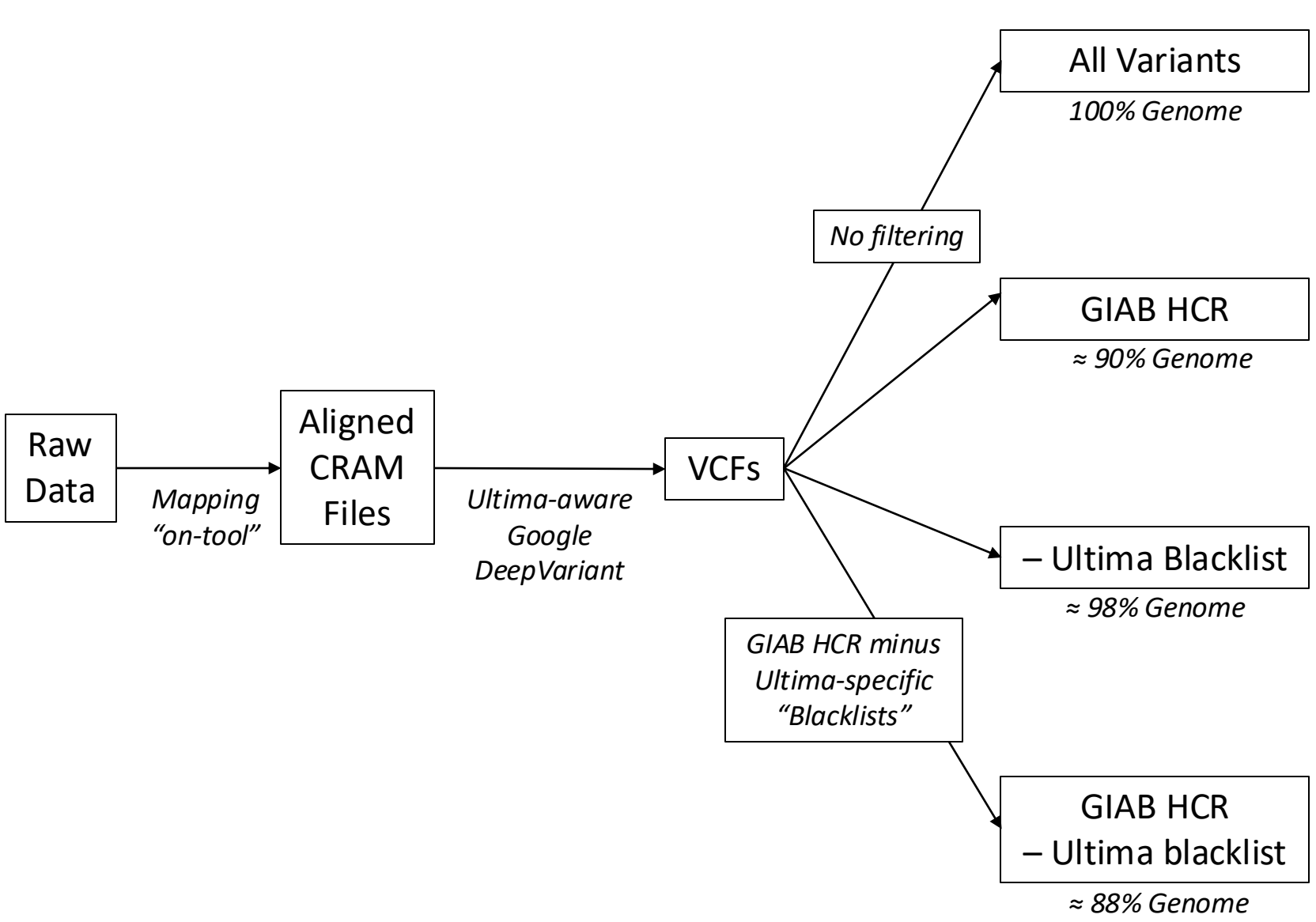
Sequence it all: The whole genome, not a capture

WGS Project #2: August-October 2025, v 1.8 chemistry
≈ 900 Human Samples: “same cost as genotyping array”
Library creation: NEB PCR-free
Loading: 24 samples / wafer (≈ 40 wafers)

First 161 Samples: mean depth = 43.8X, CV = 15%



GIAB Sample F1 Scores vs Depth



	Precision	Sensitivity	F-measure
SNPs	0.854	0.989	0.917
Indels	0.573	0.854	0.686

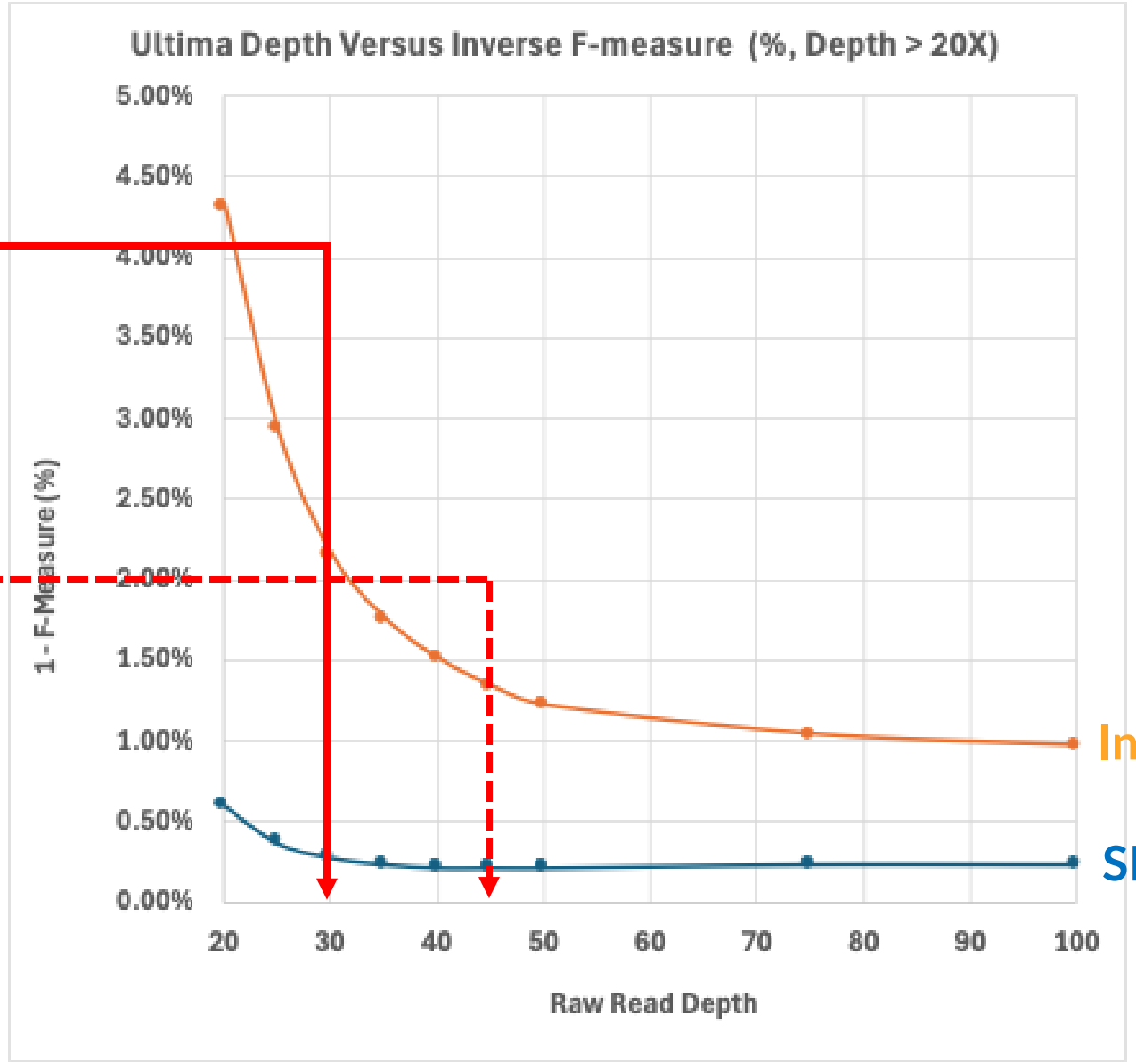
	Precision	Sensitivity	F-measure
SNPs	0.997	0.991	0.994
Indels	0.904	0.878	0.890

	Precision	Sensitivity	F-measure
SNPs	0.884	0.996	0.937
Indels	0.756	0.959	0.846

	Precision	Sensitivity	F-measure
SNPs	0.998	0.996	0.997
Indels	0.984	0.973	0.978

GIAB Sample VCF Filtering Analysis

	Precision	Sensitivity	F-measure
20X Raw			
SNPs	0.9957	0.9922	0.994
Indels	0.9715	0.9425	0.9568
25X Raw			
SNPs	0.9973	0.9954	0.9963
Indels	0.9791	0.9625	0.9707
30X raw			
SNPs	0.998	0.9964	0.9972
Indels	0.9837	0.9731	0.9784
35X raw			
SNPs	0.9984	0.9968	0.9976
Indels	0.9863	0.9786	0.9824
40X raw			
SNPs	0.9986	0.997	0.9978
Indels	0.9877	0.982	0.9849
45X raw			
SNPs	0.9987	0.997	0.9978
Indels	0.989	0.9843	0.9866
50x raw			
SNPs	0.9987	0.9969	0.9978
Indels	0.9899	0.9858	0.9878
75X raw			
SNPs	0.9988	0.9964	0.9976
Indels	0.9914	0.9878	0.9896
100X raw			
SNPs	0.9989	0.9963	0.9976
Indels	0.992	0.9886	0.9903
133X raw			
SNPs	0.9989	0.9963	0.9976
Indels	0.9919	0.9887	0.9903



Sequence them all: Model Organism Sequencing Service (MOSS)

Model Organism Sequencing Service

Applications

By driving down the cost of sequencing, the UMGC aims to enable researchers to use MOSS as a standard, efficient means to accomplish a variety of analyses:

- Mutation and insertion mapping – Identify unknown genetic variants by comparing against reference strains.
- CRISPR edit confirmation – Validate genome edits with high precision.
- Structural variant detection – Characterize genomic changes such as deletions, duplications, and translocations.
- Genotyping and strain authentication – Ensure the accuracy of genetic backgrounds in research models.

***Just* sequence:** Non-globin-depleted RNA-Seq

Whole blood

Extraction, Globin Depletion

RNA – Globin

PolyA-selection RNA-Seq

20M Reads RNA-Seq Data

+ $50 - $75
+ Bias
+ Operator Error

Extraction

RNA

PolyA-selection RNA-Seq

100 M Reads RNA-Seq Data

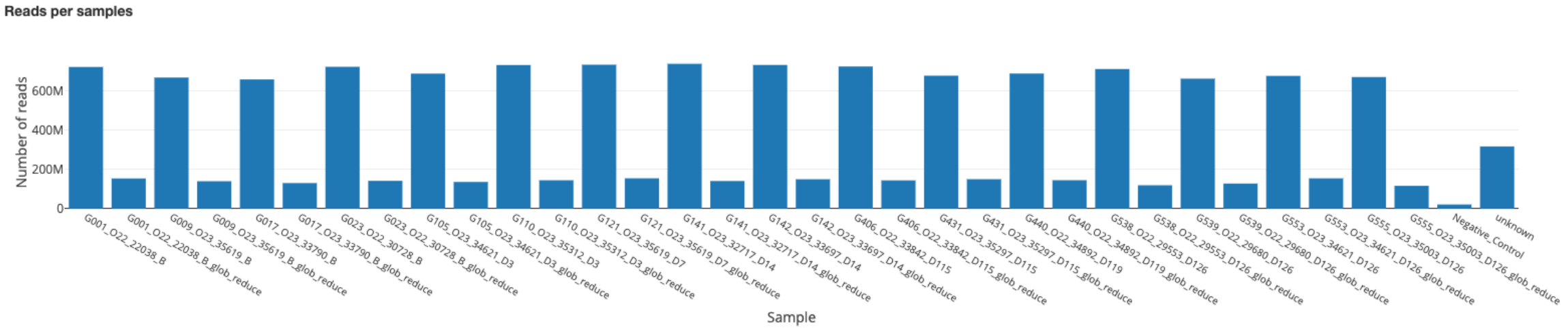
Discard Globin Reads

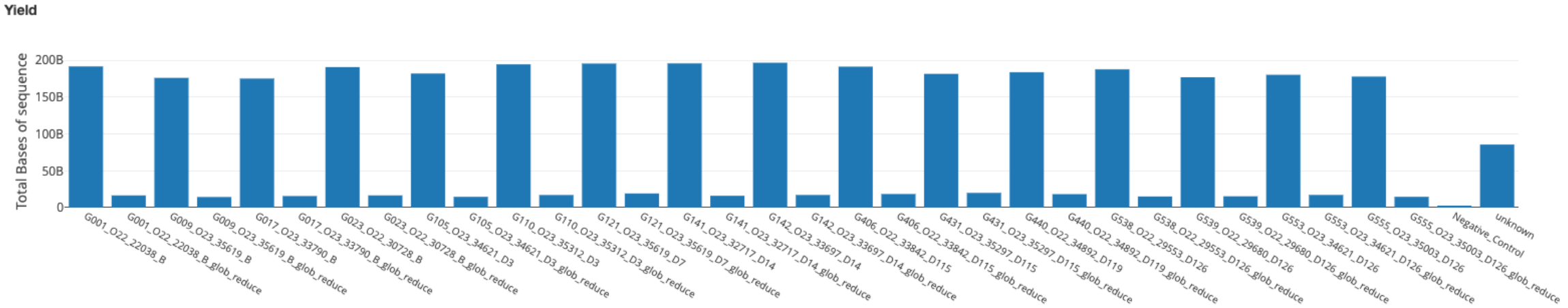
20M Reads RNA-Seq Data

+ $20
No Bias
No Operator Error

***Just* sequence:** Non-globin-depleted RNA-Seq

1 UG100 wafer, 32 samples: 13.4B reads (10-12B expected)
~ 700M reads per mRNA sample
~ 140M reads per globin-reduced sample

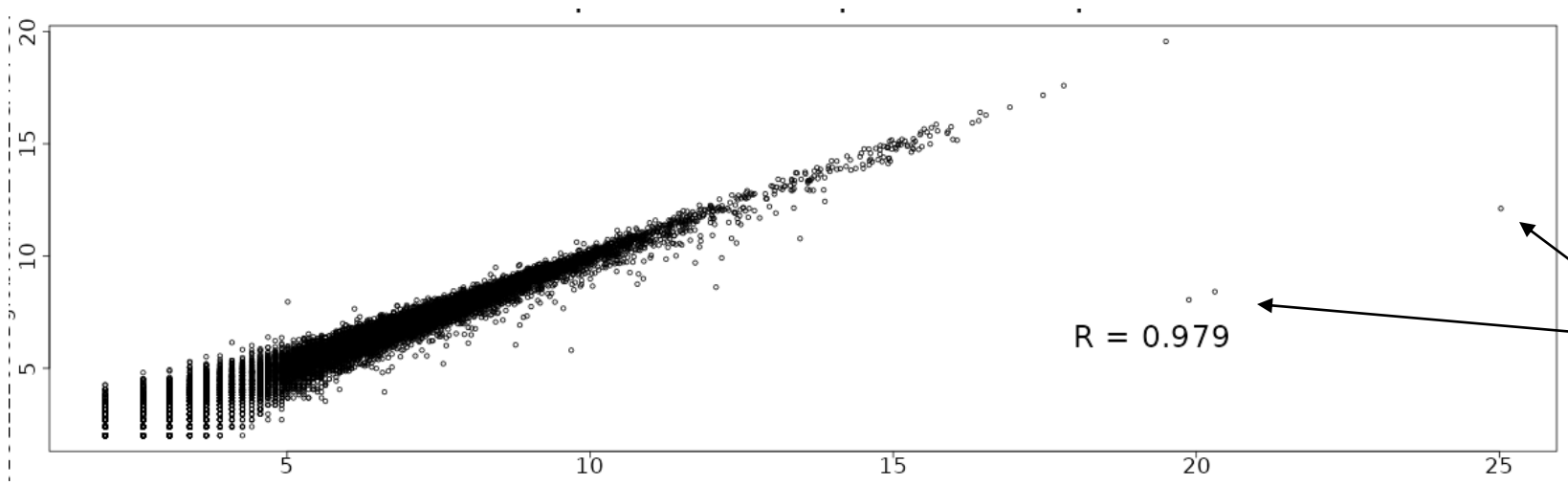




***Just* sequence:** Non-globin-depleted RNA-Seq

Scatter plot of gene expression counts for one sample

Globin-reduced

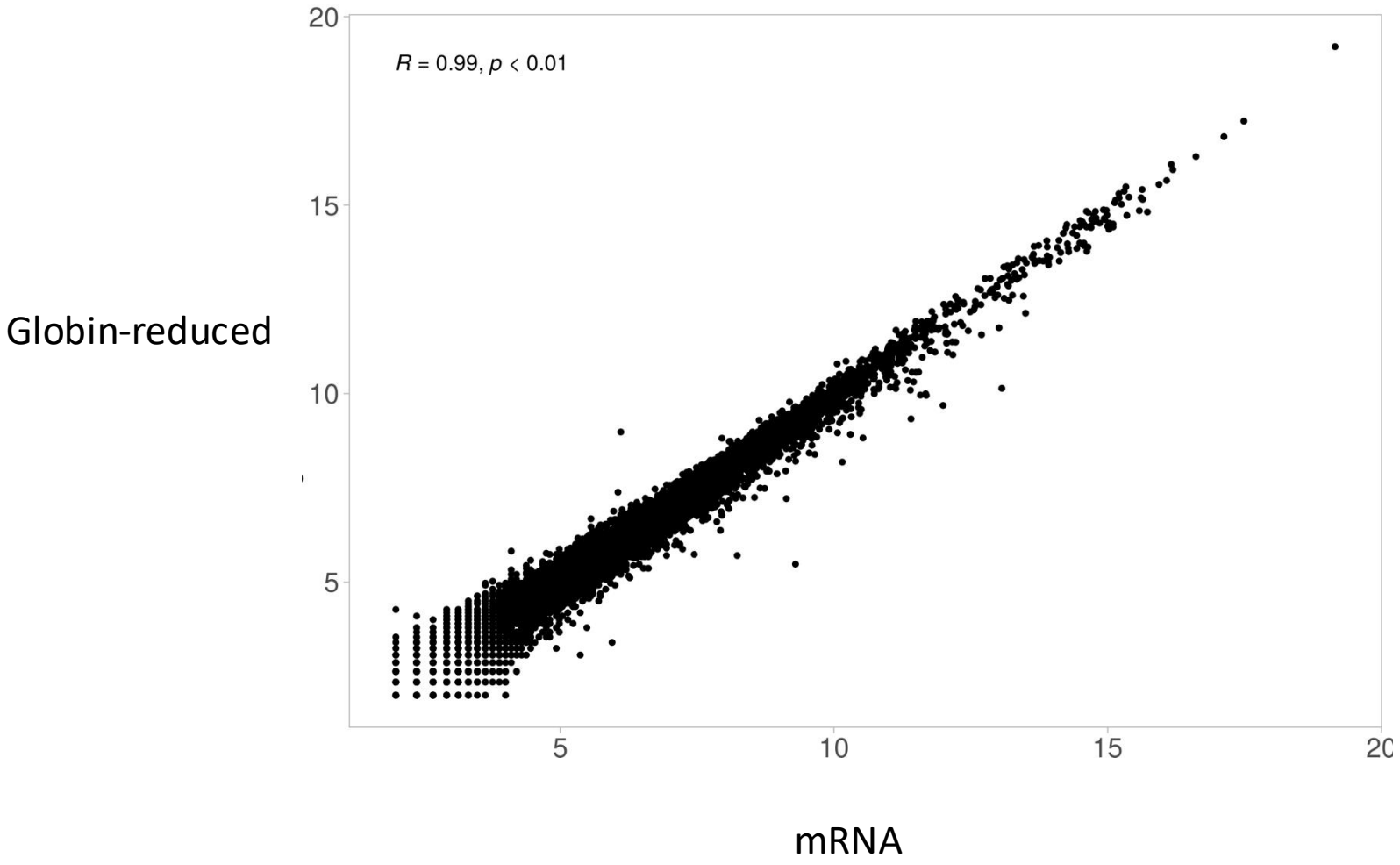


3 globin genes

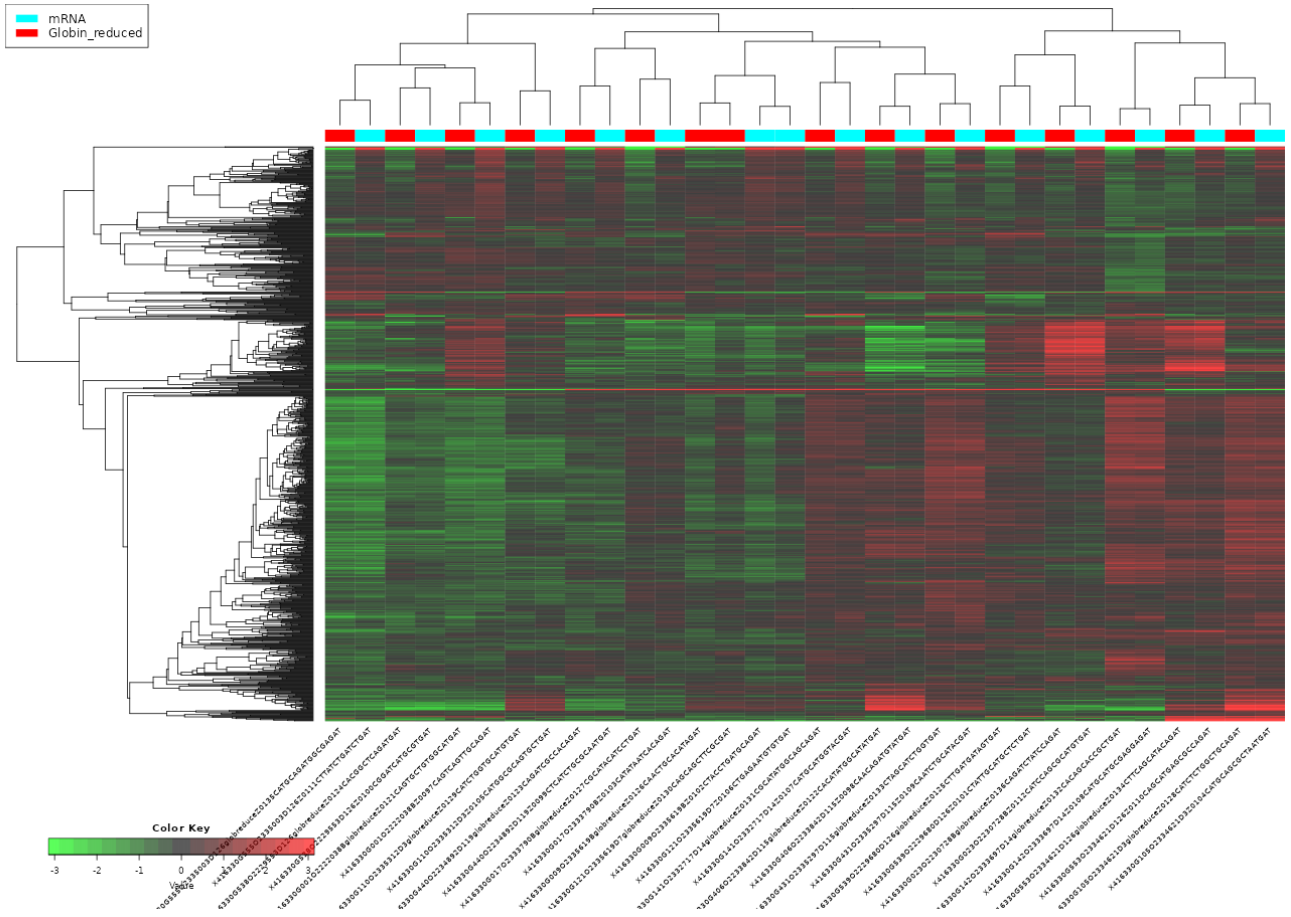
mRNA

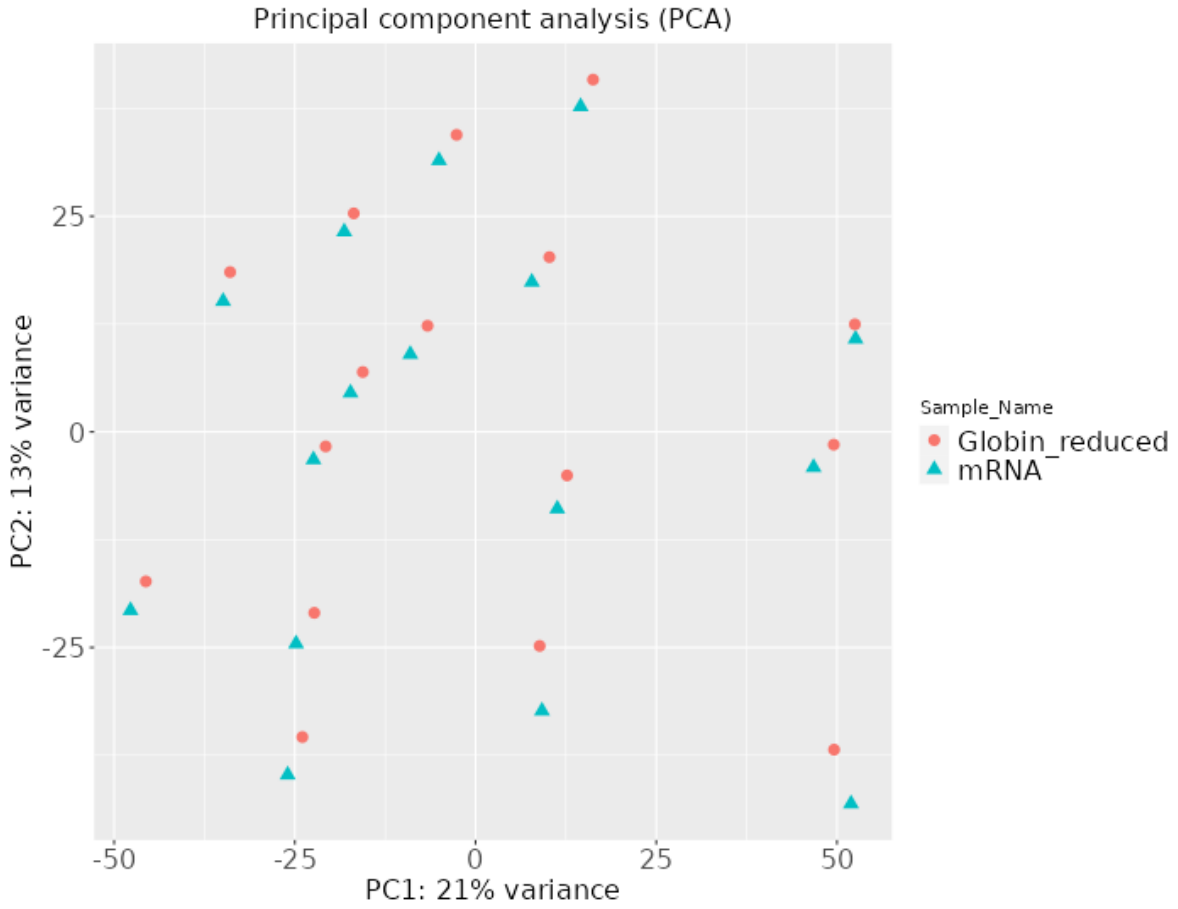
***Just* sequence:** Non-globin-depleted RNA-Seq

After *in silico* globin depletion and read depth normalization



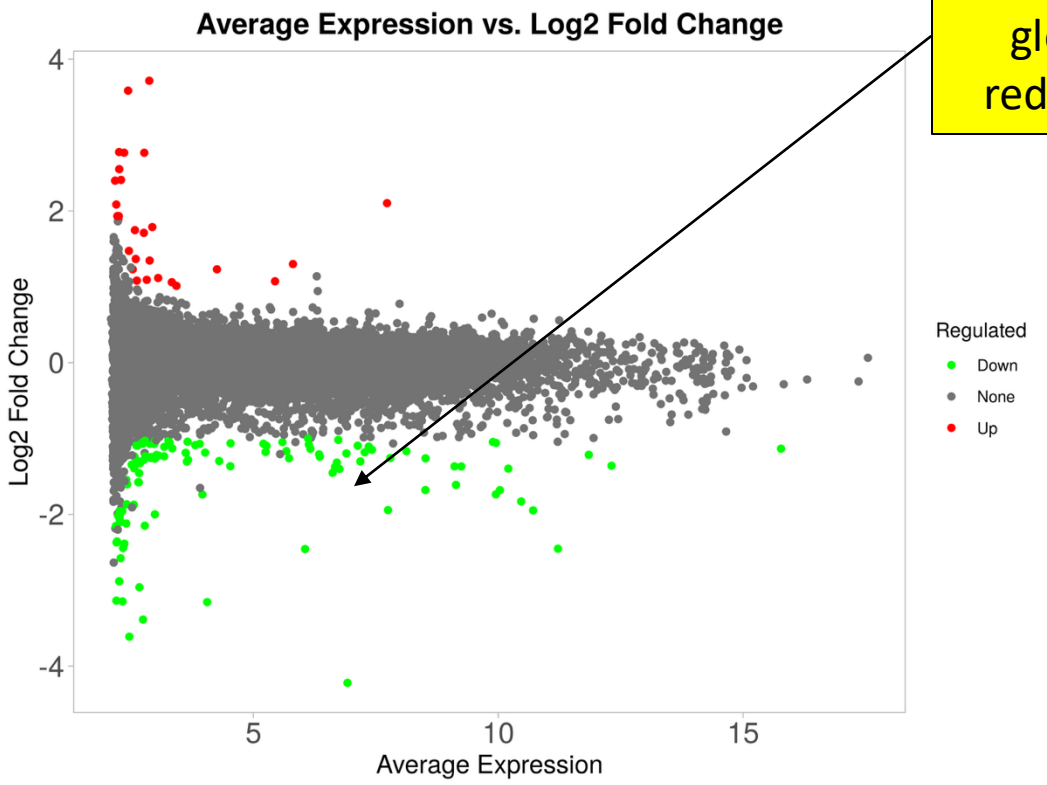
***Just* sequence:** Non-globin-depleted RNA-Seq





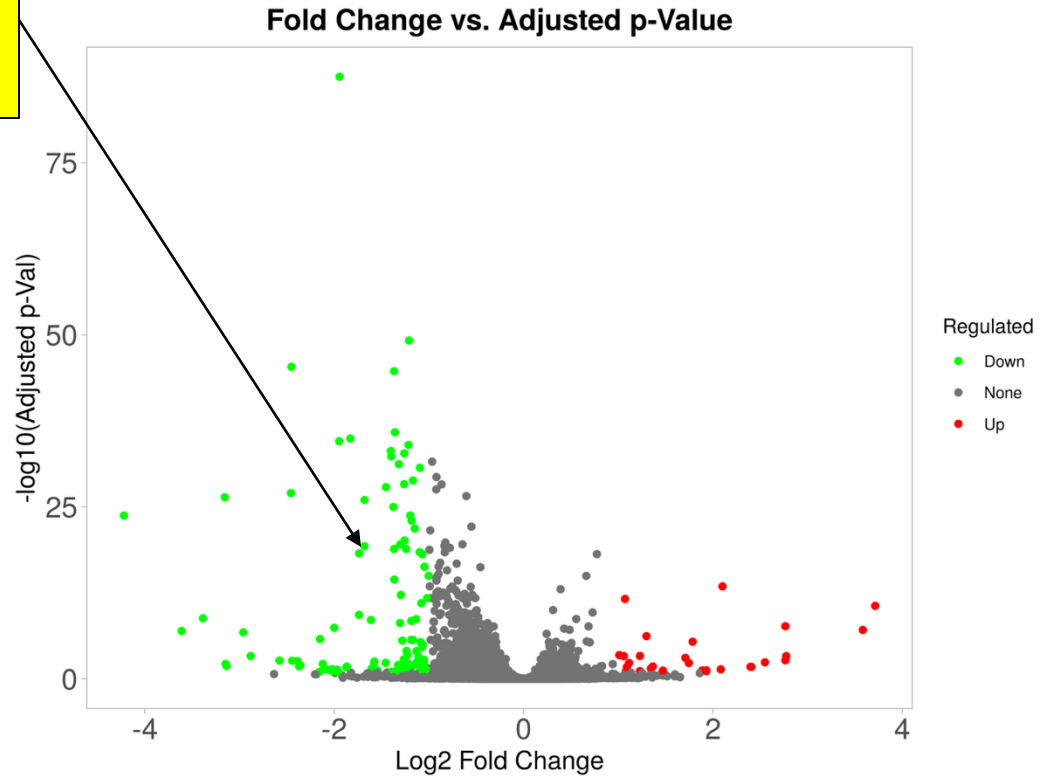
***Just* sequence:** Non-globin-depleted RNA-Seq

MA-plot: Globin-reduced vs mRNA



Lower in globin-reduction

Volcano plot: Globin-reduced vs mRNA



genes with significant difference:

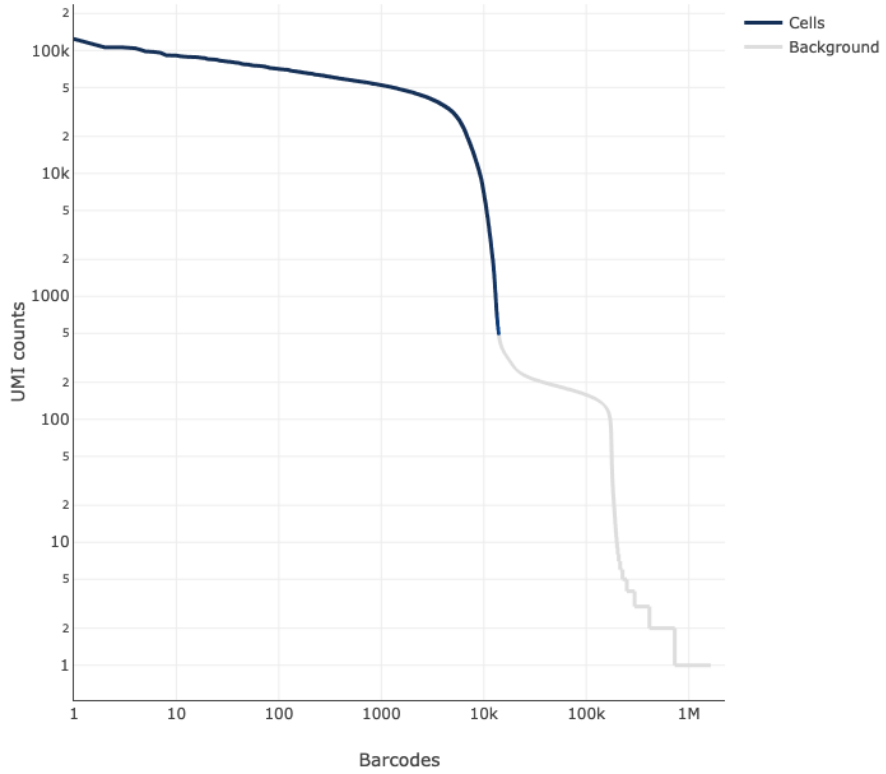
Comparison	Up	Down
GLOBIN_REDUCED-MRNA	28	112

Preponderance of expression reduction suggests off-target depletion

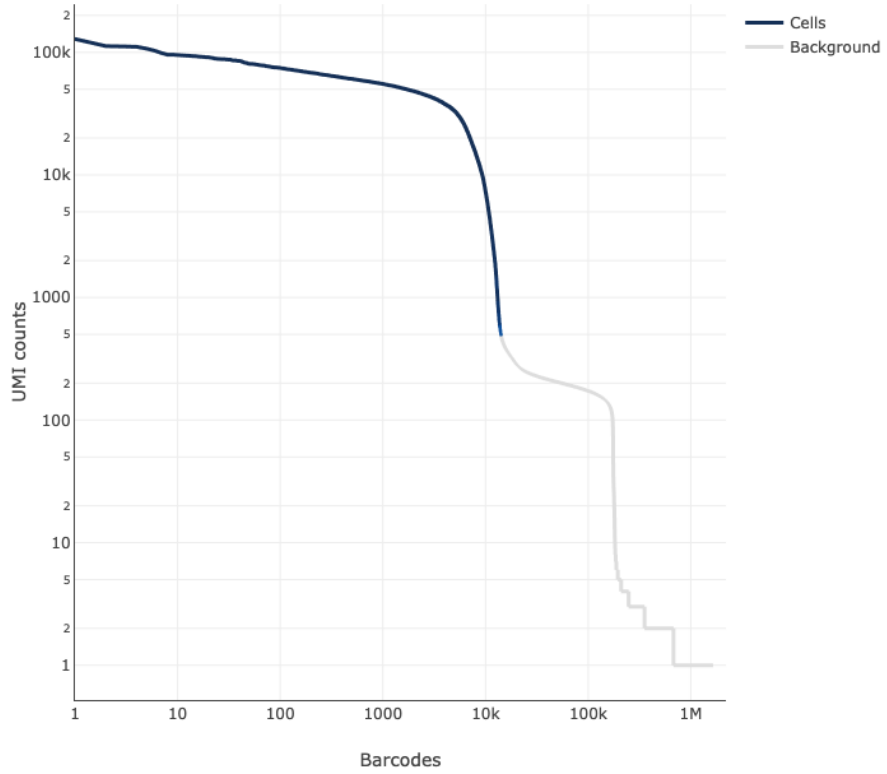
Ultima Platform Benchmarking: UMGC Single-Cell Group

The UMGC benchmarked 10X 3’ GEX on our two platforms (AVITI vs Ultima): barcode rank plots pf one library sequenced on AVITI and UG 100

UG100



Aviti



Ultima Platform Benchmarking: UMGC Single-Cell Group

The UMGC benchmarked 10X 3’ GEX on our two platforms (AVITI vs Ultima): expression metrics

	UG100	Aviti
Cells	207,378	208,896
Valid Barcodes	100%	96.1%
Valid UMIs	99.9%	99.9%
Reads Mapped confidently to Transcriptome	68.3%	67.9%
Number of Mean reads/ Cell	45,151	45,031
Fraction Reads in cells	92.5%	92.5%

Conclusion: no discernible difference between AVITI and Ultima in 3’ GEX gene expression outcomes.

Ultima Platform Benchmarking: Collaboration with 10x Genomics

10x Genomics + UMGC: understand performance and experience of UG 100 for 10x Genomics

Benchmarking: SCG and Spatial libraries created and sequenced by 10x Genomics were send to UMGC and re-sequenced on UMGC's UG 100. All analysis and interpretation by 10x Genomics.

10x Assay	Sample Type	Sequencer	Location
GEM-X 5' Gene Expression	Human PBMC rep1	NovaSeq X+	10x Genomics
		UG 100	UMN Genomics Center
	Human PBMC rep2	NovaSeq X+	10x Genomics
		UG 100	UMN Genomics Center
GEM-X Flex Gene Expression	K562 rep1	NovaSeq X+	10x Genomics
		UG 100	UMN Genomics Center
	K562 rep2	NovaSeq X+	10x Genomics
		UG 100	UMN Genomics Center
Visium HD Spatial Gene Expression	Mouse Brain	NovaSeq X+	10x Genomics
		UG 100	UMN Genomics Center
	Human Tonsil	NovaSeq X+	10x Genomics
		UG 100	UMN Genomics Center

Data courtesy of 10x Genomics

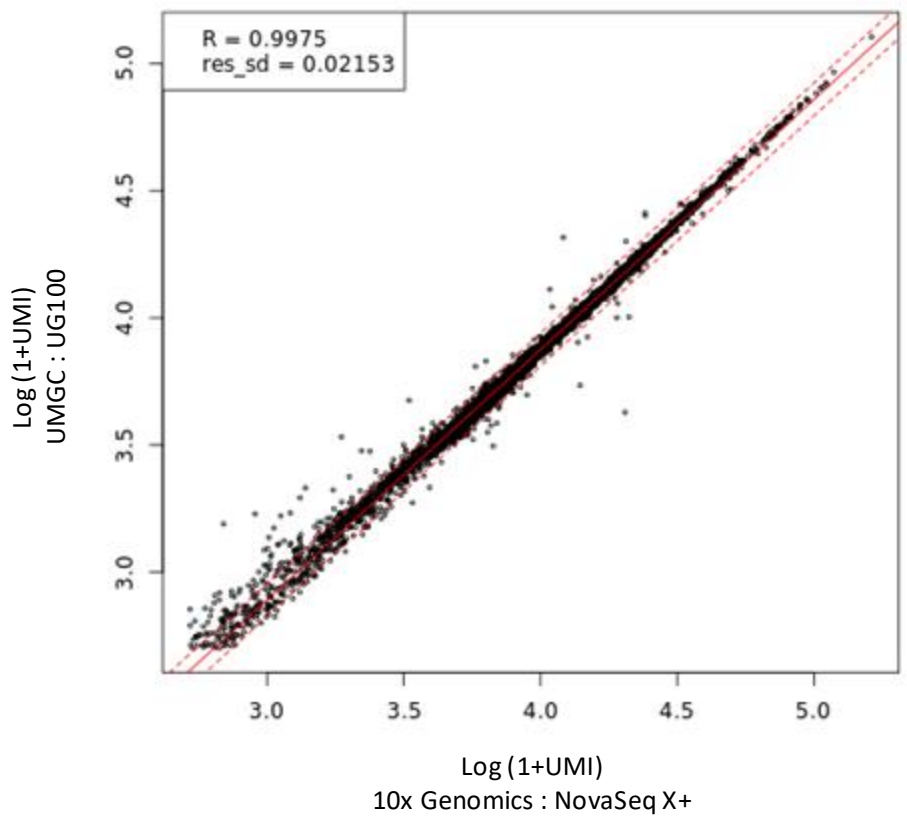
GEM-X 5’ Gene Expression Sequencing Metrics

Metric	10x Genomics NovaSeq X+ rep1	UMGC UG 100 rep1	10x Genomics NovaSeq X+ rep2	UMGC UG 100 rep2
Cells	16,170	16,025	16,263	15,944
Reads Mapped Confidently to Exonic Regions	68.3%	72.3%	68.3%	72.5%
Reads Mapped Confidently to Transcriptome	72.4%	78.7%	72.2%	78.6%
Fraction Reads in Cells	97.3%	96.9%	97.7%	97.2%
Median Genes Per Cell (10k raw reads per cell)	1,450	1,504	1,450	1,495

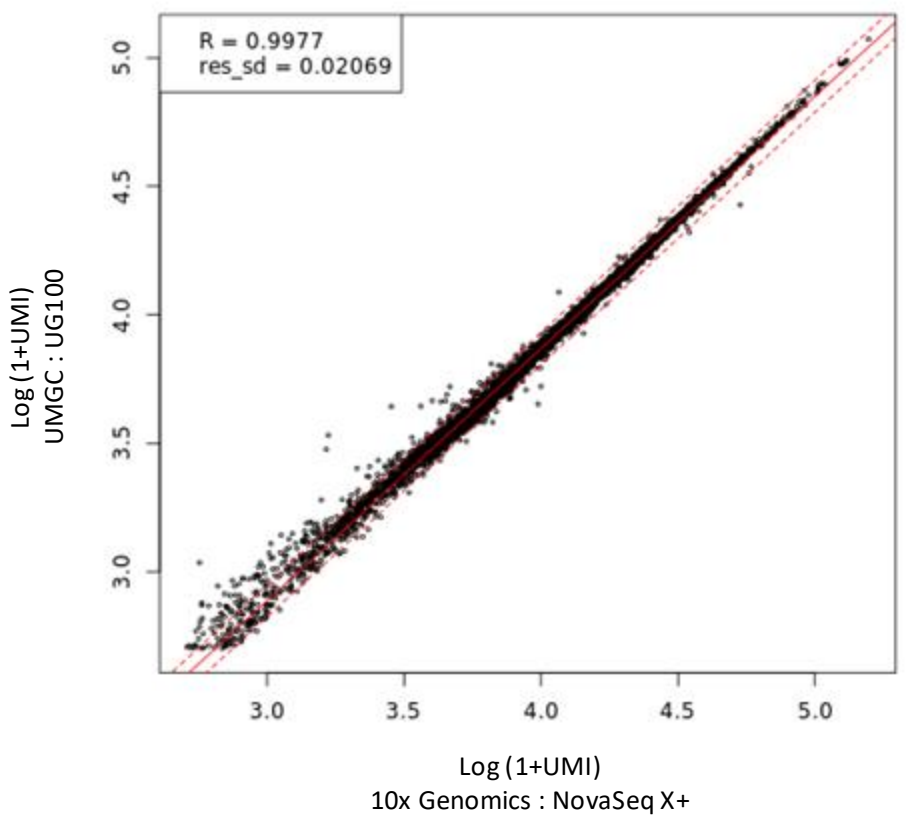
Data courtesy of 10x Genomics

GEM-X 5’ gene expression: counts per barcode

Replicate 1



Replicate 2

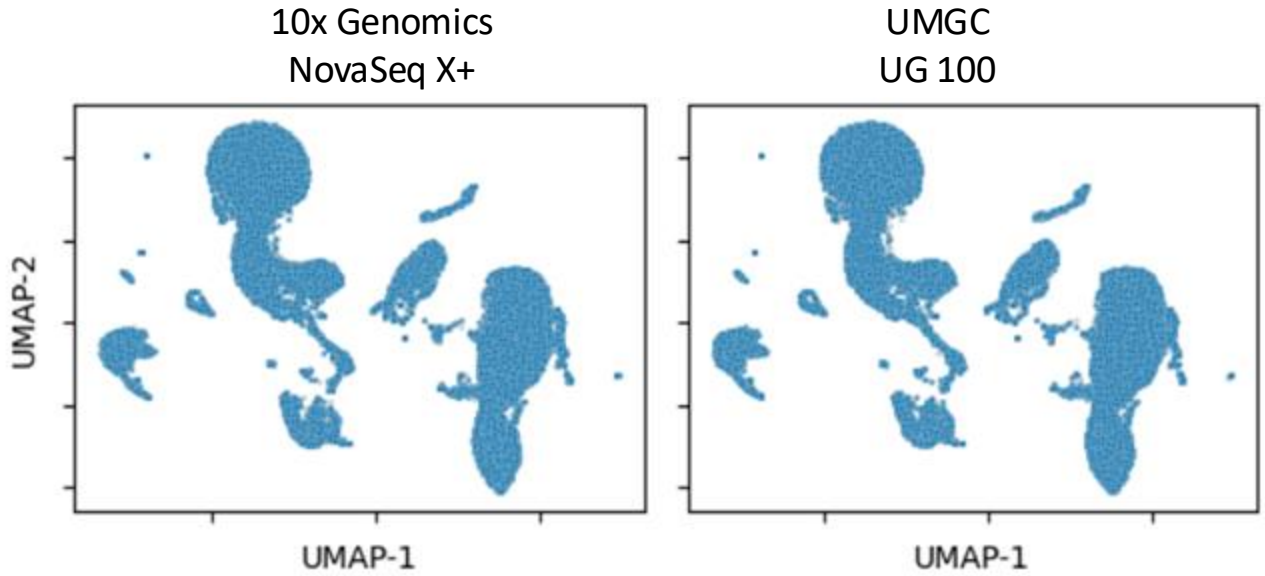


10x Genomics R&D conclusions:

- Overall UMI counts per cell-barcode highly similar across sequencers (UG 100 vs NovaSeq X+)
- Cell-barcode calls virtually identical (~99%) across sequencers

Data and analysis courtesy of 10x Genomics

GEM-X 5’ gene expression: UMAP clusters



10x Genomics R&D conclusions:

- UMAP clusters highly similar and overlapping

Data and analysis courtesy of 10x Genomics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