Breakout Session 2: Track A

SRA RNA-seq Precomputed Alignments and Gene Expression Counts

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Acting Director, NCBI, NIH/NLM

SRA RNA-seq precomputed alignments and gene expression counts

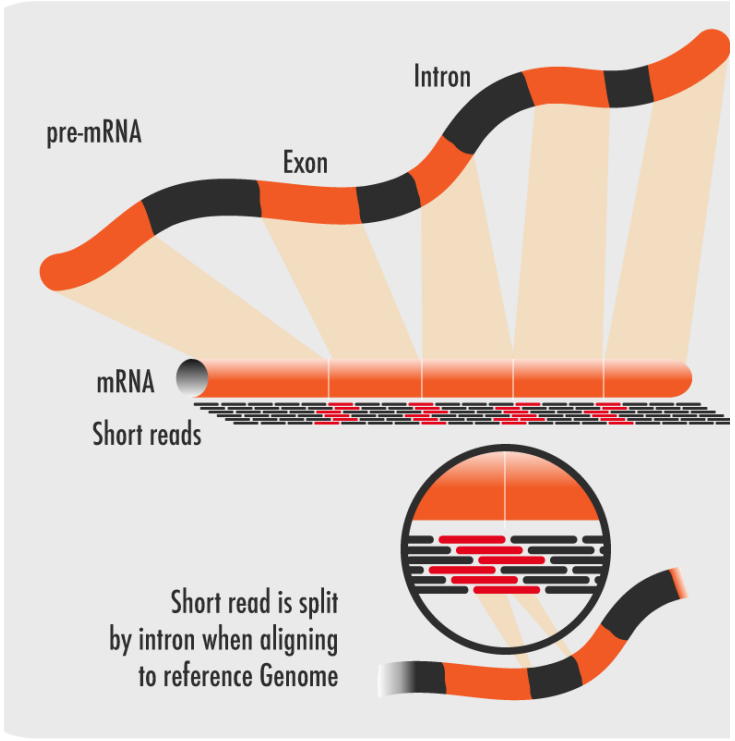
- RNA-seq Value
- Challenges
- NCBI Cloud Pipeline
- Lessons Learned
- Future Directions

Kim Pruitt, PhD; Acting Director, NCBI

ODSS Cloud Supplement PI meeting, January 18, 2024

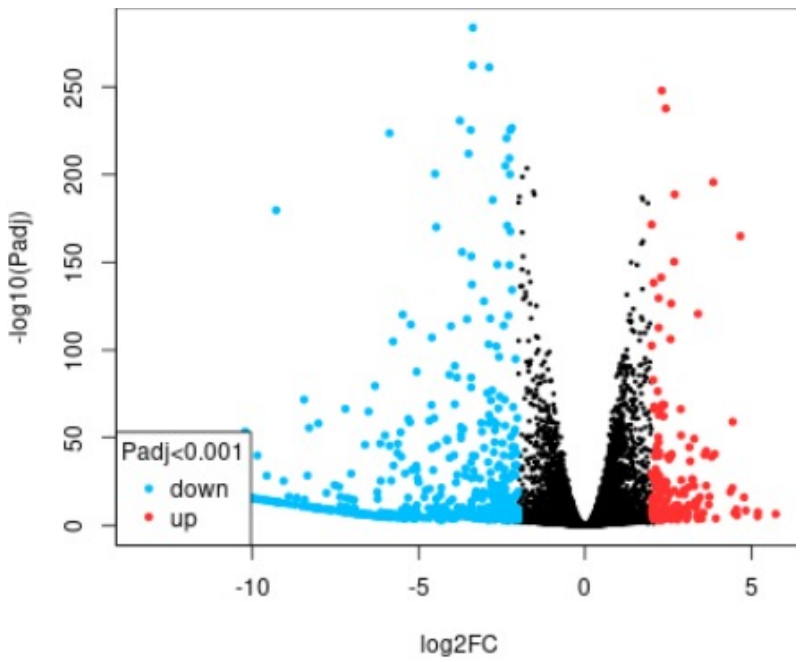
U.S. National Library of Medicine
National Center for Biotechnology Information

RNA-seq has revolutionized biomedical science

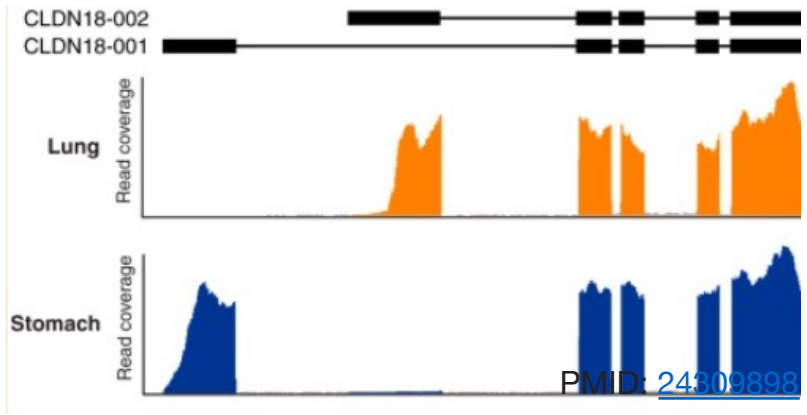


Credit: Technology Networks

RNA transformed into quantifiable short DNA sequence reads



Genomic-scale gene expression



Refined genome annotation

RNA-seq data pose challenges for data analysis and re-use

- SRA data files are large (1.5 GB average)
- Requires sufficient computing storage and power
- Requires specialized computing knowledge
- Pre-computed expression analysis results are more FAIR

NCBI's Cloud-based RNA-seq pipeline:

- ✓ Aligns SRA public human ~1.5Gb scale RNA-seq runs to genome assembly GRCh38.p13
- ✓ Analysis carried out on GCP
- ✓ Produces gene-level counts for each run in a ~560 Kb small file (*2600-fold reduction in data size*)
- ✓ Count data accessible from GEO
- ○ Count data will be given SRA unique identifiers
- ○ Count data will be delivered from the cloud, estimated Q3FY24

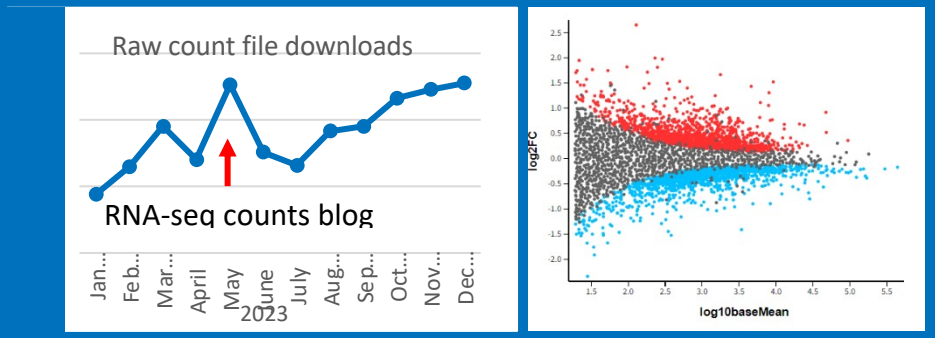
NCBI RNA-seq cloud analysis pipeline: Output to date

- 1.9 million human runs processed so far
- 1.3 million human runs passed 50% alignment threshold
- Each run processed with the pipeline:
  - Costs $0.16
  - Takes a few minutes
- ~25K GEO studies have counts available for download or analysis in GEO2R

GEO ACCESS

1. Access/analyze using GEO2R Tools
2. Download for local analysis

~3 fold-increase in file downloads from January – December 2023



RNA-seq in the cloud: lessons learned

- NCBI workforce development
- Quality control metrics design (will be released on Cloud)
- Data management (metadata, stable IDs, attribution, status, deployment)
- Advanced planning for access and search
- Learned how to better use existing data model for data delivery and provided feedback for making the model more robust

RNA-seq in the cloud: Future directions

- Release data on SRA cloud
- Pipeline will be updated to use SRA Lite format (will reduce cost by 84%, to about $.02 per run)
- Processing of 1.9 million mouse RNA-seq runs
- Improve UX with download button from GEO
- Raise awareness of data availability
- Acquire feedback with user research
- Publishing pipeline

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