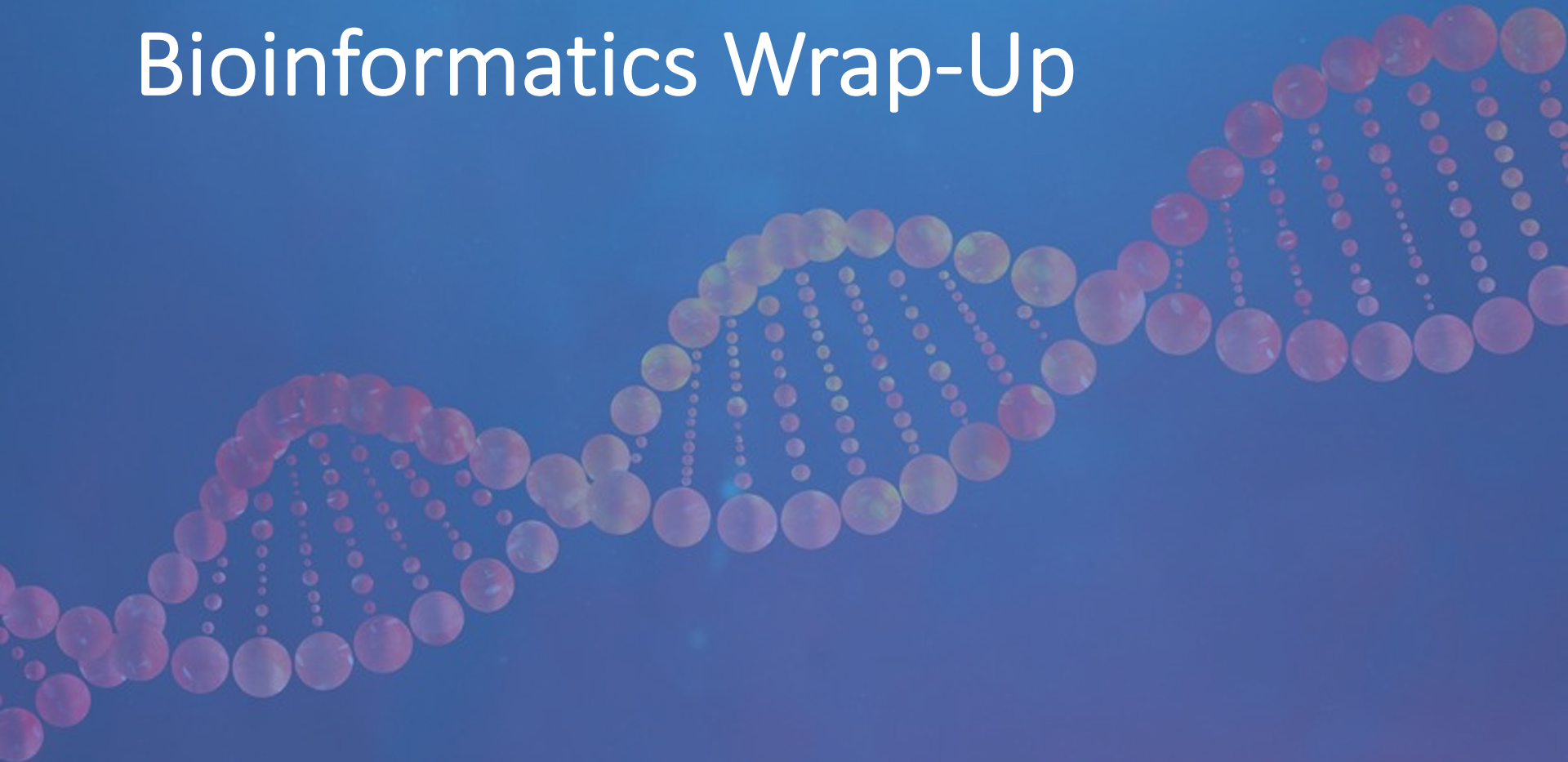


Bioinformatics Wrap-Up



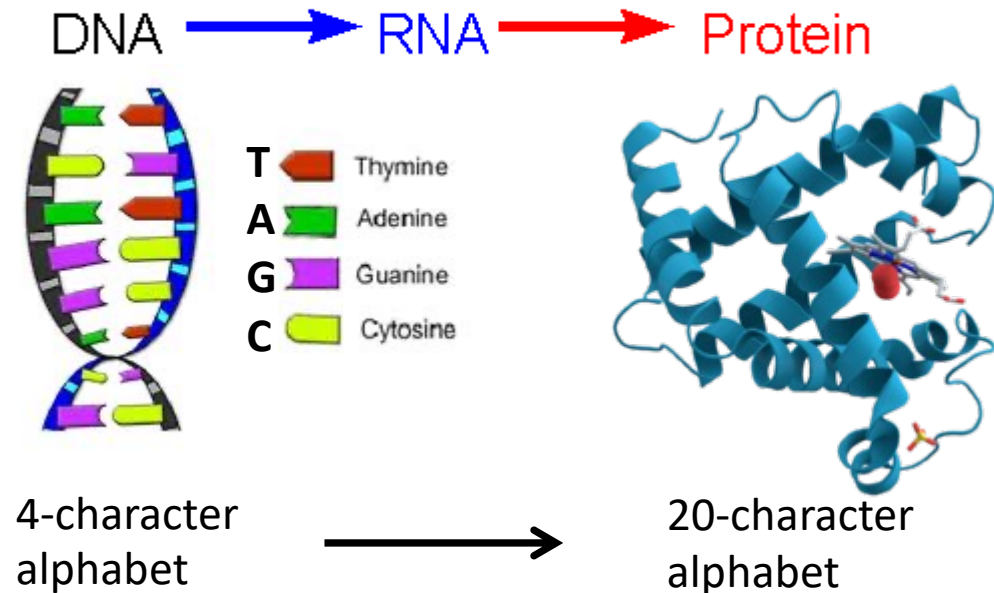
Stanislaw Pytel/Getty Images

Dr. Garrett Dancik

What is bioinformatics

- Bioinformatics:

- Biology + information
- the study and utilization of methods for storing, retrieving and analyzing biological data
- Falls under the category of Big Data



- How much information:

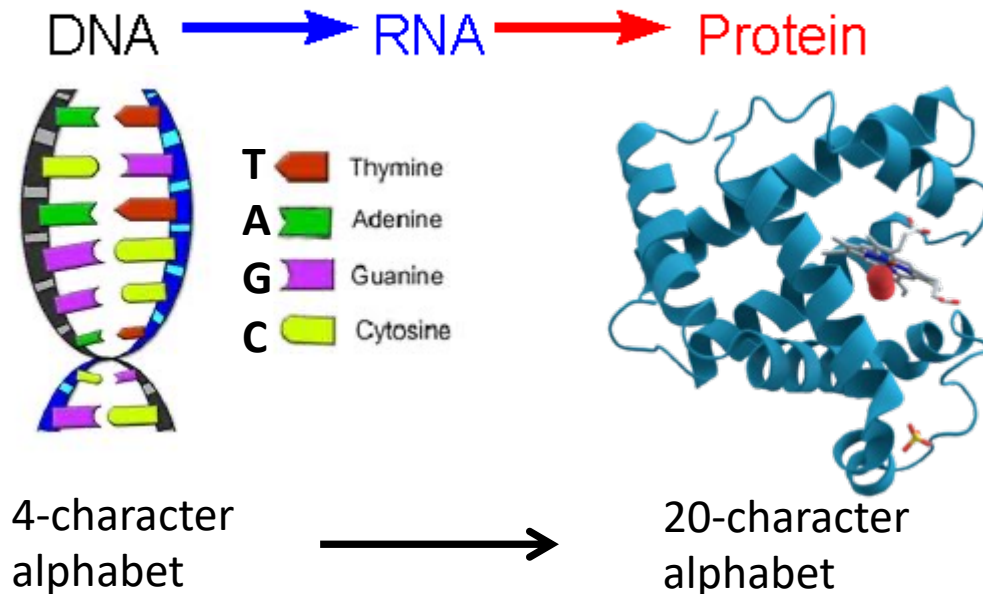
- Human genome: 3 billion nucleotides
- ~20,000 genes
 - many more when considering “junk DNA” and alternative splicing
- >10 million sites of DNA variation
- Countless possible interactions between DNA, RNA, and proteins

What is this?

```
print('hello world!')
```

- **Computer code** is a *set of instructions* that tells a computer how to process data and output results
- The **genetic code** is also a set of instructions, that tells a cell how to produce a molecule (such as a protein)
 - Information flows from DNA → RNA → protein
 - This information determines the structure/function of RNA and protein

Central Dogma of Molecular Biology



- The function of a protein can be predicted from its DNA or protein sequence
- Just like Java (or Python) is a language for computers, genetics is the language of life (DNA is the alphabet)
 - This is fundamental concept in bioinformatics

Bioinformatics example

- Let's look briefly at the gene *BRCA1* (breast cancer type 1 susceptibility protein)
 - <http://www.ncbi.nlm.nih.gov>
 - Search Nucleotide for BRCA1
 - Click on the Transcript reference sequence
- Hopefully this now makes sense!

Why do we need bioinformatics?



- To identify genetic mechanisms of diseases and other inherited (or acquired) conditions
 - Nature via nurture
- For personalized treatment of disease

Why do we need bioinformatics?

- BLAST



- Sequence Alignment

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Genome Res. 2014 Jul; 24(7): 1180–1192. PMCID: PMC4079973
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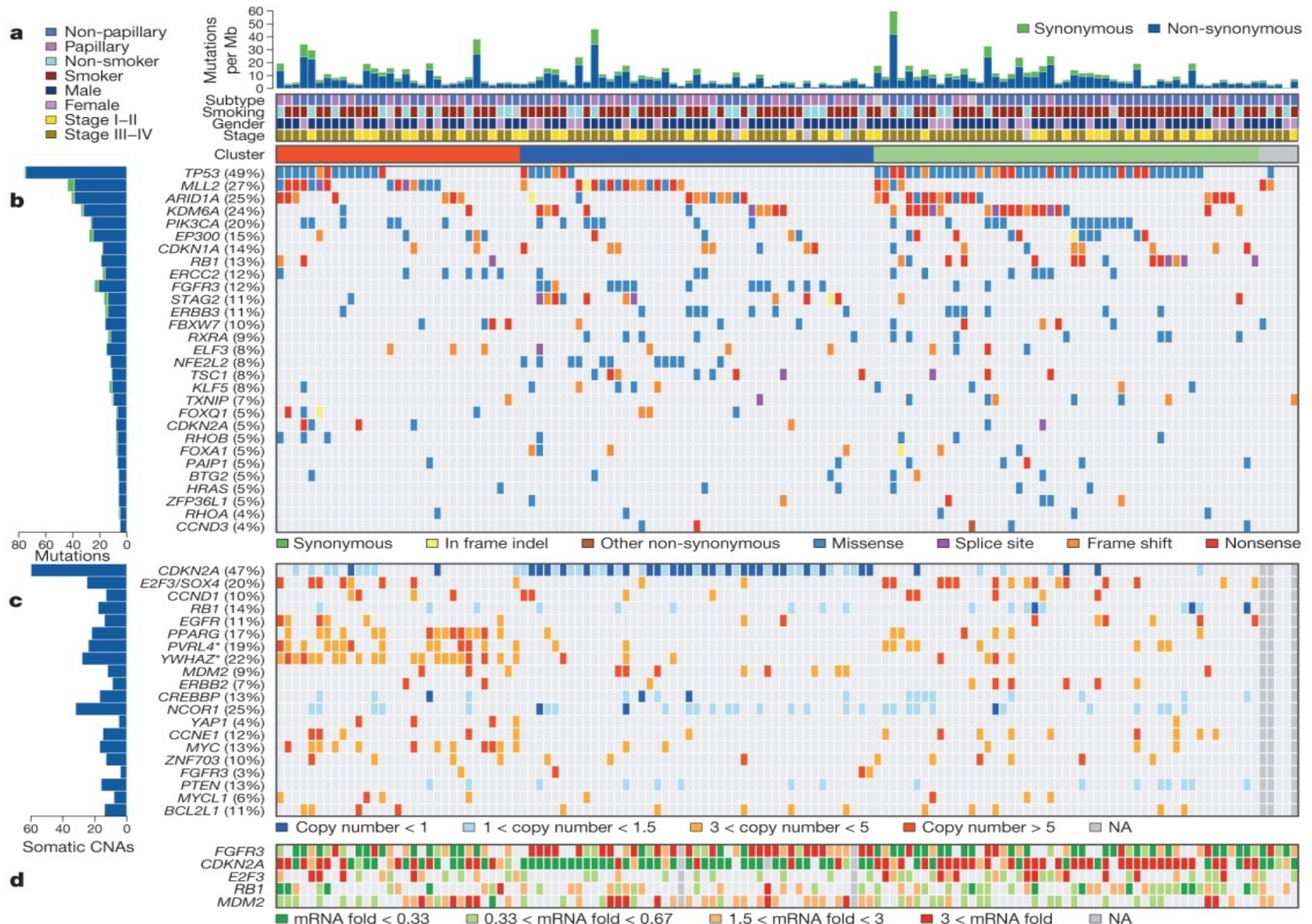
A cloud-compatible bioinformatics pipeline for ultrarapid pathogen identification from next-generation sequencing of clinical samples

[Samia N. Naccache](#),^{1,2} [Scot Federman](#),^{1,2} [Narayanan Veeraraghavan](#),^{1,2} [Matei Zaharia](#),³ [Deanna Lee](#),^{1,2} [Erik Samayoa](#),^{1,2} [Jerome Bouquet](#),^{1,2} [Alexander L. Greninger](#),⁴ [Ka-Cheung Luk](#),⁵ [Barryett Enge](#),⁶ [Debra A. Wadford](#),⁶ [Sharon L. Messenger](#),⁶ [Gillian L. Genrich](#),¹ [Kristen Pellegrino](#),⁷ [Gilda Grard](#),⁸ [Eric Leroy](#),⁸ [Bradley S. Schneider](#),⁹ [Joseph N. Fair](#),⁹ [Miguel A. Martínez](#),¹⁰ [Pavel Isa](#),¹⁰ [John A. Crump](#),^{11,12,13} [Joseph L. DeRisi](#),⁴ [Taylor Sittler](#),¹ [John Hackett, Jr.](#),⁵ [Steve Miller](#),^{1,2} and [Charles Y. Chiu](#),^{1,2,14,15}

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 Genome Res

The genomic landscape of bladder cancer



Bioinformatics and coronavirus

- Coronavirus genomic analysis suggests early spread in the United States
 - <https://www.the-scientist.com/news-opinion/coronaviruss-genetics-hint-at-its-cryptic-spread-in-communities-67233>
- Advanced bioinformatics rapidly identifies existing therapeutics for patients with coronavirus disease-2019 (COVID-19)
 - <https://translational-medicine.biomedcentral.com/articles/10.1186/s12967-020-02430-9>
- Combination of Biodata Mining and Computational Modelling in Identification and Characterization of ORF1ab Polyprotein of SARS-CoV-2 Isolated from Oronasopharynx of an Iranian Patient
 - <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7171442/>
- Computational Strategies to Combat COVID-19
 - <https://academic.oup.com/bib/advance-article/doi/10.1093/bib/bbaa232/5955939>

Additional Databases and Tools

- The Cancer Genome Atlas
 - <http://cancergenome.nih.gov>
 - Comprehensive genomic characterization of tumors
- cBioPortal for Cancer Genomics
 - <http://www.cbioportal.org/>
- Genomics of Drug Sensitivity in Cancer
 - <http://www.cancerrxgene.org>
 - Screen > 1000 cell lines with drug
 - Genomic characterization of cell lines
- And many more (current research)
 - <http://bioinformatics.oxfordjournals.org>
 - <http://www.biomedcentral.com/bmcbioinformatics>