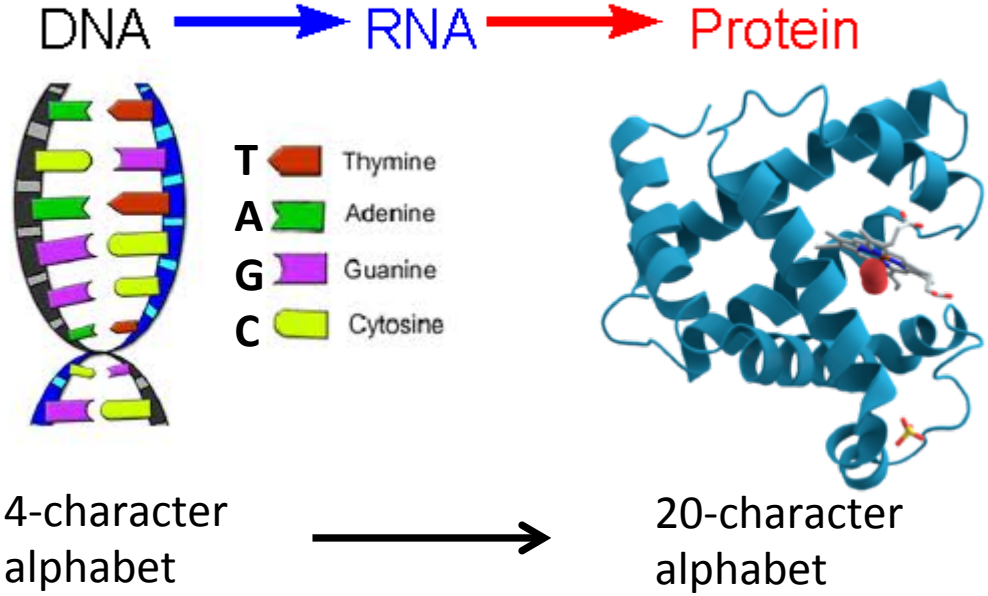
Bioinformatics Wrap-Up

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



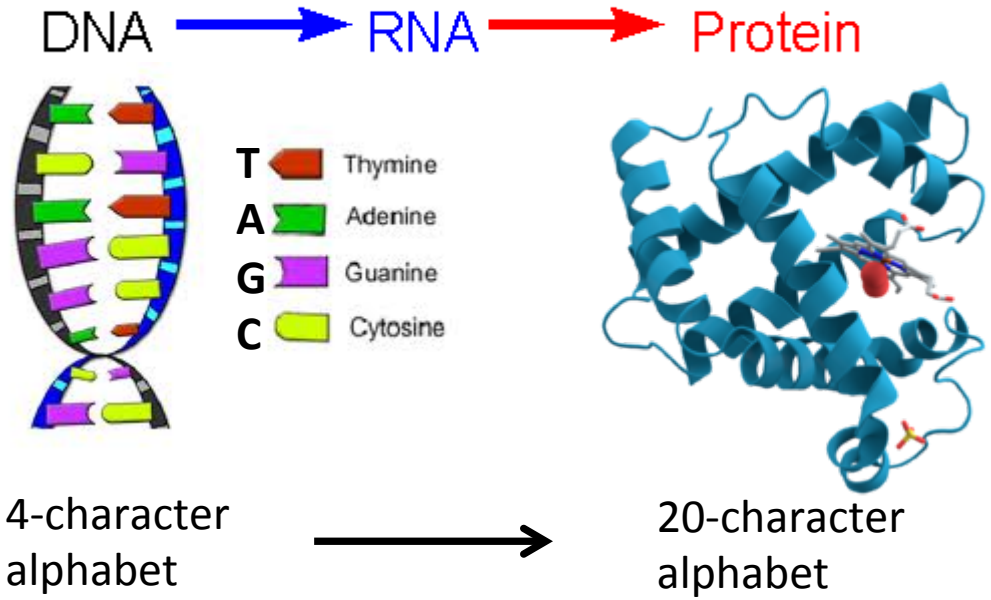
- How much information:
  - Human genome: 3 billion nucleotides
  - ~20,000 genes
    - many more when considering "junk DNA" and alternative splicing
  - >10 million sites of DNA variation
  - Countless possible interactions between DNA, RNA, and proteins

What is this?

```
#include <iostream>
using namespace std;
int main
{
cout << "hello world\n";
return 0;
}
```

- 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 C++ is a language for computers, genetics is the language of life (DNA is the alphabet)
  - This is fundamental concept in bioinformatics

Bioinformatics example

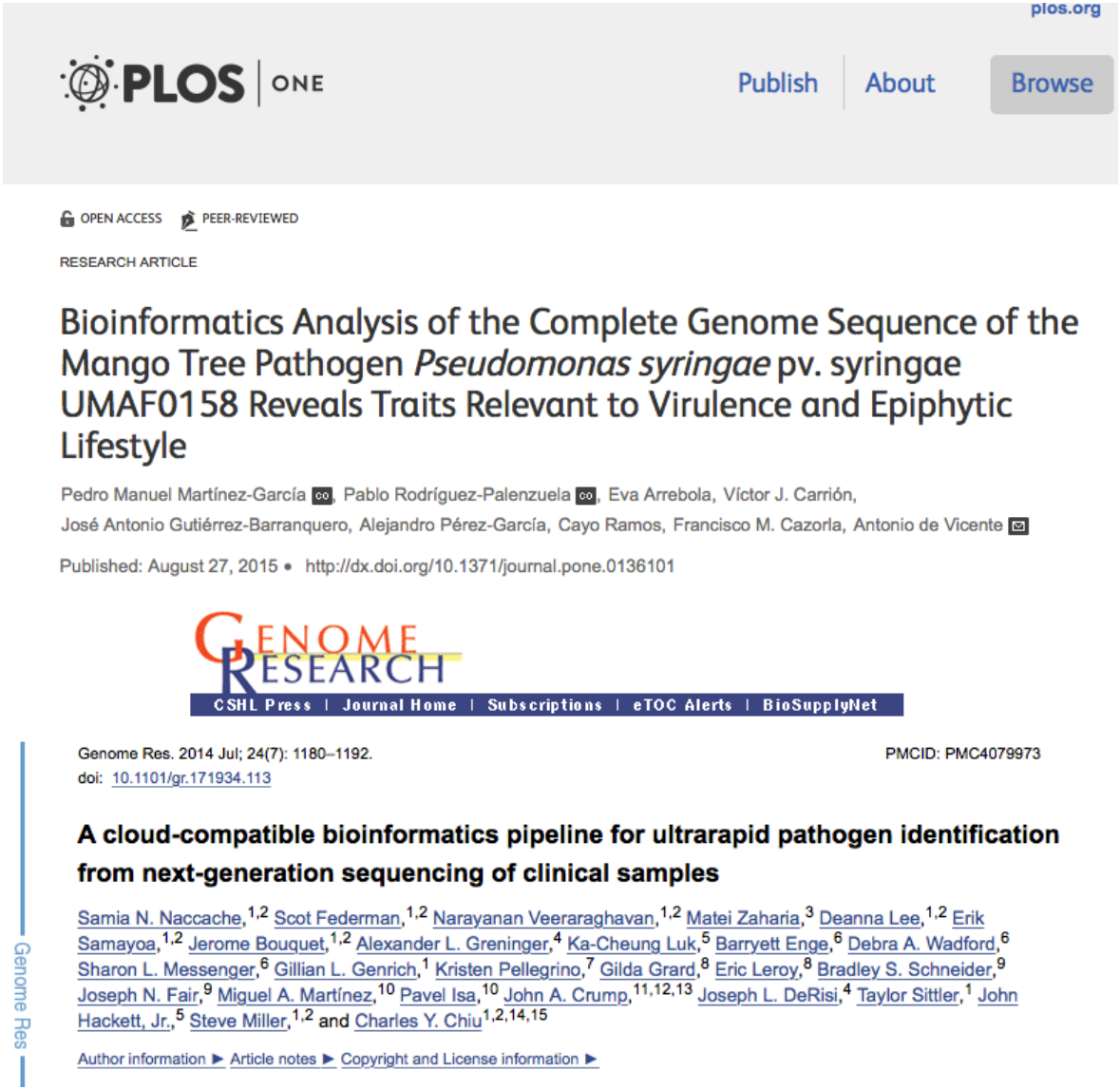
- Let's look briefly at the gene *BRCA1* (breast cancer type 1 susceptibility protein)
  - http://www.ncbi.nlm.nih.gov
  - Search Nucleotide for BRCA1
  - Click on the Genomic reference sequence
- Hopefully this now makes sense!

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 lab
- Python lab



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RESEARCH ARTICLE

Bioinformatics Analysis of the Complete Genome Sequence of the Mango Tree Pathogen *Pseudomonas syringae* pv. syringae UMAF0158 Reveals Traits Relevant to Virulence and Epiphytic Lifestyle

Pedro Manuel Martínez-García, Pablo Rodríguez-Palenzuela, Eva Arrebola, Víctor J. Carrión, José Antonio Gutiérrez-Barranquero, Alejandro Pérez-García, Cayo Ramos, Francisco M. Cazorla, Antonio de Vicente

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Genome Res. 2014 Jul; 24(7): 1180–1192. PMCID: PMC4079973

doi: 10.1101/gr.171934.113

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

- Group project

Cell Death and Differentiation (2007) 14, 662–670

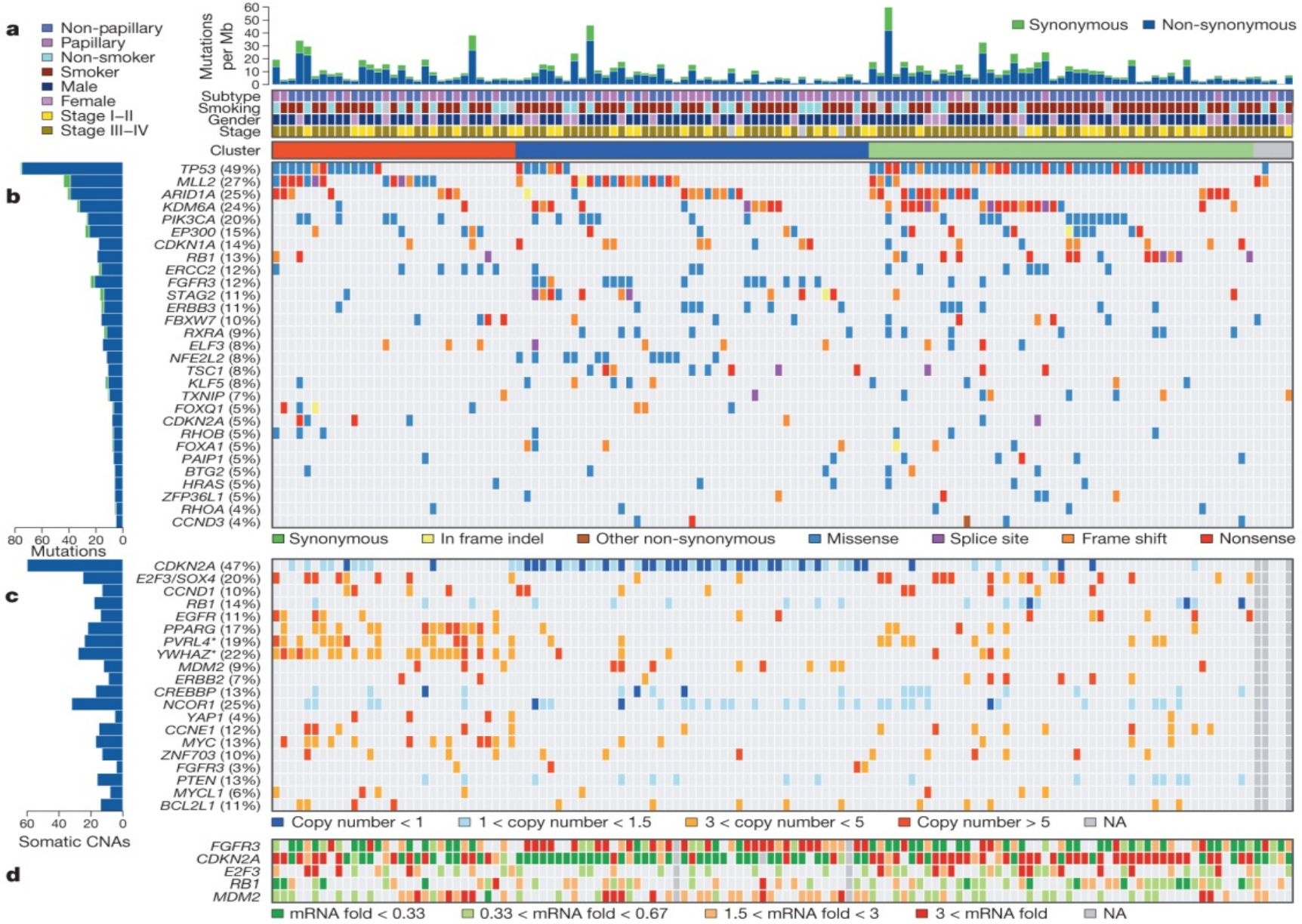
www.nature.com/cdd

Regulation of developmental rate and germ cell proliferation in *Caenorhabditis elegans* by the p53 gene network

WB Derry*,[1,2,3], R Bierings[1], M van Iersel[1], T Satkunendran[2,3], V Reinke[4] and JH Rothman*,[1]

p53 consensus binding sites in CEP-1-regulated genes. In an effort to identify putative direct targets of CEP-1, we analyzed CEP-1-activated genes for p53 consensus DNA-binding sites.[23] Vertebrate p53 binds two copies of the palindromic DNA consensus sequence RRRCWWGYYY, and CEP-1 binds an oligonucleotide containing the human p53 consensus motif *in vivo* and *in vitro*.[14,15] We searched the *C. elegans* genome for pairs of p53 consensus binding sites and found that ~25% of CEP-1-activated genes contain potential p53-binding sites in their promoters and introns (Table S2). These sites were

The genomic landscape of bladder cancer



The Cancer Genome Atlas Research Network *Nature* **507**, 315-322 (2014) doi:10.1038/nature12965

Additional Databases and Tools

- The Cancer Genome Atlas
  - http://cancergenome.nih.gov
  - Comprehensive genomic characterization of tumors
- 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