An Overview of Bioinformatics

BERDC Special Topics Talk 17

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DAKOTA COMMUNITY COLLABORATIVE ON TRANSLATIONAL ACTIVITY

Dr. Mark Williamson

Biostatistics, Epidemiology, and Research Design Core

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Introduction

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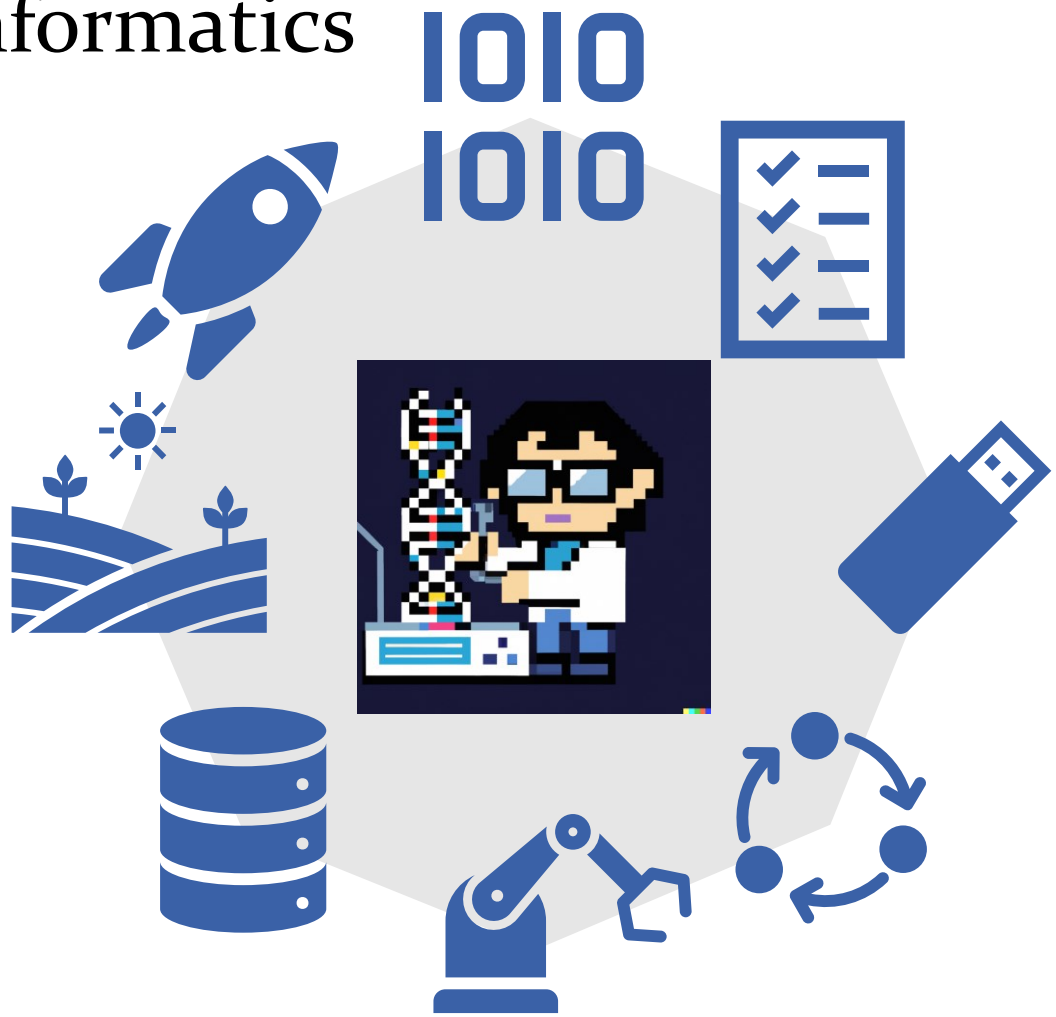
Goal: give an overview of Bioinformatics

- Definition & History
- Field & Topics
- Data
- Methods
- Machines & Software
- Databases
- Example Applications
- The Future



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Definition & History

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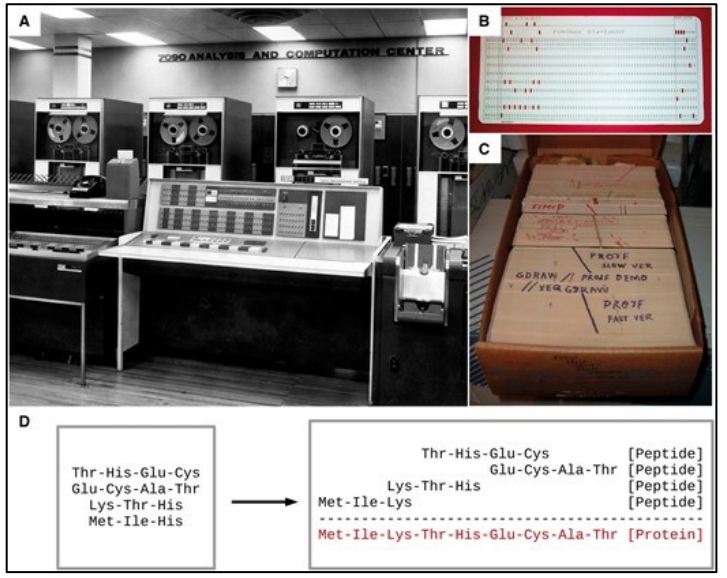
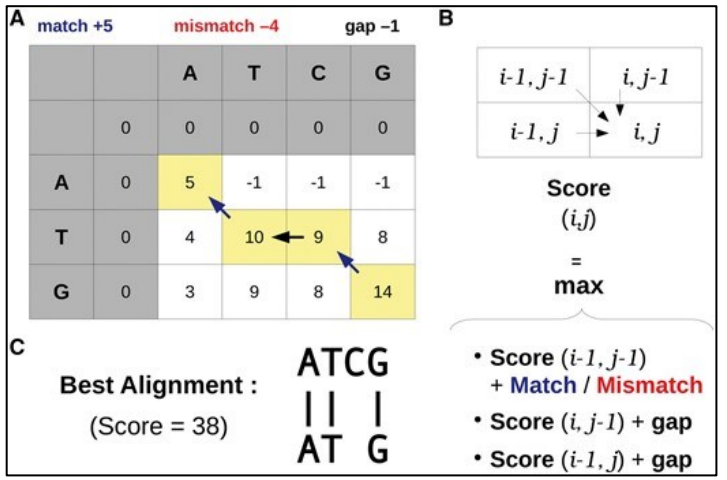
"the collection, classification, storage, and analysis of biochemical and biological information using computers especially as applied to molecular genetics and genomics" –Merriam-Webster

- 1960-1970: Protein analysis origins and Margaret Dayhoff [1]
- 1970-1980: Shift from protein to DNA analysis
- 1980-1990: Advances in biology and computing
- 1990-2000: Genomics and the internet
- 2000-2010: High-throughput bioinformatics (2nd gen sequencing)
- 2010-Today: Big data, role of bioinformaticians, systems biology





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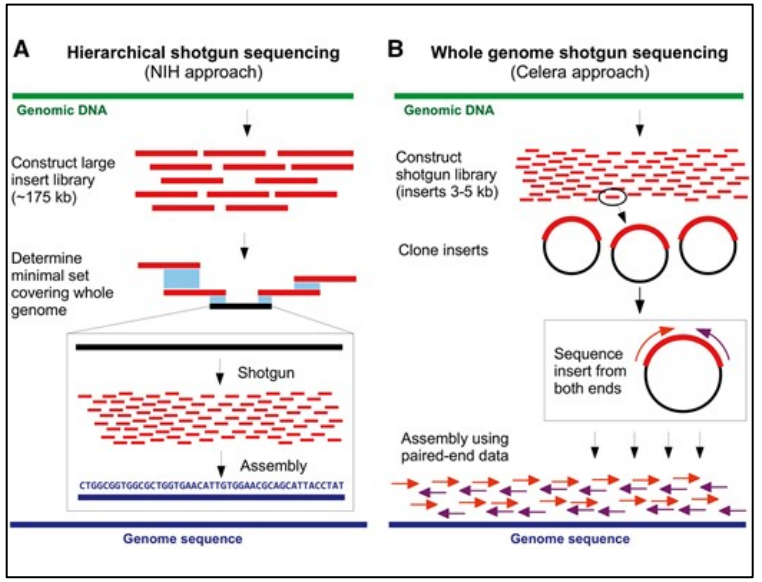

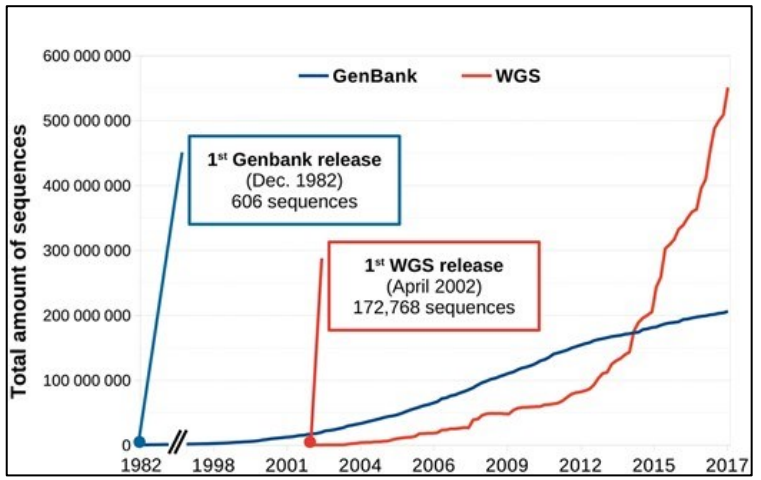

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Fields & Topics

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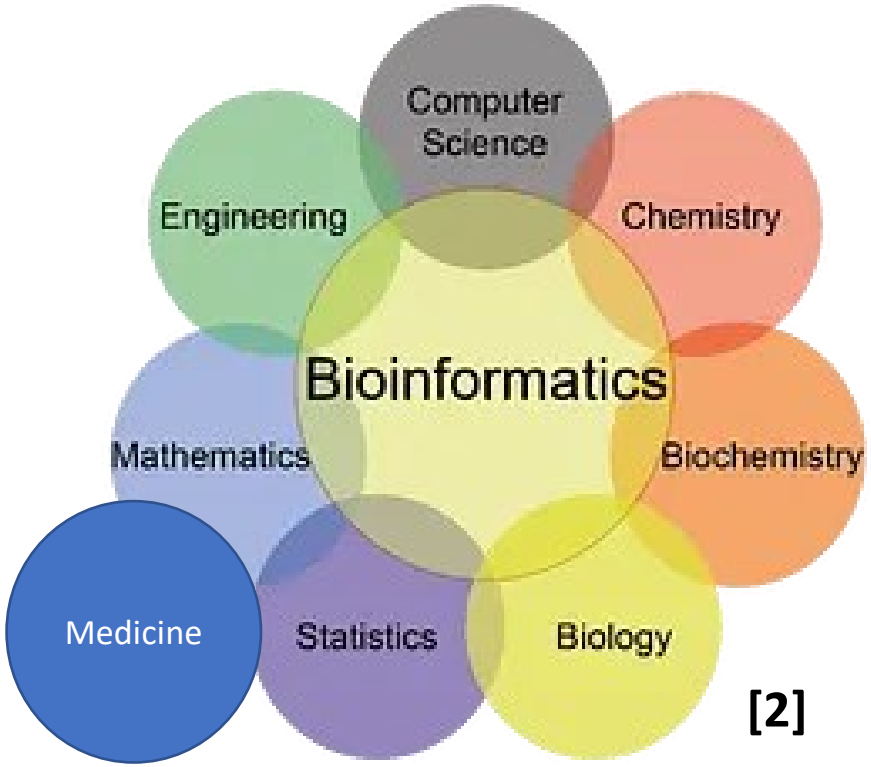

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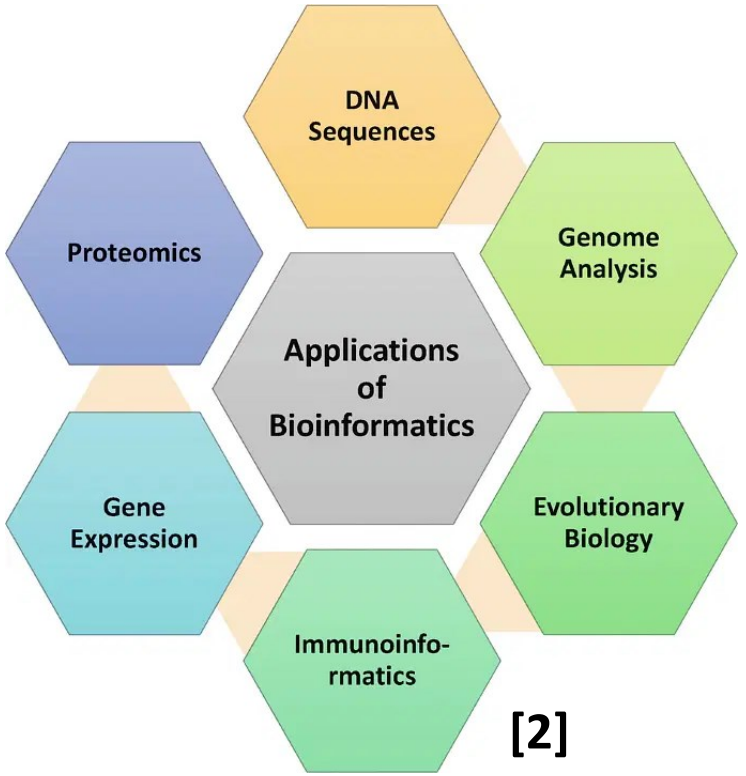

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Fields & Topics

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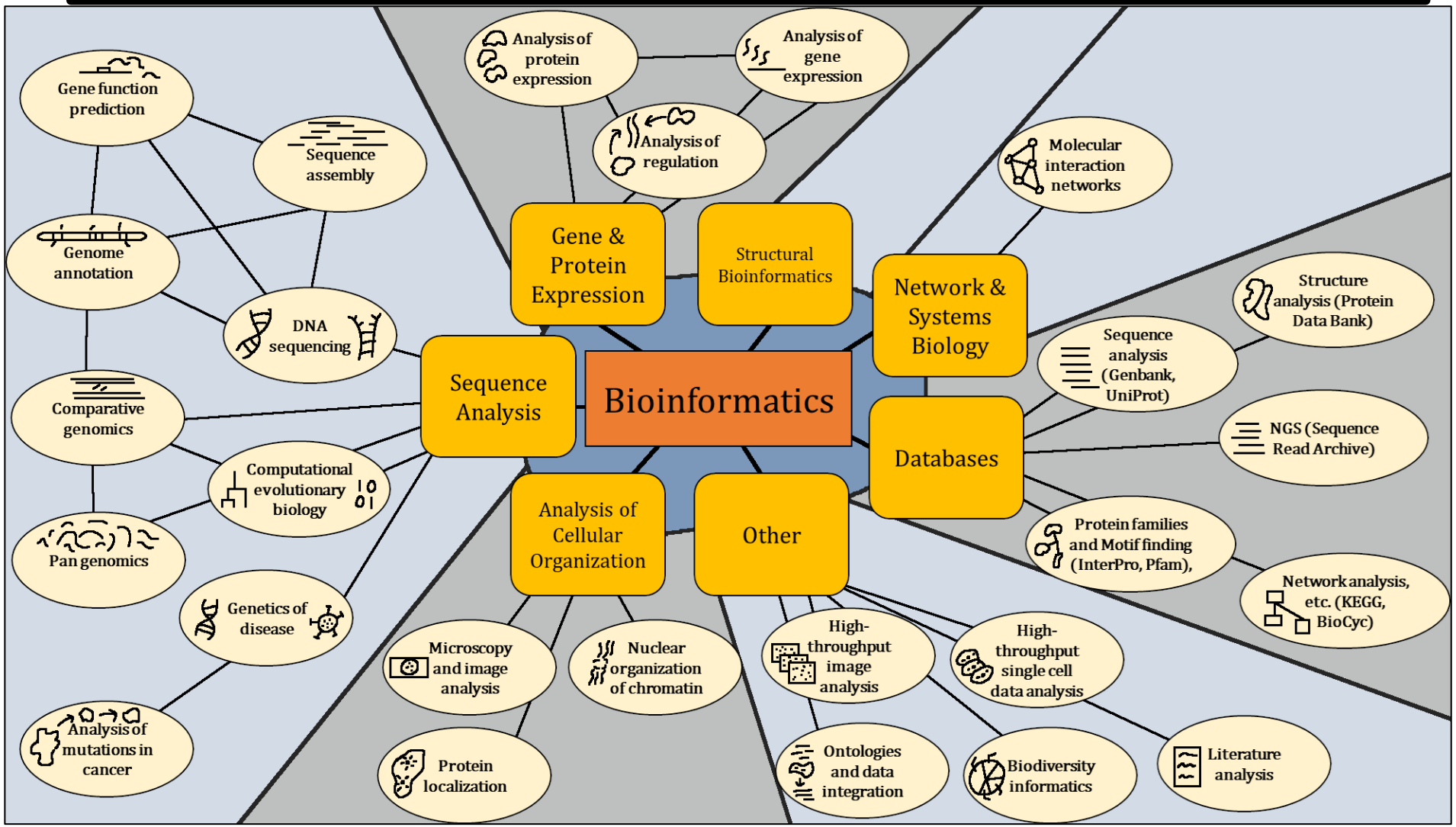

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Data

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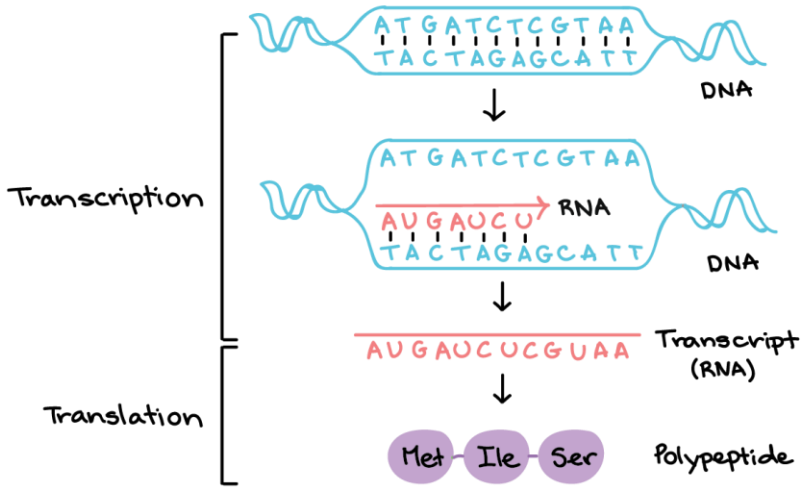
- Biological sources
  - DNA: sequences, genes, genomes, metagenomes
  - RNA and Proteins
  - Others [4]
    - Metadata
    - Images
    - Etc.
- File formats [5]
  - FASTA and FASTQ
  - Alignments (BAM, SAM, CRAM)
  - Other (VCF, GFF, GTF, BED, Tar.gz, PDB, PED, MAP, CSV, JSON)



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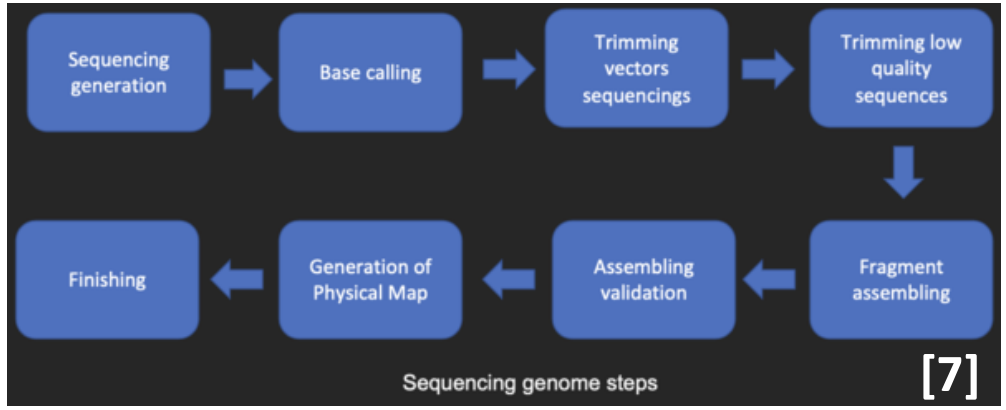
TGTTACGACTTCGTCCCAATCGCCAGTCCCACCTTCGAC
AGCTCCCTCCCACAAGGGGTTGGGCCACCGGCTTCGGGTG
TTACCGACTTTCGTGACGTGACGGGCGGTGTGTACAAGG
CCCGGGAACGTATTCACCGCAGCAATGCTGATCTGCGATT
ACTAGCAACTCCGACTTCATGGGGTCGAGTTGCAGACCC
CAATCCGAACTGAGACCGGCTTTTTGAGATTCGCTCCGCCT
CGCGGCATCGCAGCTCATTGTACCGGCCATTGTAGCAC
GTGTGCAGCCCAAGACATAAGGGGCATGATGACTTGACG
TCGTCCCCACCTTCCTCCGAGTTGACCCCGGCAGTCTCCT
GTGAGTCCCCATCACCCCGAAGGGCATGCTGGCAACACA
GAACAAGGGTTGCGCTCGTTGCGGGACTTAACCCAACATC
TCACGACACGAGCTGACGACAGCCATGCACCACCTGTATA
CCGACCACAAGGGGGGCACCATCTCTGATGCTTTCCGGT

Methods

- Pipelines
- DNA Sequencing
  - Amplicon sequencing: 16/18S, single copy, etc.
  - Genome sequencing: reference based, *de novo*
  - Annotation (what's there)
  - Comparison (what's different between samples/organisms)
- Gene and Protein expression
  - RNA-Seq
  - Microarray
- Phylogeny
- Networks
- Omics



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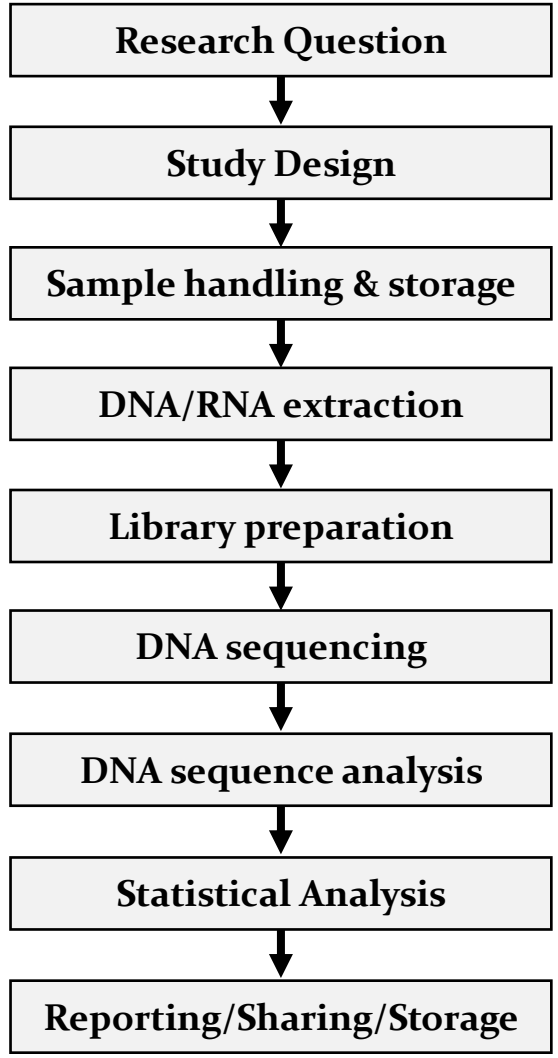

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Machines

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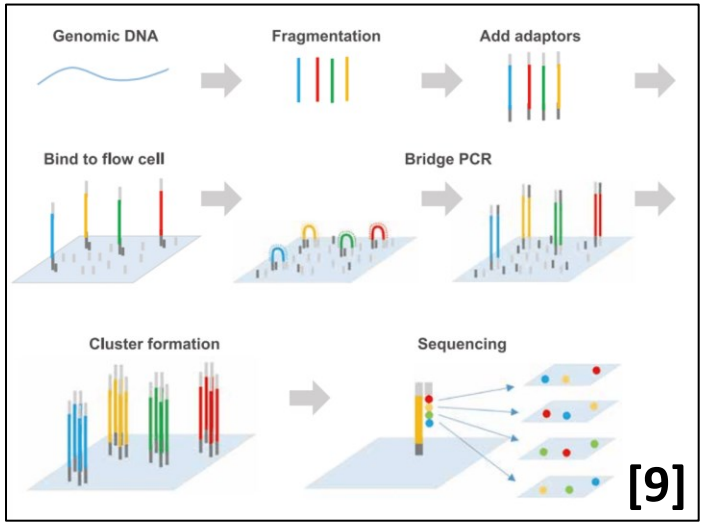
- DNA Sequencing [8]:
  - Illumina
  - Ion Torrent
  - Oxford Nanopore
  - Pacific Biosystems
- Mass Spectrometry
- Flow Cytometry



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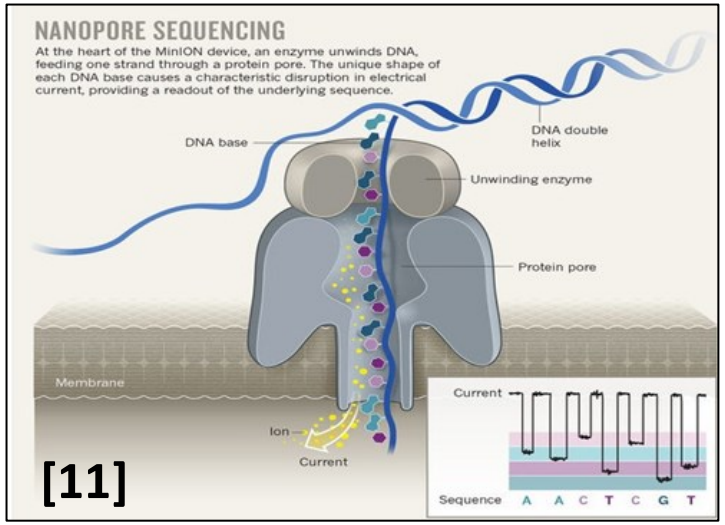

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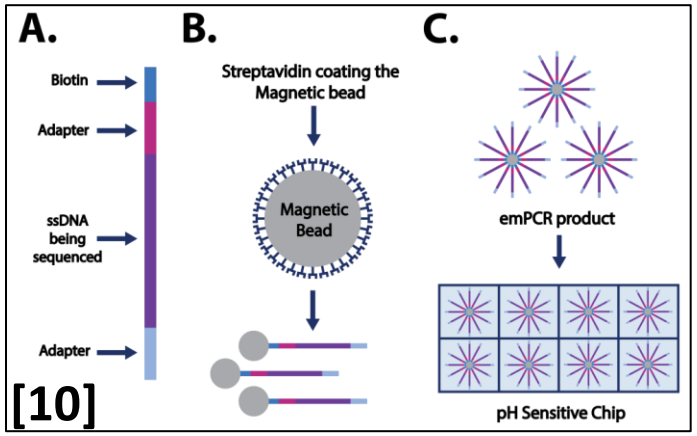

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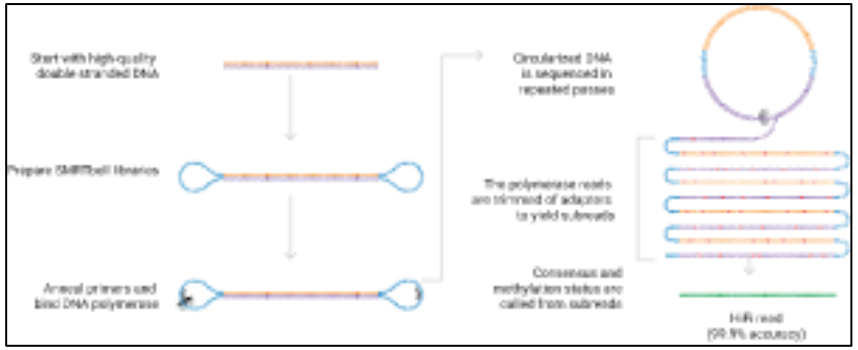
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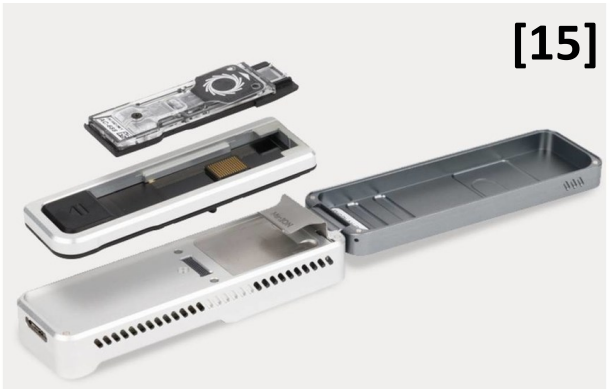
[13]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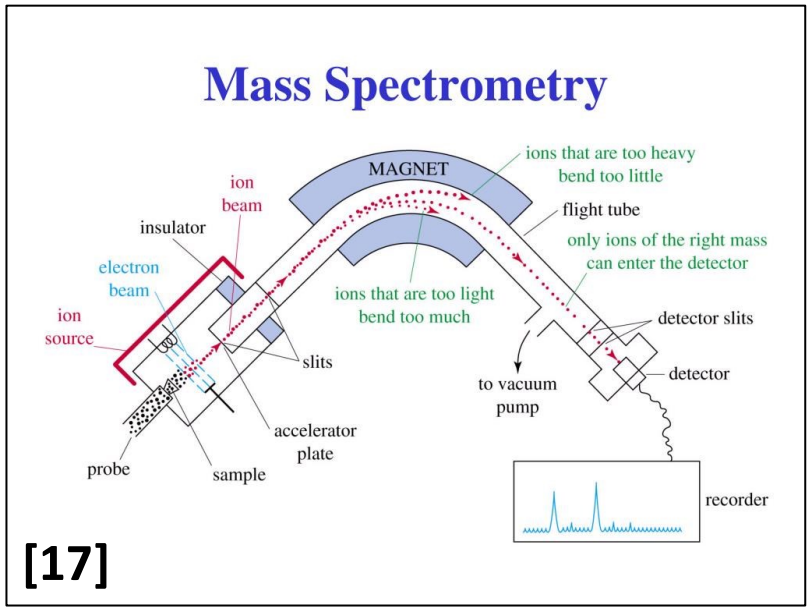

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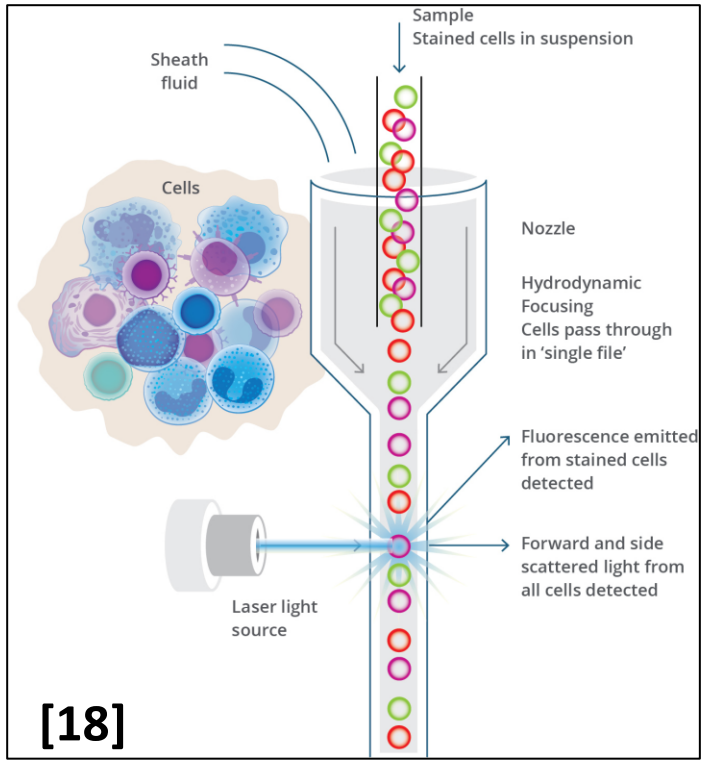

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Software

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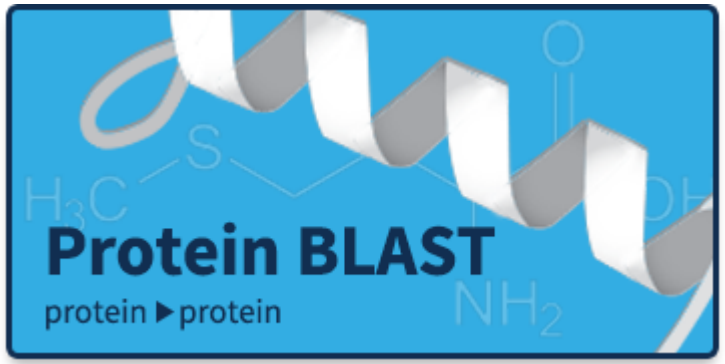
- **Core Tools:** BLAST, EMBOSS, Clustal Omega, PROSPECT, Ensembl, Usearch, SAMtools, Velvet, Bowtie, TopHat, Muscle, ProteinTools, etc.
- **Underlying Code:** Needleman-Wunch, Borrows-Wheeler Transform, UPGMA, Neighbor-joining, etc.
- **Software Environments:** Biopython, Bioconductor, BioPerl, etc.
- **Pipelines:** Dada2, DNA-seq Analysis, Center for Genomic Epidemiology, Qiime2, Mothur, Workflow Managers, etc.



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biopython

Bioconductor
OPEN SOURCE SOFTWARE FOR BIOINFORMATICS

BioPerl

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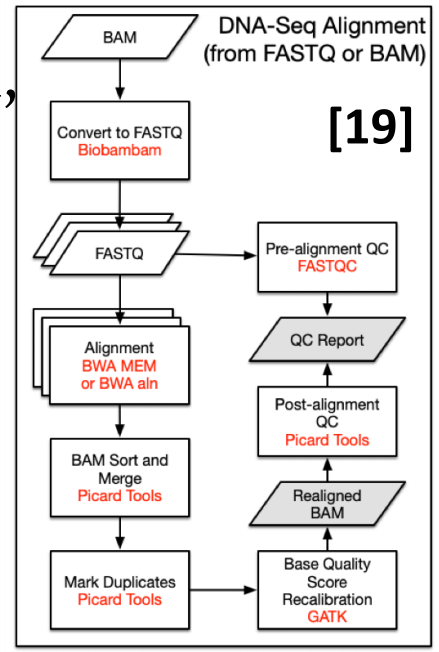
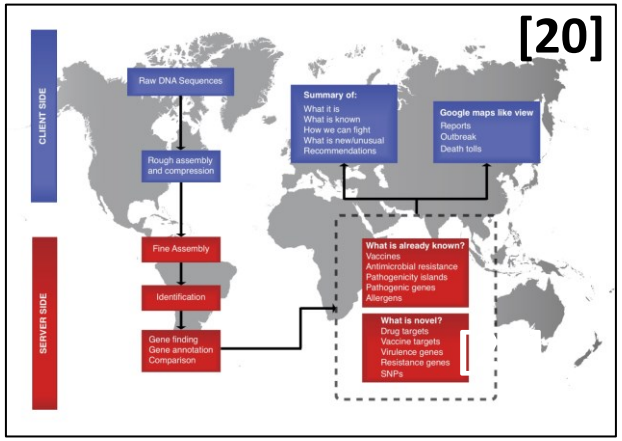


qiime2



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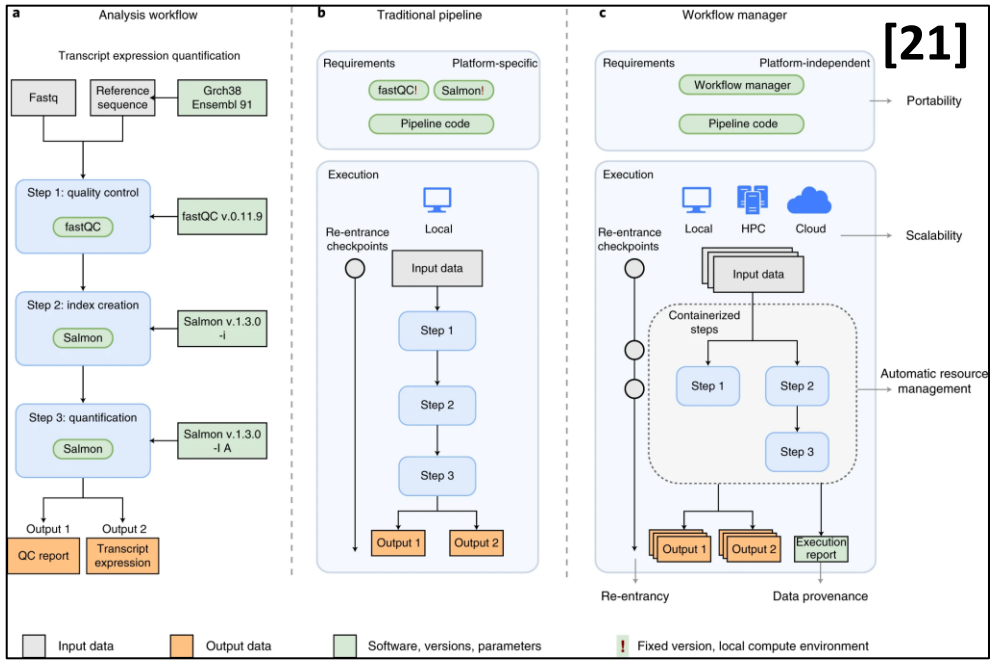

[21]

Galaxy PROJECT [22]

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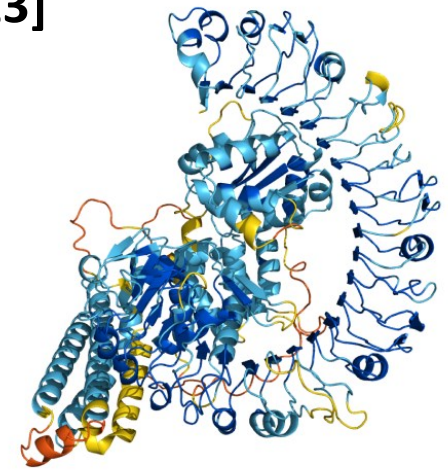
Databases

- **Entities:** NCBI, EMBL, Japanese Institute of Genetics
- **DNA:** GenBank, RefSeq
- **Proteins:** UniProt, Alphafold
- **Pathways:** KEGG
- **Taxonomy:** NCBI Taxonomy, Catalogue of Life
- **Interesting:** rrnDB, GTDB, silva

NCBI

[23]



KEGG Kyoto Encyclopedia of Genes and Genomes

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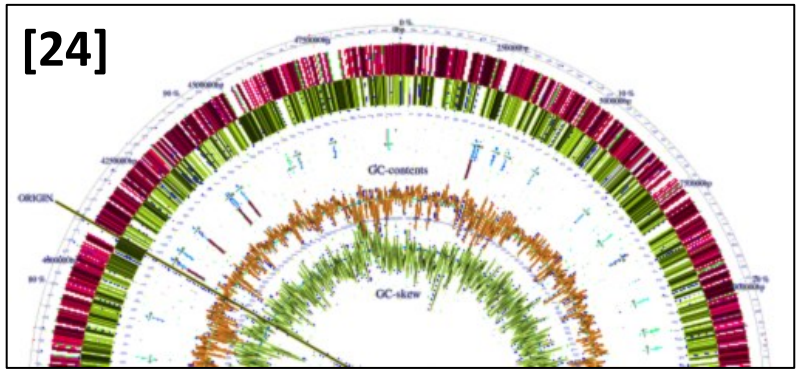

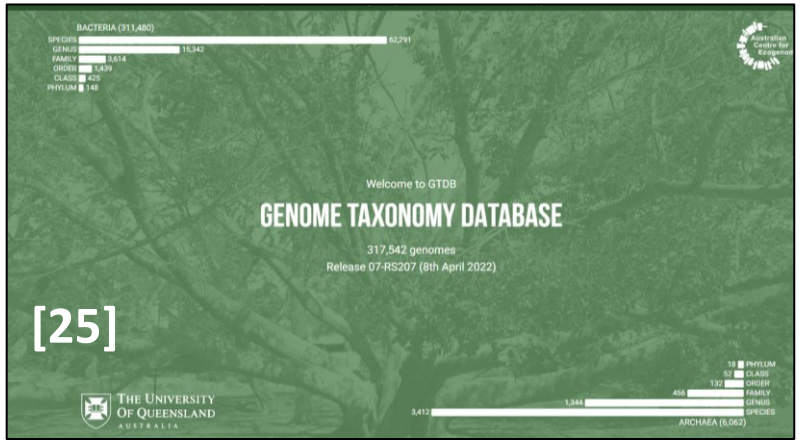

[25]

silva
high quality ribosomal RNA databases

[26]

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Example Applications

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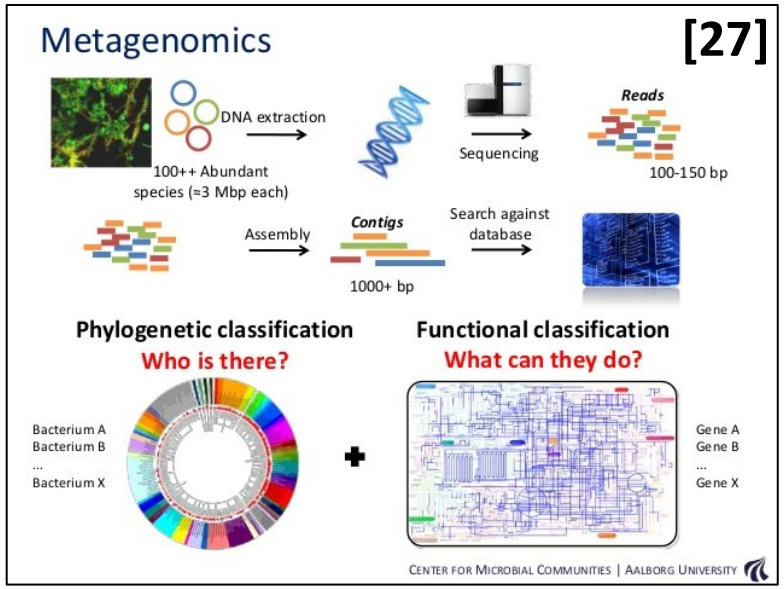
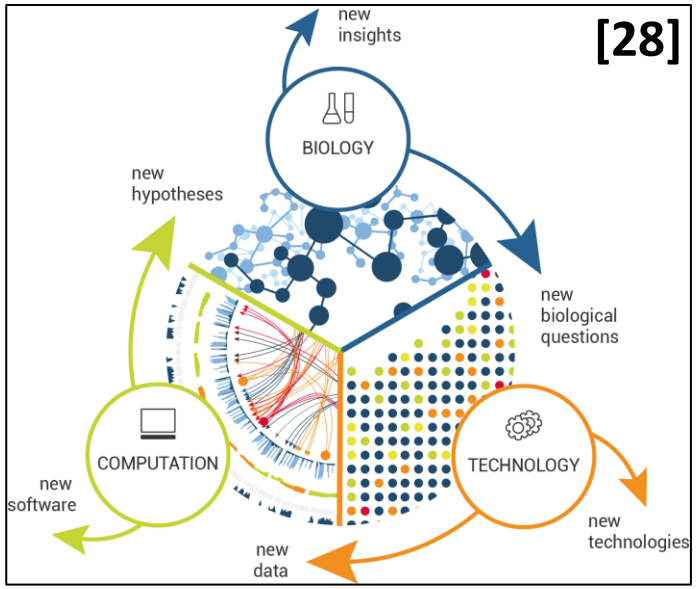
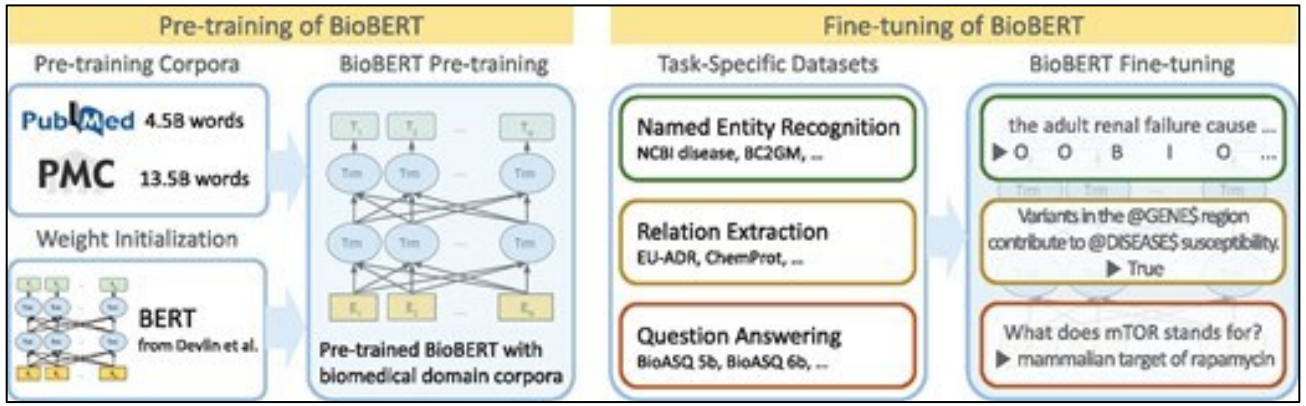
- Metagenomics
- Systems Biology
- Literature Analysis
- Microbiome







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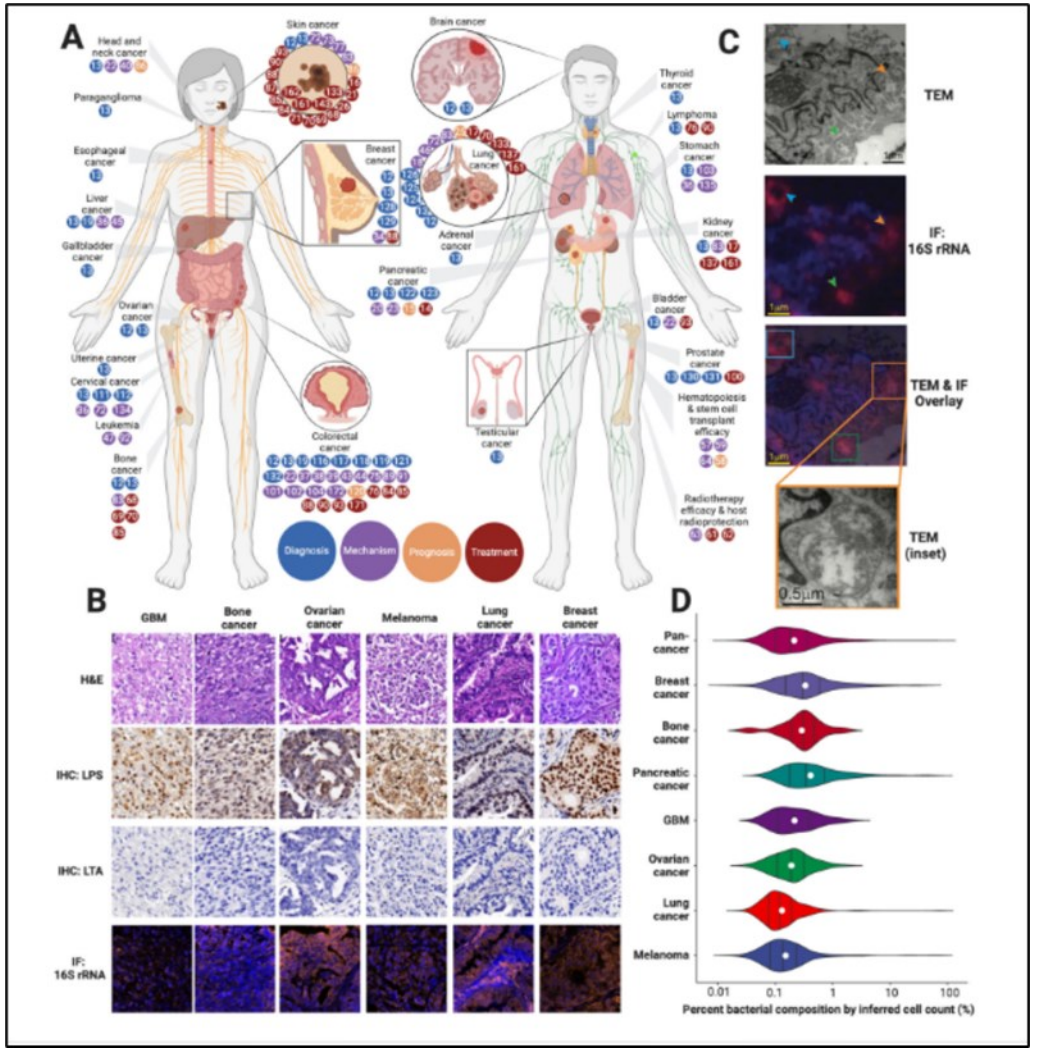

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The Future

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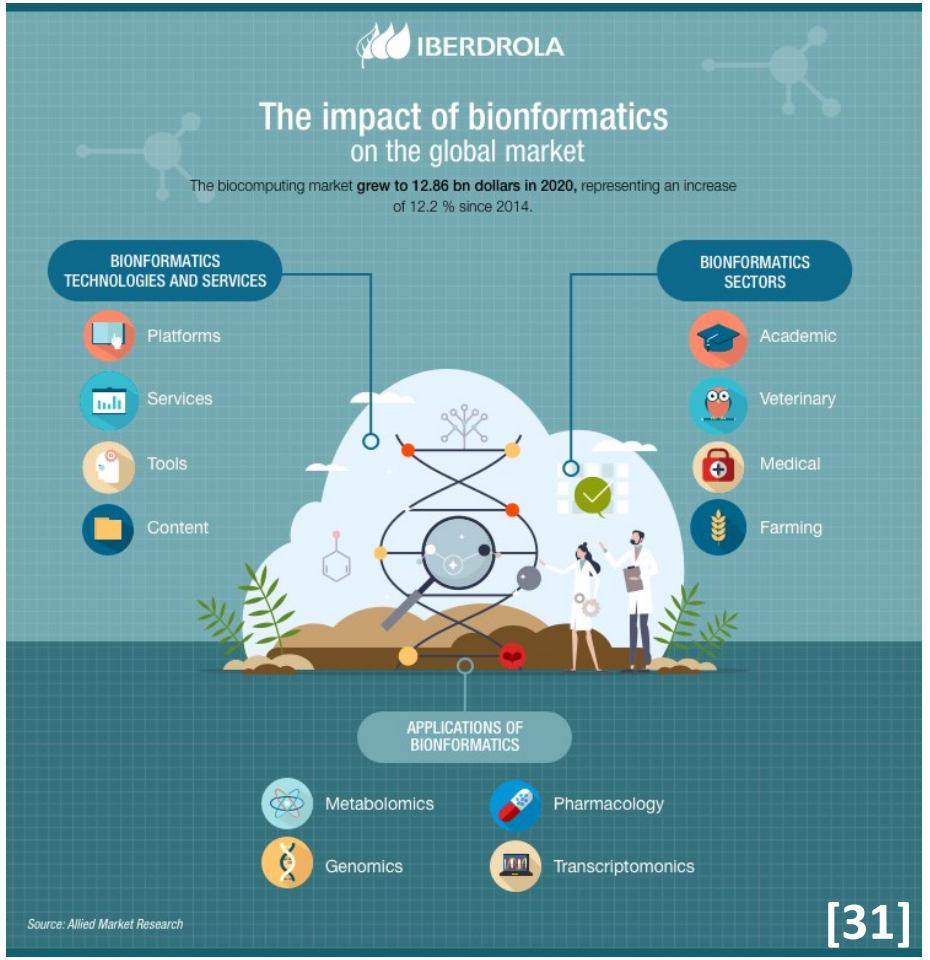
- Increasing market share
- Use of robotics, machine learning, deep learning, AI, etc. [32]
- More data, more integration
- The future is long? [33]



[31]

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Resources

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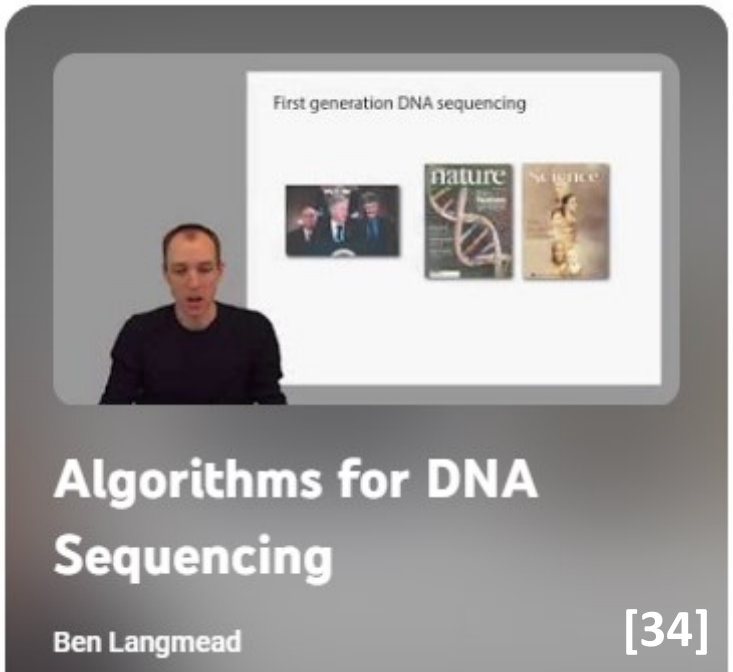
- Algorithms for DNA Sequencing
- Joint Genome Institute
  - NeLLi 2021 [35]
  - VEGA Symposium
  - Annual Genomics of Energy & Environment Meeting
- Center for Genomic Epidemiology (TDU)
  - Metagenomics applied to surveillance of pathogens and antimicrobial resistance
  - Whole genome sequencing of bacterial genomes-tools and applications
- Bioinformatics.ca [36]

bioinformatics.ca

JGI JOINT GENOME INSTITUTE
A DOE OFFICE OF SCIENCE USER FACILITY

Center for Genomic Epidemiology

Home Services Publications Contact



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References 1

[1] https://pubmed.ncbi.nlm.nih.gov/30084940/

[2] https://collegedunia.com/exams/bioinformatics-biology-articleid-1454

[3] https://sites.google.com/site/obenscience7e/_/rsrc/1490190066963/unit-7/from-dna-to-protein/dna%20to%20rna%20to%20protein.png

[4] https://www.ebi.ac.uk/ols/ontologies/edam

[5] https://www.formbio.com/blog/your-essential-guide-different-file-formats-bioinformatics

[6] https://www.coursera.org/learn/wgs-bacteria/

[7] https://en.wikipedia.org/wiki/Bioinformatics

[8] https://genohub.com/ngs-instrument-guide/

[9] https://blogs.iu.edu/ncgas/files/2021/09/Picture1.png

[10] https://i0.wp.com/apollo-institute.org/wp-content/uploads/2021/11/IonTSeq-Figure-1-01-01.png?resize=1024%2C636&ssl=1

[11] https://www.medgadget.com/wp-content/uploads/2020/04/nanopore.png

[12] https://www.pacb.com/technology/hifi-sequencing/

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References 2

[13] https://img.medicalexpo.com/images_me/photo-g/83632-12919744.jpg

[14] http://genearrays.com/wp-content/uploads/2014/03/pgm.jpg

[15] https://mma.prnewswire.com/media/834434/Oxford_Nanopore_Flongle.jpg?p=facebook

[16] https://www.pacb.com/revio/

[17] https://image.slideserve.com/464232/mass-spectrometry-l.jpg

[18] https://www.streck.com/wp-content/uploads/2020/11/flow-cytometry-light-scatter-illustration.jpg

[19] https://docs.gdc.cancer.gov/Data/Bioinformatics_Pipelines/DNA_Seq_Variant_Calling_Pipeline/

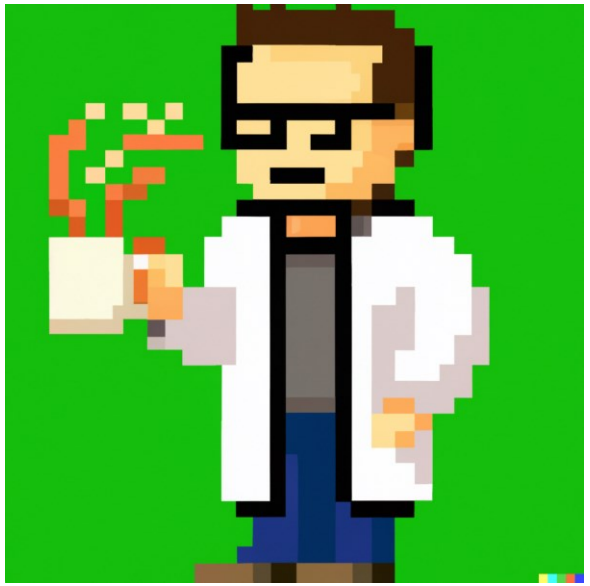
[20] https://www.genomicepidemiology.org/

[21] https://www.nature.com/articles/s41592-021-01254-9/figures/1

[22] By Anton Nekrutenko, http://galaxyproject.org/, Fair use, https://en.wikipedia.org/w/index.php?curid=32778899

[23] https://alphafold.ebi.ac.uk/

[24] https://rrndb.umms.med.umich.edu/



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References 3

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Acknowledgements

The DaCCoTA is supported by the National Institute of General Medical Sciences of the National Institutes of Health under Award Number U54GM128729.

For the labs that use the Biostatistics, Epidemiology, and Research Design Core in any way, including this Module, please acknowledge us for publications: ***"Research reported in this publication was supported by DaCCoTA (the National Institute of General Medical Sciences of the National Institutes of Health under Award Number U54GM128729)"***

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