



# Foresights on Human Genomics in 2030

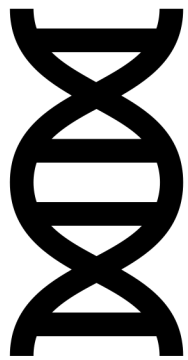
**Benilton S Carvalho**

May, 16<sup>th</sup> 2024

Regional meeting  
Human Genomics for Health:  
Enhancing the Impact of Effective Research

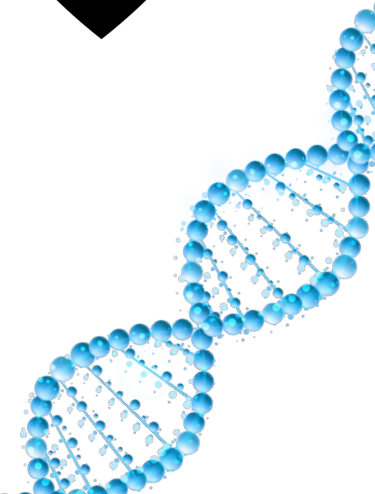
# Some Recent Advances

## NGS



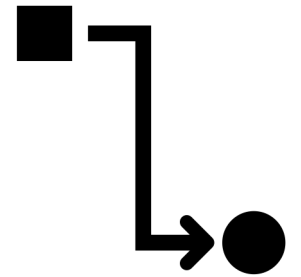
- Accelerated genetic mapping
- Cost reductions
- Accessibility increase
- Personalized treatments based on genomic profiles
- Increased therapeutic efficacy

## Precision Medicine



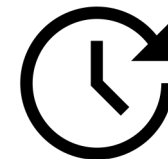
# Impacts on Research and Treatments

- Early identification of genetic diseases and cancers
- Improvement of prognoses
- Development of specific treatments for individuals
- Increase of success rates
- Reduction of side effects
- Advances in understanding complex diseases



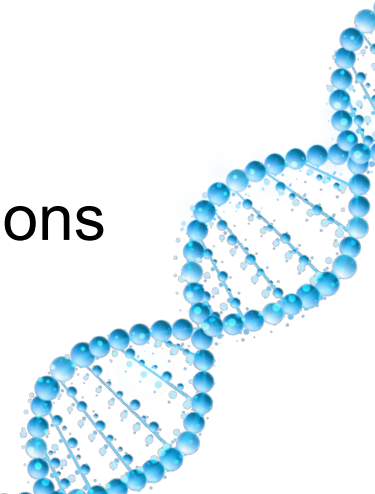
# Technological Predictions for 2030

- Faster and more accessible sequencing
  - Continued cost reduction
  - Increased sequencing speed
  - Greater throughput
- Advances in genomics
  - Powerful data analysis algorithms
  - Improved data-sharing policies
  - More statistical power
- AI for genomics
  - Identify patterns in large genomic datasets
  - Better disease predisposition prediction
  - Treatment suggestions



# Future of Personalized Medicine

- Gene therapies (will)
  - become more common
  - be cheaper
  - offer cure to some genetic diseases
  - treat some cancers
- Preventive medicine
  - Genomic profiles will allow for personalized disease prediction and prevention
  - Early interventions and lifestyle changes will avoid chronic conditions



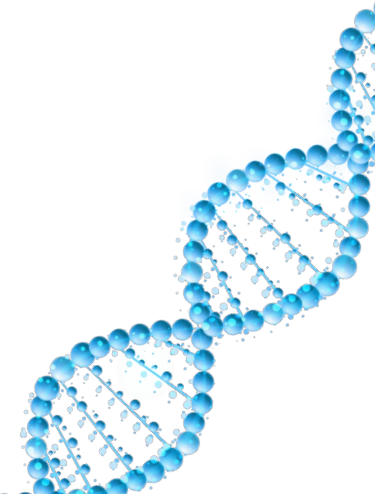
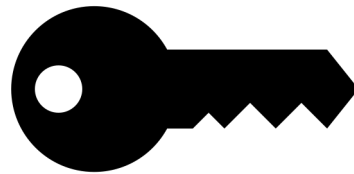
# Genomic Data Management

- Robust infrastructure
  - Hardware
  - Software
- Advanced tools for
  - Data storage
  - Data processing
  - Data analysis
- Advanced Statistical Methods
  - Data collections
  - Correlation mapping
  - Bigger cohorts and  $n = 1$



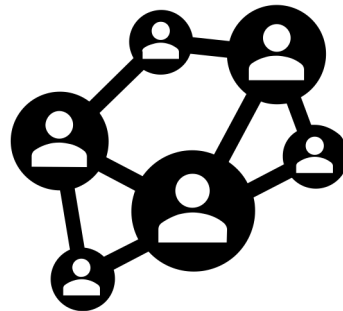
# Ensuring Equitable Access

- Public Policies
  - Ensuring everyone has equal access to all benefits from genomics
  - Access should be the same regardless: population, location, socioeconomics
- International Collaboration
  - Sharing knowledge and resources between countries and institutions
  - Collaborations and other efforts must promote genomic technologies and scientific findings/advances for all



# Federated Learning

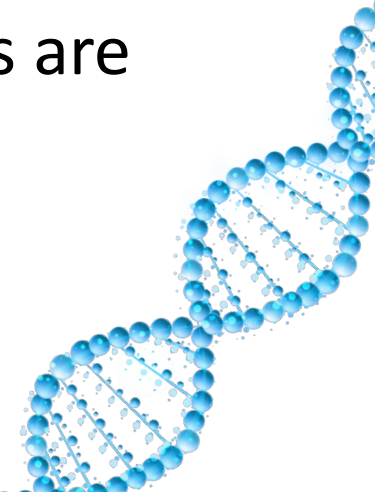
- Data is distributed by nodes (countries, universities, hospitals)
- Algorithms are trained in a decentralized way
- Data does not leave the node
- Privacy-preserving analytics





# Ethical and Legal Challenges

- Data privacy
  - Protects genetic, clinical and other information
  - Avoids discrimination
  - Ensures individual trust
- Informed consent
  - Ensures understanding of risks and benefits prior to consent
  - Empowering the individual
- Intellectual Property
  - Intellectual property
  - Clear definition of rights and discoveries Encourage innovation
  - Ensure that benefits are widely distributed



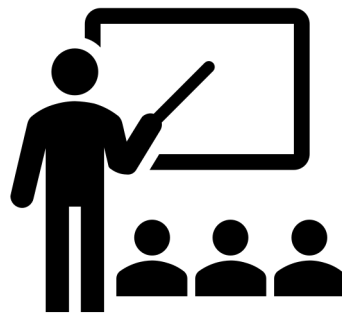
# Education and Training

- Professional Training

- Data literacy
- Genomics professionals
- Clinical bioinformaticians
- Genetic Counselors

- Continued Education

- Updating knowledge and skills
- Technologies are fast-evolving and so must be (the knowledge of) the professionals





# Thank you