THE EVOLUTION OF GENOMICS IN THE LAST DECADE IN ROMANIA

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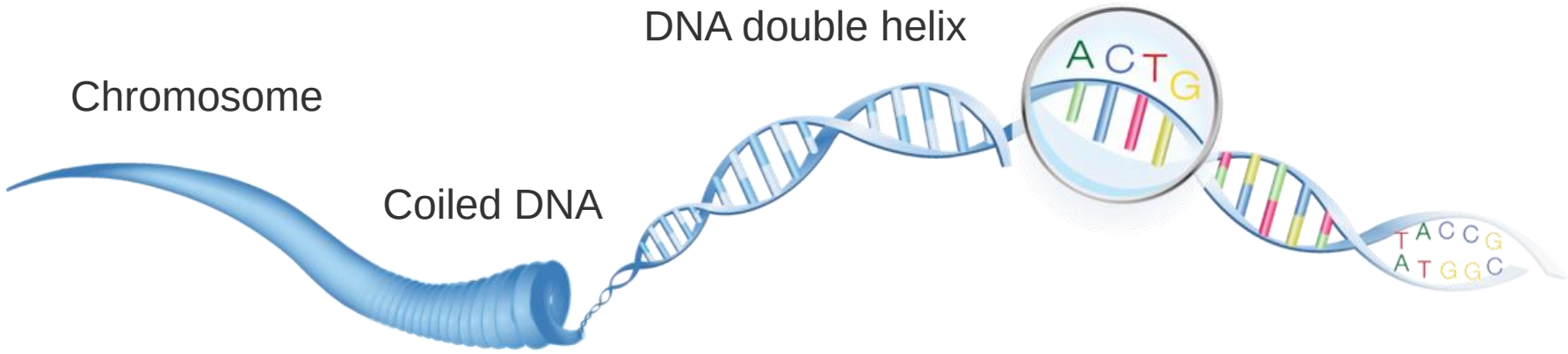
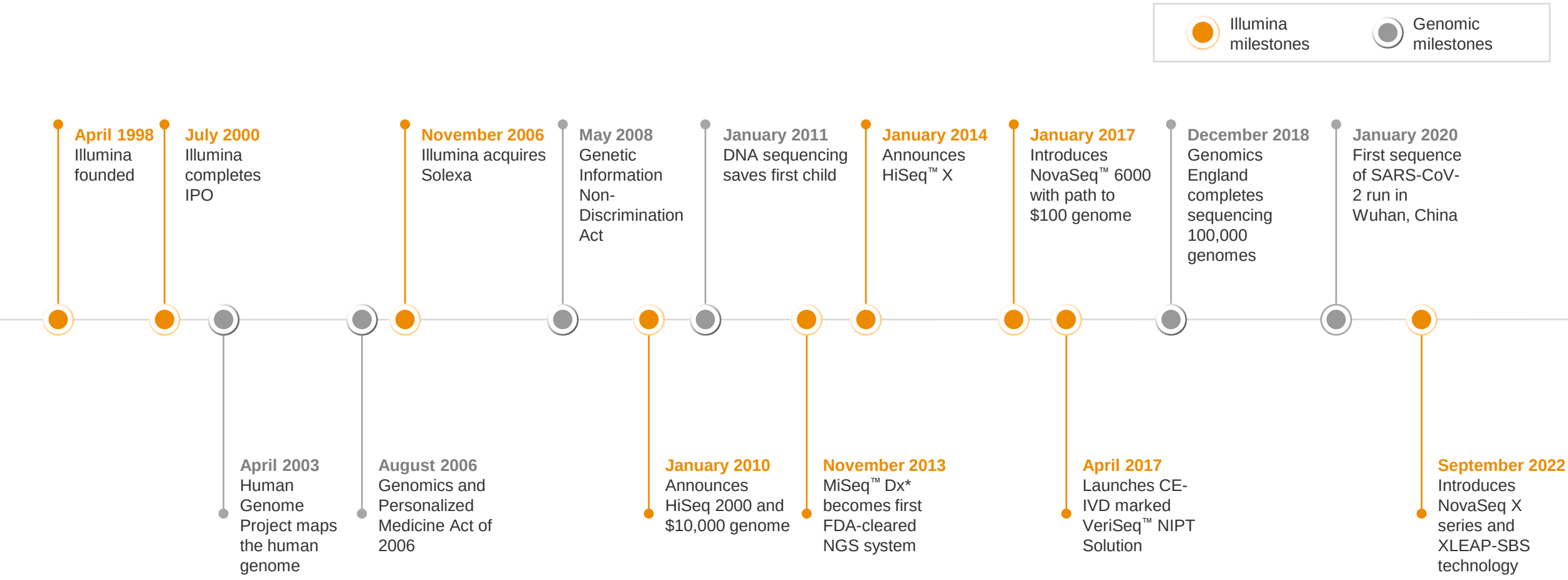
Understanding genomics



The four bases

Adenine	Cytosine	Thymine	Guanine

Illumina milestones in the era of genomic discovery



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Accelerating access to genomics

We are driven by our conviction that genomics should be available to the many, not the few.

We believe we have an obligation to make our technology affordable and accessible to as many patients as possible, while setting the highest standard for the ethical use and security of data.

Cost of sequencing, per human whole genome

Since 2001, with innovation the cost of DNA sequencing has dropped more than 100,000×, from $100 million USD per human genome to $200 USD in 2023.

1. Wetterstrand KA. DNA Sequencing Costs: Data from the NHGRI Genome Sequencing Program (GSP). Available at: www.genome.gov/sequencingcosts; 2. NovaSeq™ X series. Data on file.

The latest in population-scale sequencing

Enabled by Illumina technologies' speed and throughput

US: All of Us Research Program

All of Us RESEARCH PROGRAM

Goal: Speed health research discoveries, enabling new kinds of individualized health care

1 million participants; aiming for 70% of samples from underrepresented groups

China: One Million Whole Genome Sequencing Project

Six cohorts in four years

Three genetic data bases specific to the Chinese population

UK: Our Future Health

The UK's largest-ever health research program

DNA samples from over 5 million adults

Singapore: National Precision Medicine

PRECISE
Precision Health Research Singapore

Pilot: 2018 – 10,000 genomes

Phase 2: 2022 – 100,000 genomes

Phase 3: 2025 – 1,000,000 genomes

NGS is emerging as standard of care for genetic disease in the United States

NICUSeq randomized study

Results demonstrate that clinical whole-genome sequencing (cWGS) outperforms usual care by two-fold both in terms of a diagnostic efficacy and change of clinical management.[1]

Clinical guidelines

The ACMG[2] recommends WES/WGS for infants under one year of age with congenital anomalies or patients under 18 years of age with an intellectual disability/developmental delay

228 million

Covered lives for WES and/or WGS

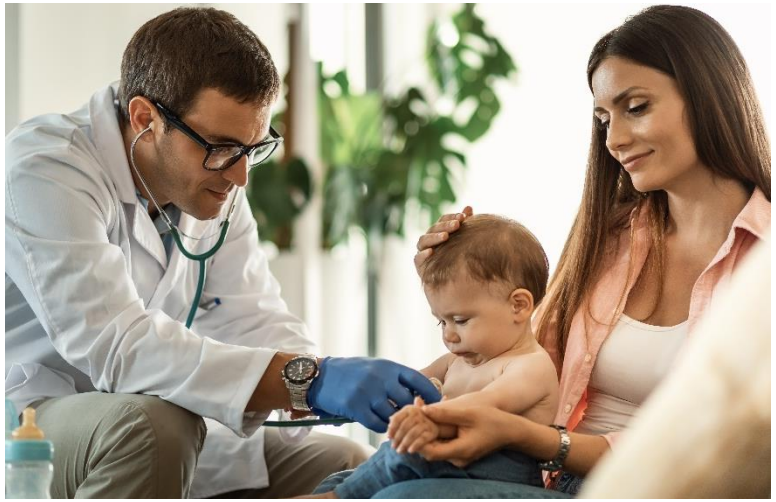
1. NICUSeq Study Group, Krantz ID, Medne L, et al. Effect of Whole-Genome Sequencing on the Clinical Management of Acutely Ill Infants With Suspected Genetic Disease: A Randomized Clinical Trial. JAMA Pediatr. 2021;175(12):1218-1226. doi:10.1001/jamapediatrics.2021.3496 2. American College of Medical Genetics and Genomics

NGS is becoming the standard of care for genetic disease diagnosis around the globe

474 million

Covered lives for WES and/or WGS globally

Payers across the globe continue to open coverage for WES/WGS in various geographies. These include the US, Canadian, Australian, and major Western European markets.



Multiple studies demonstrate cost effectiveness

WGS and WES provide improved clinical and economic value for patients and health care systems.[1-4]

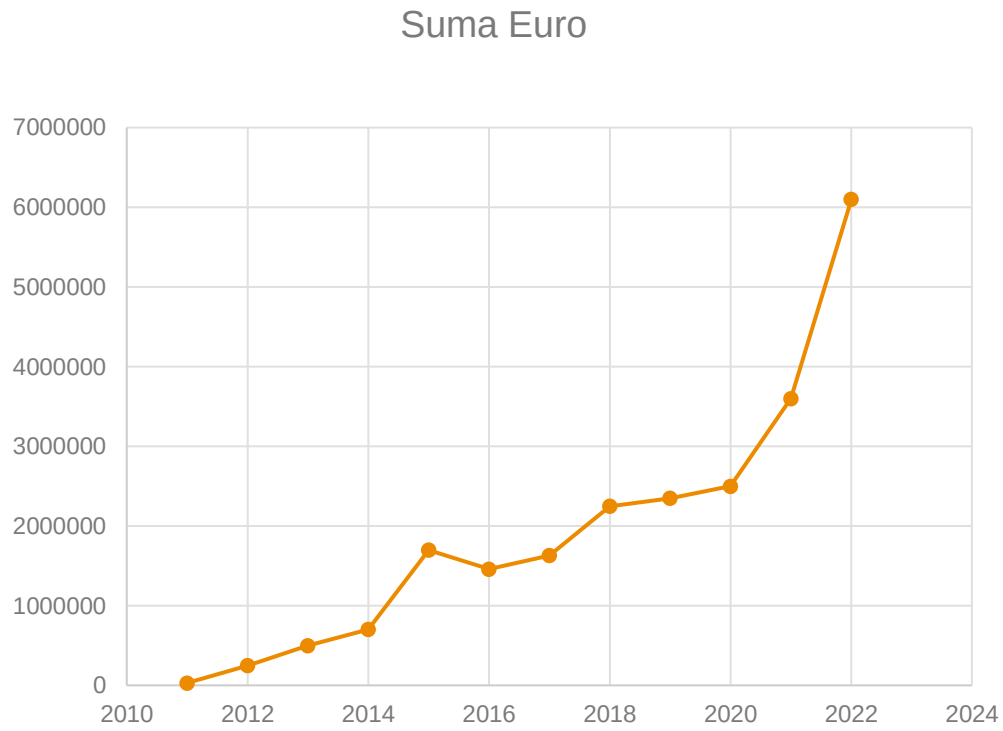
Supporting nationwide programs to assess the impact of WGS

BeSolveRD,[5] Project Baby Bambi,[6] and other projects continue to build evidence to establish the utility of WGS in routine care.

1. Incerti D, et al. Genet Med. 2022 Jan;24(1):109-118. doi: 10.1016/j.gim.2021.08.015. Epub 2021 Nov 30. PMID: 34906478. 2. Li C, et al. Genet Med. 2021 Mar;23(3):451-460. doi: 10.1038/s41436-020-01012-w. Epub 2020 Oct 28. PMID: 33110268. 3. Goranitis I, et al. Genet Med. 2022 Feb 15:S1098-3600(22)00029-6. doi: 10.1016/j.gim.2022.01.013. Epub ahead of print. PMID: 35181209. 4. Yeung A, et al.. Genet Med. 2020 Dec;22(12):1986-1993. doi: 10.1038/s41436-020-0929-8. Epub 2020 Aug 10. 5. Illumina Supports Nationwide Program across Belgium to Assess Whole Genome Sequencing for Developmental Disorders Diagnosis 6. Illumina Supports Israel's National Program to Accelerate Diagnoses of Critically-Ill Newborns with Suspected Genetic Disease

Romania-Past, Present, Future

1. First NGS sequencer in 2012-MiSeq Bucharest
2. First big capacity system in 2015- HiSeq2500 Timisoara
3. First NIPT lab in 2021-NextSeq550Dx Craiova
4. First NovaSeq6000 2021- Bucharest



Growth factors

- Faster increase of number of samples
- National plan for Oncology
- Rare Desease National Program
- Non Invasive Prenatal Test NIPT
- Big number of platforms- around 100 systems for NGS

Slowing factors

- Big number of platforms
- Not enough samples to use the maximum capacity
- No founds

Thank You

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