

**Day 2**

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# **Next Generation Sequencing**

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29 January 2010

# ACKNOWLEDGEMENTS

Early enthusiasm of speakers

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Massive attendance

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Hakim and Jo



## Technology maturity

accuracy >99%, 25 to 1000 bp, paired end, high and ultra high throughput  
kits for genomics, transcriptomics, epigenomics

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## Software puberty

Maq, Bowtie, Velvet

## Analysis infancy

heterozygotes 30x, CNVs, PTCs/Missense/Nonsense, epigenetics profiles, small RNAs, etc

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## Hardware specialization according to needs

length of reads, length of runs, throughput, accuracy

## Technology

library reliability, amplification or recapture bias  
throughput too high or still too small

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## Software

CPU time

## Analysis

no automation, manual validation, still in the discovery phase, CPU power, data transfer

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## High Costs

hardware, reagents, CPU, data storage, analysis time

## Technology

Expansion of hardware portfolio (very low to ultra high), expansion of protocols (eg, RNA-Seq)

Complete Genomics (small groups, healthy costs competition), Helicos (more niche effect)

Recapture maturity (exomes, exomes, exomes)

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## Software

Best CPU time, Cloud or External Computing

## Analysis

Pathogenicity automation (Exomes), Cancer Genomes, New genomes (eg, pathogens, new models, evol)

Integration of different layers (SNPs, CNVs, Epigenetics, Transcriptomics, Proteomics, Metabolomics)

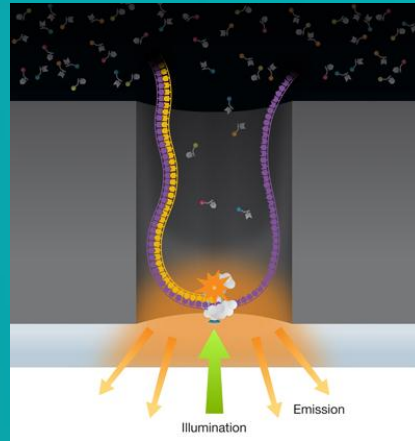
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## Lower Costs

More adapted hardware/reagents costs whatever the (small or big) needs

## Technology

Polymerase sequencing?  
Nanopore sequencing?  
Other method?



## Software

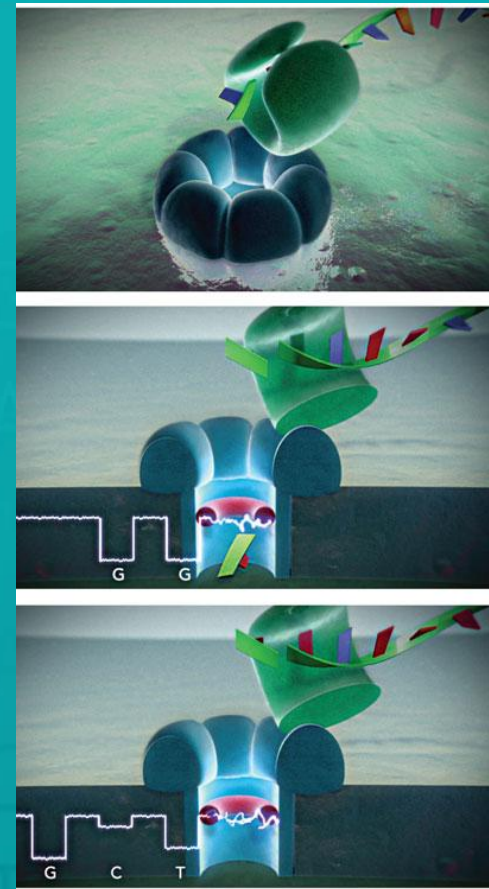
External computing is the norm? Desktop supercomputers?

## Analysis

Full automation of the analysis?  
Full integration of different layers? Automated Systems Biology?

## Lower Costs

\$1000 genome or X prize (100 genomes, 10 days, \$10,000 per G, 1 error per 100,000, 98% coverage)?  
Personal Genomics and Preventive Medicine Era? (Governments-MDs preparedness? Health Care Insurance?)



# Outline of the Day

4 Sessions	AM	Technology then Biology
	Lunch	Posters
	PM	Recapture then WGS Medical (+ Awards)

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Speakers: timer = 2 min left, chairman stands up = go to conclusion

Public: Award for interaction (each question a ticket, draw at the end)

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Mobiles on silent mode

Thanks again to all of you and enjoy the day !





