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# Current state of DNA sequencing technologies and the consequential tasks in computational life sciences

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- What is DNA?
- Polymerase Chain Reaction (PCR)
- Sanger Sequencing
- Next-Generation Sequencing (NGS)
- DNA Sequence Alignment
- Nanopore Sequencing
- Oxford Nanopore Technologies MinION Sequencer
- Selective Sequencing
- Project Group Goals
- Acknowledgements

### What is DNA?



M.-T. Hütt and M. Dehnert, Methoden der Bioinformatik. Berlin, Heidelberg: Springer Berlin Heidelberg, 2016, Chapter 1.

# What is DNA?



M.-T. Hütt and M. Dehnert, Methoden der Bioinformatik. Berlin, Heidelberg: Springer Berlin Heidelberg, 2016, Chapter 1.

## Chain Reaction (PCR)



Linda K. Park, Paul Moran, and Robin S. Waples, Eds., "Generalized schematic of polymerase enzyme activity," in Application of DNA Technology to the Management of Pacific Salmon, 1994 [Online]. Available: http://www.nwfsc.noaa.gov/publications/scipubs/techmemos/tm17/figures/moranfig4.htm. [Accessed: 07-Mar-2015]

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# Sanger Sequencing



Linda K. Park, Paul Moran, and Robin S. Waples, Eds., "Sanger dideoxy chain termination sequencing," in Application of DNA Technology to the Management of Pacific Salmon, 1994 [Online]. Available: http://www.nwfsc.noaa.gov/publications/scipubs/techmemos/tm17/figures/moranfig4.htm. [Accessed: 07-Mar-2015]

# Next-Generation Sequencing (NGS)



J. M. Johnsen, D. A. Nickerson, and A. P. Reiner, "Massively parallel sequencing: the new frontier of hematologic genomics," Blood, vol. 122, no. 19, pp. 3268–3275, Nov. 2013 [Online]. Available: http://www.bloodjournal.org/content/122/19/3268. [Accessed: 17-Mar-2015]

## **DNA Sequence Alignment**

Data Analysis

### Variant identification

#### Reference:



Illumina Sequencing Technology. 2013 [Online].

Available: https://www.youtube.com/watch?v=womKfikWlxM&feature=youtube\_gdata\_player. [Accessed: 07-Mar-2015]

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## Nanopore Sequencing



V. Greenwood, "How It Works: The Fastest DNA Sequencer," Popular Science. [Online]. Available: http://www.popsci.com/science/article/2013-03/fastest-dna-sequencer. [Accessed: 19-Sep-2016]

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## Oxford Nanopore Technologies MinION Sequencer



Image credit: Oxford Nanopore Technologies

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# **Selective Sequencing**

### Problem

- DNA samples may be contaminated
  - Different species
- Only small parts of the whole genome of interest
  - Single genes are examined

### Possible solution

- Locate the current read within the whole genome
- Assign the curent read to a specific species
  - Continue sequencing when positive result
  - Otherwise eject the strand from the pore

### Requirements

- Real-Time alignment of the reads to one or multiple references
  - Assignement to a gene or species

- Enable Selective Sequencing on the Human Genome
  - Over 3 billion base pairs
  - Optimize or find algorithms to enable it
- Work towards a fully mobile solution
  - Enable DNA analysis in the field
  - Benchtop device for laboratories

- Dr. Franz-Josef Müller
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- Benedicte Willert and Prof. Dr. Ole Ammerpol Institute for Human Genetics at the University Hospital Schleswig Holstein Campus Kiel

Thank you for your patience.