

NGS - variant analysis

Geert van Geest
Interfaculty Bioinformatics Unit, UniBe
Training group, SIB

Teachers

- **Diana Marek:** Training manager at SIB
- **Irene Keller:** bioinformatician at DBMR Bern
- **Geert van Geest:** trainer at SIB/bioinformatician at IBU Bern

Learning outcomes

- Understand important **aspects** of **NGS** and read **alignment** for variant analysis
- **Perform** a read **alignment** ready for variant analysis
- **Perform variant calling** according to **GATK** best practices
- **Perform** a variant **annotation**

Learning experiences

- Lectures
- Quiz questions
- Exercises

Communication

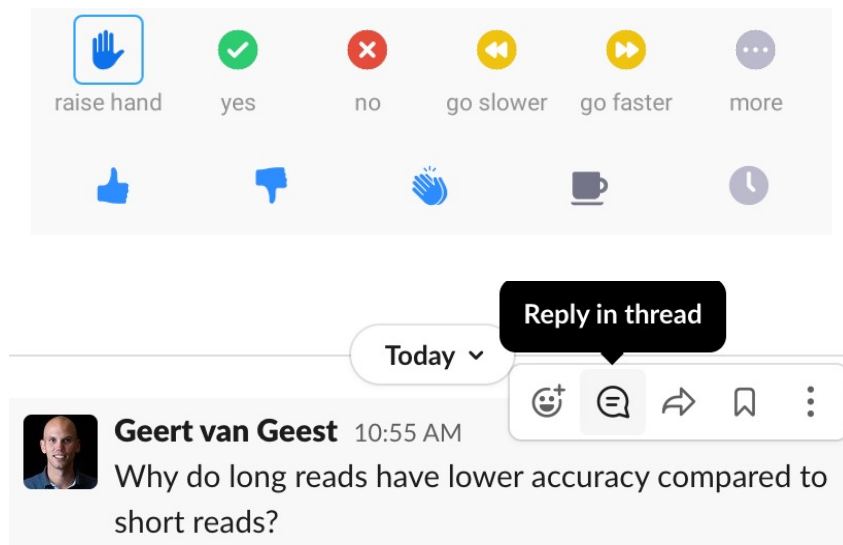
- Course website:

<https://sib-swiss.github.io/NGS-variants-training/>

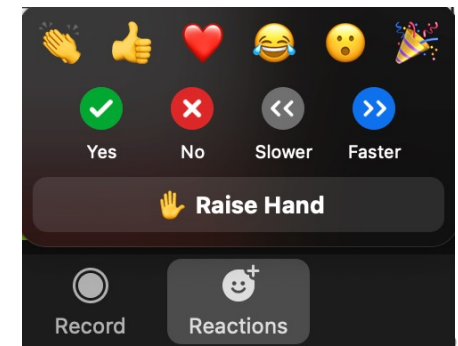
- Slack
- Google docs

Asking questions

- During lectures: zoom functionality
- Personal interest questions: [#background](#)
- During exercises: [#q-and-a](#) on slack



OR



Learning outcomes – per chapter



Long-read sequencing analysis



Introduction



Learning outcomes

After having completed this chapter you will be able to:

- Illustrate the difference between short-read and long-read sequencing
- Explain which type of invention led to development of long-read sequencing
- Describe the basic techniques behind Oxford Nanopore sequencing and PacBio sequencing
- Choose based on the characteristics of the discussed sequencing platforms which one is most suited for different situations

Get to know each other

- Write in the google doc (5 minutes):
 - Three keywords about yourself
 - Why you are joining this course, and what you want to learn
- You will discuss them in breakout rooms afterwards (15 minutes)
 - Introduce yourself based on what you've written in the doc