Bioinformatics for Precision Medicine

Introduction to the course

Carolina Barra Quaglia

Associate Professor at DTU Bioinformatics
Protein Immunoinformatics
carolet@dtu.dk

Research Interests

#Immunoinformatics
#machine-learning #deep-learning
#antigen processing and presentation
#protein-clustering #proteomics
#allergy
#autoimmunity

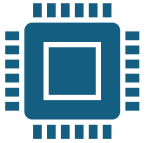
Welcome to Bioinformatics for MPM

Bioinformatics tools in the context of precision medicine, focus on the **clinical practice** and **therapeutics**

Learning the tools in a **hands-on** manner supported by a light **theoretical foundation** to understand key concepts

The online part of the course will explore **additional essential bioinformatics tools** broadly applicable across many domains

Practical course, apply the tools on a **case study project** work during the last 4 weeks of the course



No prior coding experience is required. We will use state-of-the-art **web-based** tools

Structure of the course

Block 1: Bioinformatics for genomic medicine - variant calling and classification

Block 2: Immunoinformatics for precision therapeutics – Epitope prediction and Immunotherapy

Block 3: Fundamental bioinformatics algorithms: BLAST and MSA

Block 4: Project work

Course Plan

Block 1: Bioinformatics for genomic medicine - variant calling and classification

Day 1: DNA sequencing and germline variant calling

The role of bioinformatics in genomic medicine and the workflow from DNA sequencing to variant identification. Exercise on germline variant calling and classification, including key file formats and commonly used databases. Interpret variant data and apply relevant tools in a clinical context

Day 2: Variant Interpretation and Classification Methods

How genomic data can be interpreted and used for classification and molecular subtyping. Fundamental concepts of clustering and classification algorithms and how these methods are applied to biological data. Through hands-on exercises, students will be able to apply and evaluate these methods in a practical setting.

Course Plan

Block 2: Immunoinformatics for precision therapeutics – Epitope prediction and Immunotherapy

Day 3: Assessing chimeric antigen receptor targets for cancer immunotherapy

How bioinformatics tools are used to evaluate targets for chimeric antigen receptor (CAR) therapy. hands-on experience with relevant databases and web servers to analyze protein features, expression, and suitability as therapeutic targets. Integrate multiple tools to assess targets in a biologically meaningful context

Day 4: Neo epitope prediction and vaccine development (on-line)

understand the principles of T cell epitope prediction and the role of MHC binding in immune recognition. How computational methods, including neural networks, are used to predict peptide–MHC interactions. Apply epitope prediction tools and interpret results in the context of immunotherapy

Course Plan

Block 3: Fundamental bioinformatics algorithms: BLAST and MSA

Day 5: Pairwise sequence alignment and BLAST (on-line)

understand the principles of pairwise sequence alignment and how sequence similarity is assessed. They will learn how BLAST is used to identify homologous sequences and interpret key output metrics such as e-values and percent identity. Through exercises, students will be able to apply these tools to analyze biological sequences.

Day 6: Multiple Sequence alignment and Phylogenetic trees (on-line)

understand how multiple sequence alignment is used to compare sequences and identify conserved regions. They will learn how phylogenetic trees are constructed and how to interpret evolutionary relationships between sequences. Through hands-on exercises, students will be able to build and analyze phylogenetic trees.

Course Plan

Block 4: Project work

The project forms the basis of the exam and is carried out in groups of 3–5 students. It is open-ended but must include methods from the course. Each group selects at least one gene of interest and analyzes it in a diagnostic or therapeutic context using at least five web services or databases

The report

The report should be 10–12 pages and include a summary, introduction, materials and methods, results and discussion, and conclusion. It should demonstrate understanding of the tools used, how they work, and where the data comes from, as well as critically evaluate results and limitations

The presentation

The group gives a ~20-minute presentation covering the project, methods, and key findings. All group members should contribute and demonstrate understanding

The exam

The group presentation is followed by ~20 minutes of oral examination with questions to individual group members. Other participants may attend, but only examiners ask questions

Exam

Generelle regler for eksamensregistrering og afmelding
Registrering til et kursus medfører automatisk registrering til den ordinære eksamen.
Deadline for afmelding af eksamen er 14 dage før eksamen.
Deadline for tilmelding til re-eksamen er 14 dage før reeksamen.
Eksamens til- og afmelding sker til lifelonglearning@adm.ku.dk

Bioinformatics
Kursus kode: SPMM21006U

Ordinary exam	Submission of written assignment in Digital Eksamen: 12:00 noon, Monday, May 25, 2026. Oral exam: Friday, May 29, 2026
Grades, ordinary exam	Same date as the oral exam.
Re-exam	Submission of written assignment in Digital Eksamen: 12:00 noon, Monday, June 15, 2026. Oral Exam: Friday, June 19, 2026
Grades, re-exam	Same date as the oral re-examination.
Exam venue	Technical University of Denmark – DTU
Exam form	Written assignment based on a group project followed by an oral examination.
External examiner	No
Grading scale	Passed/not passed
Exam responsible	Carolina Barra Quaglia carolet@dtu.dk

Agenda for today

Day 1: DNA sequencing and germline variant calling

When: April 20, 10.00-17.00

Where: Building 202, room R1020 how to get there?

Program:

10.00 - 10.15: Intro to the course (Carolina Barra Quaglia)

10.15 - 11.00: Bioinformatics in Genomic Medicine (Frederik Otzen Bagger)

11.00 - 11.15: Coffee Break

11.15 - 12.00: Somatic variant calling and RNA sequencing (Frederik Otzen Bagger)

12.00 - 13.00: Lunch

13.00 - 13.30: Introduction to germline variant characterization (Carolina Barra Quaglia)

13.30 - 17.00: Exercises in germline variant classification (Carolina Barra Quaglia)

15.00 - 15.15: Coffee Break

Agenda for today

Day 2: Variant Interpretation and Classification Methods

When: April 21, 9.00-17.00

Where: Building 202, room R1014

Program:

9.00 - 9.30: Exercise summary

9.30 - 10.00: Clinical variant interpretation (Carolina Barra Quaglia)

10.00 - 10.15: Coffee Break

10.15 - 11.15: Clustering + Classification algorithms (Carolina Barra Quaglia)

11.15 - 12.00: Microbial genomics in personalized medicine (Rasmus Lykke Marvig)

12.00 - 13.00: Lunch

13.00 - 17.00: Exercises in variant interpretation, clustering and classification (Carolina Barra Quaglia)

15.00 - 15.15: Coffee Break

Agenda for today

Day 3: Assessing chimeric antigen receptor targets for cancer immunotherapy

When: April 22, 9.00-17.00

Where: Building 202, room R1014

Program:

9.00 - 9.30: Exercise summary

9.30 - 10.00: Immunoinformatics and CAR T cells

10.00 - 10.15: Coffee Break

10.15 - 11.15: Bioinformatics tools for assessing CAR targets (Lars Rønn Olsen)

11.15 - 12.00: Exercises in CAR target assessment (Carolina Barra Quaglia)

12.00 - 13.00: Lunch

13.00 - 17.00: Exercises in CAR target assessment (Carolina Barra Quaglia)

15.00 - 15.15: Coffee Break

Agenda for today

Day 4: Neo epitope prediction and vaccine development (on-line)

When: April 25, 9.00-15.00

Where: Zoom link

Program:

9.00 - 9.30: Exercise summary

9.30 - 10.00: MHC binding and T cell epitopes (Carolina Barra Quaglia)

10.00 - 10.15: Coffee Break

10.15 - 11.00: Technical note: Building a sequence logo (Carolina Barra Quaglia)

11.15 - 12.00: MHC binding predictions and medical applications (Carolina Barra Quaglia)

12.00 - 13.00: Lunch

13.00 - 15.00: Exercises T cell Epitopes prediction (Carolina Barra Quaglia)

Links

Detailed Course Program