

# BIO392

# Bioinformatics of Sequence Variations

Genome Variation | Function | Data Formats | Resources | Privacy



1992



2001



2003



2006



2007



Heidelberg

Stanford

Gainesville

Aachen

Zürich

Student of medicine | doctoral thesis in molecular cytogenetics @ DKFZ (Peter Licherter) | resident in clinical hematology/oncology | data, clinical studies & cancer systematics

Post-doc in hemato-pathology (Michael Cleary) | molecular mechanisms of leukemogenesis | transgenic models | expression arrays | systematic cancer genome data collection | *Progenetix* website

Assistant professor in paediatric haematology | molecular mechanisms of leukemogenesis | focus on bioinformatics for cancer genome data analysis

Research group leader in genetics | genomic array analysis for germline alterations | descriptive analysis of copy number aberration patterns in cancer entities

Professor of bioinformatics @ IMLS (2015) | systematic assembly of oncogenomic data | databases and software tools | patterns in cancer genomes | *arraymap* online resource | GA4GH | SPHN

# BIO392: Course Schedule

	Tue Sep 15	Wed Sep 16	Thu Sep 17	Fri Sep 18	Tue Sep 22	Wed Sep 23	Thu Sep 24	Fri Sep 25	Tue Sep 29	Wed Sep 30	Thu Oct 1	Fri Oct 2	Tue Oct 6	Wed Oct 7
<b>09:00 - 10:00</b>		Michael: short introductory lecture about genome variation	Izaskun: Terminal / Unix / Files	Michael: genomic file formats		Izaskun: File typers / manipulations	Bo: Introduction to UCSC genome browser	Genome versions & liftover (continued)		Intro online platform Rahel?	Intro to R ...	Genome variants & populations		Exam
<b>10:00 - 11:00</b>		Github exercise: create user specific directories & upload/edit test files using Markdown	Izaskun: Terminal / Unix / Files	ToDo: File formats, calculations		Izaskun: File typers / manipulations	Bo: Introduction to UCSC genome browser	A taste of BLAST and scripting		GATK online course	DNAcopy ... exercise	plink		Exam
<b>11:00 - 12:00</b>			Izaskun: SIB online introduction to Unix			Izaskun: File typers / manipulations	Bo: Introduction to UCSC genome browser	A taste of BLAST and scripting		GATK online course	DNAcopy ... exercise			Exam
<b>13:00 - 14:00</b>	* Room information * Administrative - discuss times/days - exam	Literature (genome variant review papers ...)	Izaskun: SIB online introduction to Unix	ToDo: File formats, calculations	Recap W1; Q&A	Izaskun: file typers/manipulations reading/literature	genome browser related exercises	Python, R ...	Michael: genomic variants w/ respect to schedule etc.	GATK online course	Progenetix as tool for CNV frequencies etc.		Recap; Q&A; genomic privacy (?)	
<b>14:00 - 15:00</b>	Tina Siegenthaler: technical introduction (room,					Literature (genome analysis techniques ...)		read up/look up other genome resources, writing about use cases	Q&A, (Exercises???)			CNV literature etc.	Populations / Ethnicities /	
<b>15:00 - 16:30</b>	* explore course site * create Github accounts and forward to bio392@compbiozurich.org	Exercise: Q&A using Github				Genome technologies - brief notes about usage scenarios, pro & con								



# BIO392: Course Resources



- Course repository & website on Github
    - links to articles and information resources
    - downloads

<https://compbiozurich.org/UZH-BIO392/>

<https://github.com/compbiozurich/UZH-BIO392/>

## UZH BIO392

Bioinformatics of Sequence Variation

[Course Info](#)

[Course Days](#)

[Teachers](#)

[Examples, Guides & FAQ](#)

[Related Sites](#)

[CompbioZurich](#)

[UZH390 lectures](#)

[Baudisgroup at UZH](#)

[Github Projects](#)

[compbiozurich](#)

[progenetix](#)

[Tags](#)

[FAQ](#)

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## UZH BIO392 - Bioinformatics of Sequence Variation

This is a repository for materials related to the BIO392 *Bioinformatics of Sequence Variation* introductory course at the University of Zürich.

### Summary

One of the fastest growing areas of bioinformatics is in the analysis, warehousing and representation of genomic and protein sequence variants, particularly with view on the use of molecular data in personalised health and biomedical applications in general. This course will engage participants to explore common data formats, online resources and analysis techniques, with a focus on human genome variation data.

### Learning Goals

- Core [Learning Goals](#), relevant for passing the test...

### Links

- BIO392 HS 2019 in the [UZH OLAT system](#)
- BIO392 HS 2019 in the [UZH directory](#)

### Literature and Resources

- [Literature links](#) and recommendations
- [Resource links](#) (browsers and online repositories)

### Schedule

### Course feedback pages

### Location

- Room info

The image shows a 3D architectural floor plan of the Irchel and Stockwerk F buildings. The Irchel building is on the left, featuring several rooms numbered 1 through 14. A green arrow points from room 1 towards the Stockwerk F building. The Stockwerk F building is on the right, with rooms labeled Y11 through Y55. A legend at the bottom provides details for specific rooms:

- 1 Zugang zu Y-01F-50 Computerarbeitsplätze Zwischengeschoss
- 8 Hörsaal Y22-F-68
- 10 Seminarraum/Sitzungszimmer Y35-F-08A

A small inset map in the bottom right corner shows the location of the buildings relative to other campus areas.

University of  
Zurich<sup>UZH</sup>

[Display a menu](#)

Terminal?

UNiX?

Github?

R?

Python?

Perl?

YAML?

Skill assessment