

# VarFish Training

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# Course Outline

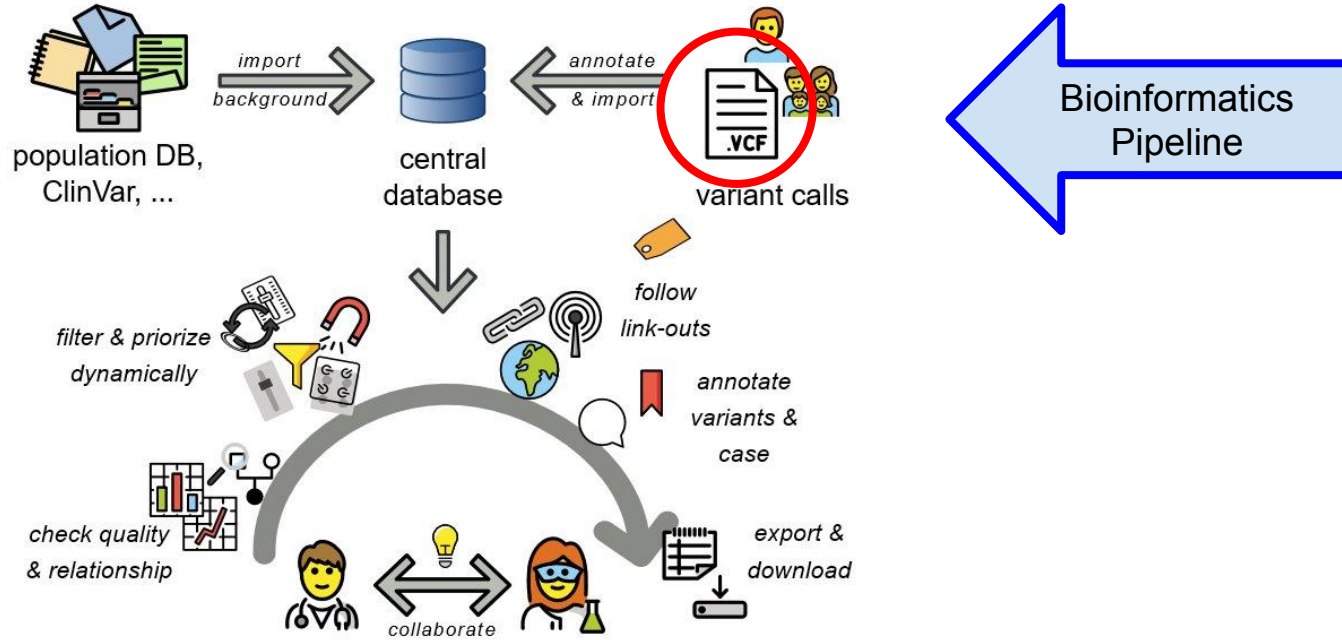
1.	VarFish Introduction	15 min	09:00	
2.	VarFish Walk-Through	40 min	09:15-09:55	5 min BREAK
3.	Hands-On VarFish	90 min	10:00-11:30	5 min BREAK
4.	Discussion / Reflection	25 min	11:35-12:00	

# Course Audience

We assume that

- have good experience with genetics
- have basic experience with NGS data
- little or no experience with VarFish

# 1.1 Overview (Introduction to VarFish)



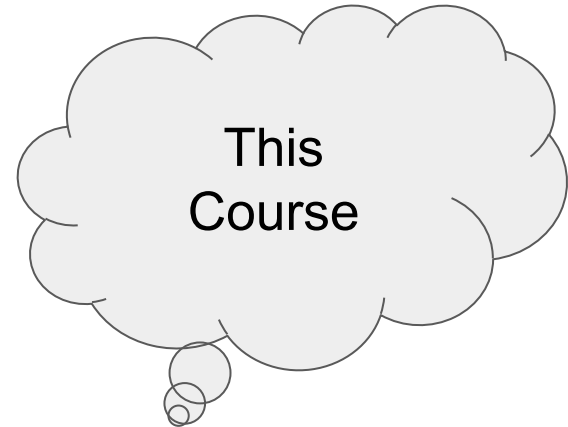
# 1.1 Analysis Workflow (VarFish Introduction)

## Before VarFish (Bioinformatics Staff)

- NGS Processing, Variant calling
- Variant Import

## With VarFish

1. Quality Control
2. Variant Filtration
  - a. Assess Variants
  - b. Create User Annotations (flags and comments)



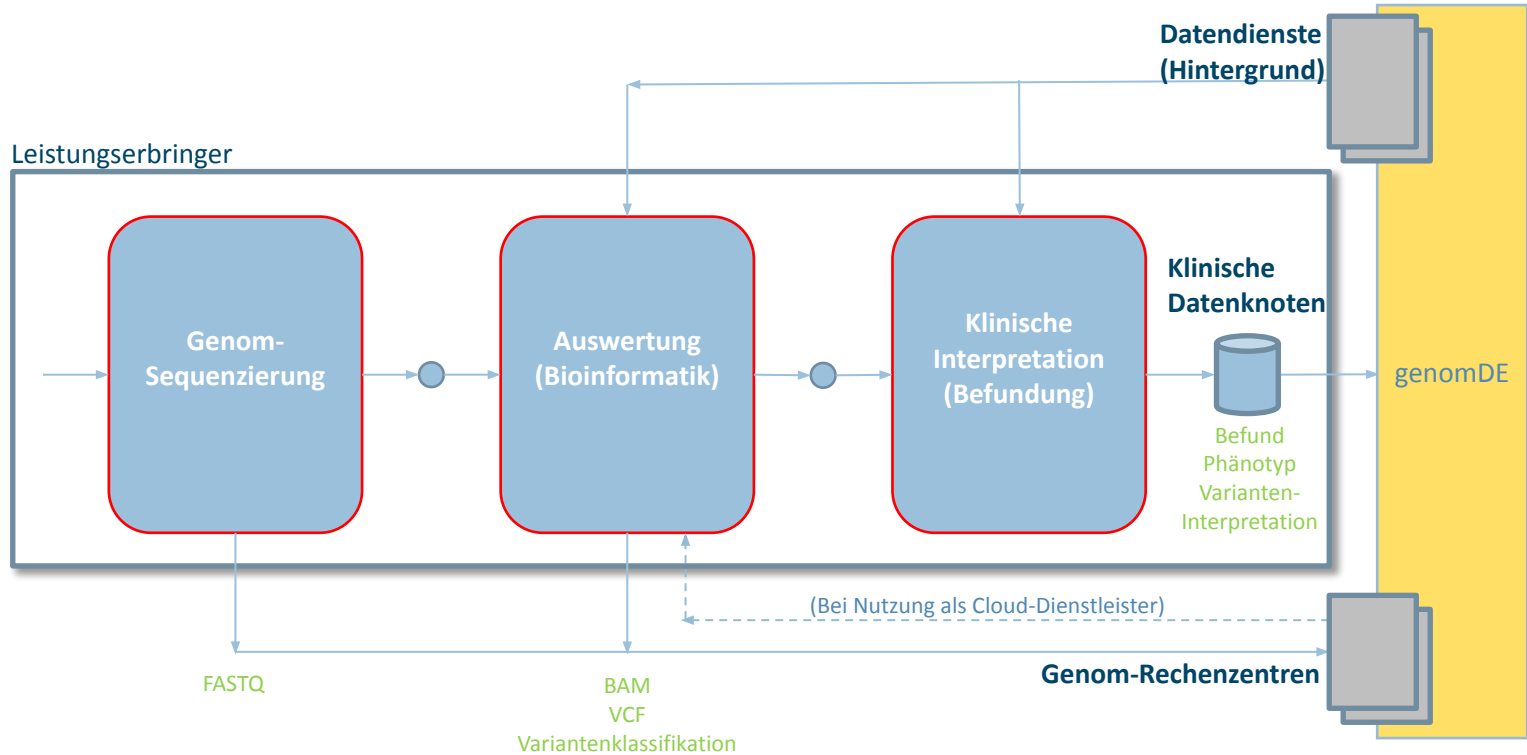
## 1.2 VarFish in Context of genomDE

Klärung von UV3 und match making (Kernleistung):

“[...] Bereitstellung eines Services welcher die Analyse von Genotype-Phenotype Kombinationen über Standorte hinweg erlaubt (“Wurde Variante X im Zusammenhang mit Phenotyp Y schon mal beobachtet”) insbesondere um bisher unbekannte krankheitsverursachende Varianten zu identifizieren. Es muss möglich sein den entsprechenden Kontakt zum anderen Standort herstellen (Match-Making, mit der behandelnden Ärzt:in oder Klinik) um die weitere gemeinsame Fallaufklärung zu ermöglichen. Dieser Service ist damit zentrales Element für die standortübergreifende wissensgenerierende Krankenversorgung. Anders als in A werden hier Daten mit hohem Schutzbedarf zwischen Standorten abgeglichen. [...]”

=> VarFish bietet diese Funktionalität über Beacon queries

## Ausschnitt aus dem Versorgungsprozess



## 1.3 Getting More Information

### **VarFish User Manual**

<https://varfish-server.readthedocs.io/en/latest/>

### **VarFish GitHub Project (Download)**

<https://github.com/bihealth/varfish-server>

### **VarFish Users Mailing List**

<https://mailman.charite.de/mailman/listinfo/varfish-users>

### **VarFish Operators Mailing List**

<https://mailman.charite.de/mailman/listinfo/varfish-operators>



# VarFish Walk-Through

Demo Time!



# Hands-On VarFish

- Log into VarFish
  - Before the course we have sent you a user name (your email address) and a password to log into at <https://varfish-ext.cubi.bihealth.org/>
- Consult your hand-out for examples to work on
  - Go to Section 3.1 and, e.g., start with Case 2 (or 1 if you have not done so yet)
- We will split you into 6 smaller break-out groups Zoom
  - Work through the cases and try to handle questions first with your group
  - In case of issues: send us a chat Zoom message (or email ;-))
  - We will also move through the break-out Zoom rooms and answer any questions
- We will ask some questions afterwards ;-)
  - What did you like the most/least in VarFish?
  - What was most remarkable to you?
  - What was the greatest challenge that you found?

# VarFish & the IVDR

## IVDR includes regulation of software in lab process

- Situation 2022/09: no notified body exist yet, no IVDR accredited software for variant analysis yet

## Plan for VarFish

- History as “for research use only software”
- Aim 2022: Software Development with QMS and 13485, 62304, 62366  
=> IVDR Class A self-certification
- Aim 2023-2024: Lift this up to IVDR Class C  
=> Actual certification and registration as IVD device in EU

# Discussion / Reflection

1. What did you like the most/least in VarFish?
2. What was most remarkable to you?
3. What was the greatest challenge that you found?

Thank you for your interest!  
**Questions?**

## Contact

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