

# INTERPRETATION OF SEQUENCE VARIANTS

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# INTERVAR

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- ▶ SQLite db
  - ▶ tabeller: pasientinfo, varianter, tolkning
- ▶ Python backend
  - ▶ Kjøre spørringer, lage rapporter, entre data i db
- ▶ Python + (django, flask etc. ) frontend
  - ▶ webgui hvor bruker kan legge inn data, få oversikt printe rapporter etc.

## SCHEMA FOR DB

### ▶ Pasienter

- ▶ pasientID
- ▶ Fødselsår
- ▶ Kjønn
- ▶ Kliniske opplysninger
- ▶ Slektsnummer
- ▶ Panel (hvilken analyse er utført (APN, HSP, Exom, Filtex))

### ▶ Varianter (Som VCF-format?)

- ▶ Chr
- ▶ Start
- ▶ Stop

### ▶ Ref

- ▶ Alt
- ▶ rsID

### ▶ Tolkninger

- ▶ JSON fra Alamut
- ▶ Tolket av / Dato
- ▶ Klasse

### ▶ Kjøringer

- ▶ Mean target cov
- ▶ % Target > 20 & 30 X
- ▶ SBS



## Login screen

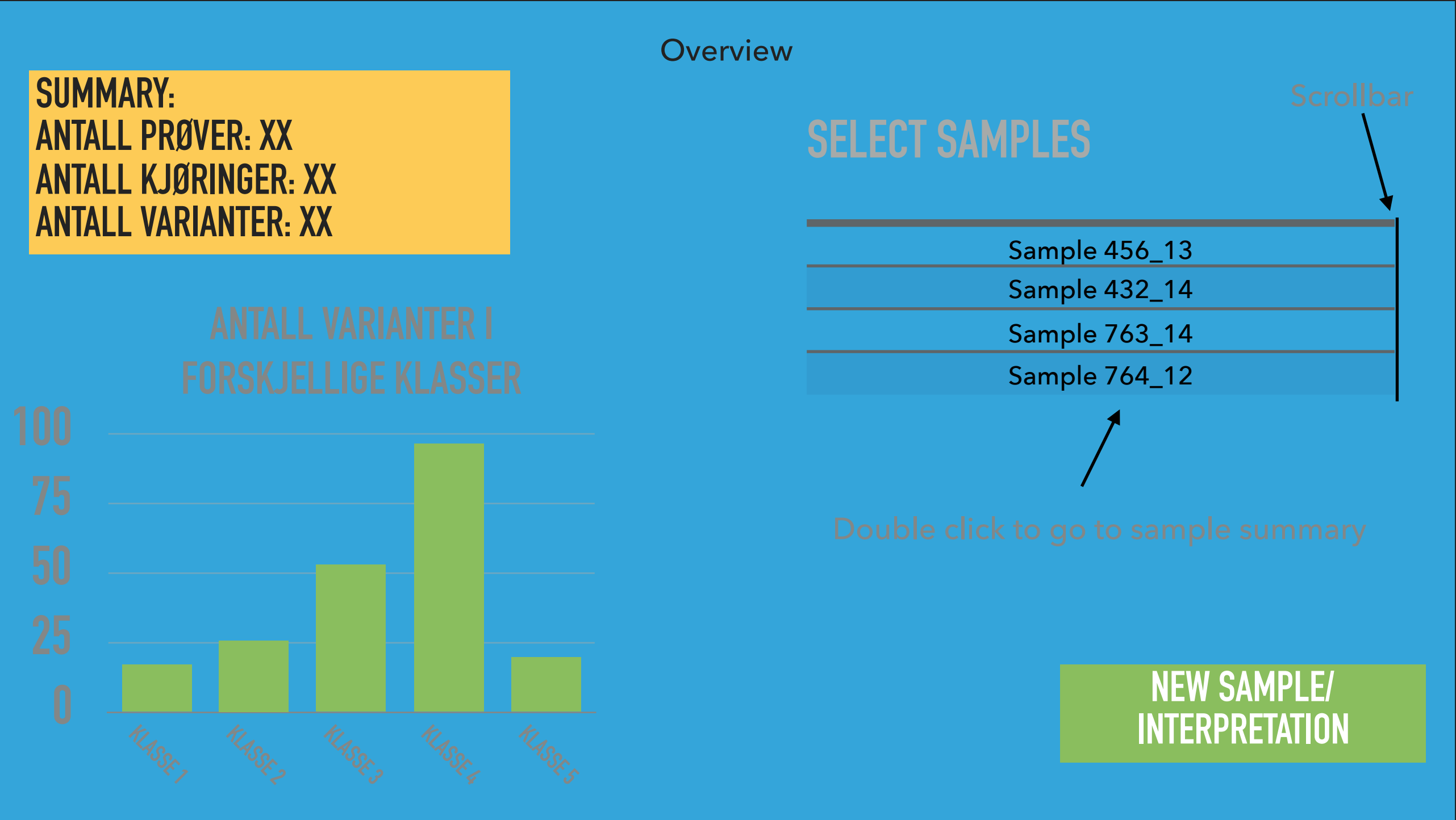
InterVar login

Username

Password

**DONE**

Overview screen



► Patient screen

Patient screen

**PATIENT INFO:**

PATIENT ID

BIRTHDATE

FAMILY ID

CLINICAL INFO

PANEL

UPDATE

VARIANTS CONNECTED TO THIS SAMPLE:

Chr	Start	Stop	Mer info
1	123	1234	blabla
2	123	123	blabla
X	321	231	blabla
2	231	312	blabla

Scrollbar

Double click to go to variant summary

ADD VARIANT

REPORT

Variant screen (Popup from patient screen button)

Variant screen

VARIANT INFO FOR PATIENT :

CHR:

START:

STOP:

REF:

ALT:

TRANSCRIPT:

CDNA:

TYPE:

PROTEIN:

DOMAIN:

SPLICING:

ZYGOZITY:

EXAC:

1000KG:

ESP:

HGMD:

REFS (PMID):

COMMENT

CLASS:

SEEN BEFORE:

PREVIOUS CLASS

GET INFO FROM ALAMUT

UPDATE

DONE