Swiss Variant Interpretation Platform

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THE CHALLENGE

- Cancer is a **rare** disease
- Cancer is most **common**
- **Not** enough data
- **Too much** data
- There is **no** standard
- There are **many** standards
TIME OUT, DANIEL! WHAT ARE YOU TALKING ABOUT?

Skin cancer

Ion Torrent

KIT W557R

MycancerGenome.org

sunitinib: confers increased sensitivity

sunitinib

ClinVar

COSMIC

Varsome

CIViC

OncoKB

pathogenic

pathogenic

Uncertain Significance

Sensitivity/Response

Likely oncogenic
THE WISHLIST

- **Joint repository**: Shared between all partner hospitals with single point of entry.
- **Posting of variants**: To avoid unnecessary manual labor and increase efficiency.
- **Validation of clinical variants**: By manual curation (SIB) and clinical experts consensus (hospitals).
- **Common annotation**: Approved by all partners, ensuring consistent assessment of pathogenicity.
- **Discrepancy management**: To solve conflicting annotations between labs when new variants are identified.
- **Feedback mechanism**: In case pathogenicity changes over time.

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SWISS VARIANT INTERPRETATION PLATFORM FOR ONCOLOGY

THE IDEA

variants

relevance
THE EXAMPLES
SWISS VARIANT INTERPRETATION PLATFORM FOR ONCOLOGY

THE PROJECT

call
initial ramp up
harmonise
applications
testing

Clinical-grade variant database
Retrieval system
Validation process
Submission system

call
initial ramp up
harmonise
applications
testing

extend
reconcile
curate

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THE PARTNERS

SVIP

SOCIBP  SPON

other

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All participating institutions
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