# Whole genome sequencing based identification of tumor tissue of origin





Abstract #: EACR21v-0293

### Introduction

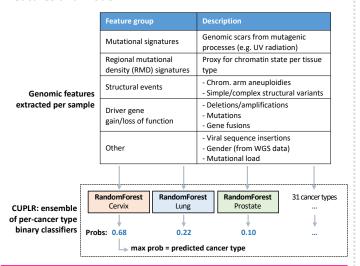
- Primary tumor tissue of origin (TOO) guides treatment decisions
- ... typically determined by histopathology
- ... cannot be determined for 3-5% of metastatic tumors
- A whole genome sequencing (WGS) based machine learning classifier can aid pathologists with clarifying TOO

## CUPLR (Cancer of Unknown Primary Location Resolver)

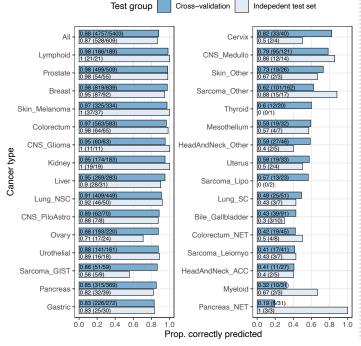
Samples and training



#### Features and model

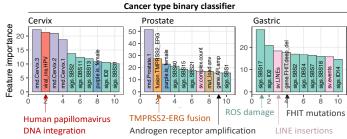


## CUPLR predicts TOO with 87% overall accuracy



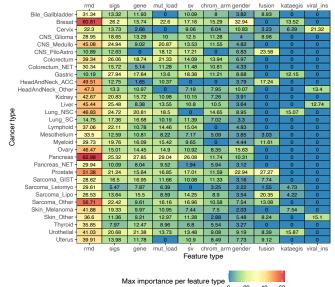
## Features predictive of TOO

Certain features used by CUPLR are known to be associated with the respective cancer types



Overall, the most predictive feature types are:

- Chromatin state (RMD signatures)
- Mutational scars from mutagenic processes (mutational signatures)



# Patient report

For a patient, the top predicted cancer types are shown. In cases with no clear top prediction, info on the features supporting a prediction still allows a pathologist to make an informed decision on the TOO of a patient

HMF002928A (actual cancer type: Unknown)

