# EXPLORING GENOTYPE-PHENOTYPE RELATIONSHIPS OF BREAST CANCER

OBJECTIVE

- To predict the genomic biomarkers - hormone receptor status, subtype, and mutations, from breast cancer histopathology images
- Explore the results in terms of human interpretable features
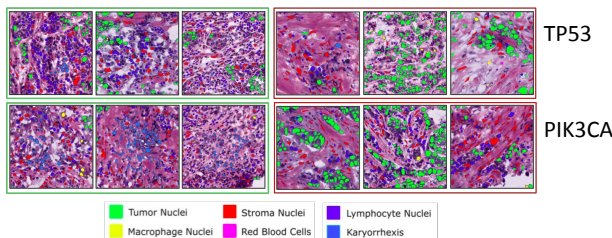
METHOD

- Used a proxy task (Cancer vs Normal Classification), to extract the cancerous regions from the image for biomarker classification
- Inception Net v3 pretrained on imageNet as the classifier
- The *discriminative patches* were further analysed on their intensity, spatial, and morphological features using a random forest classifier
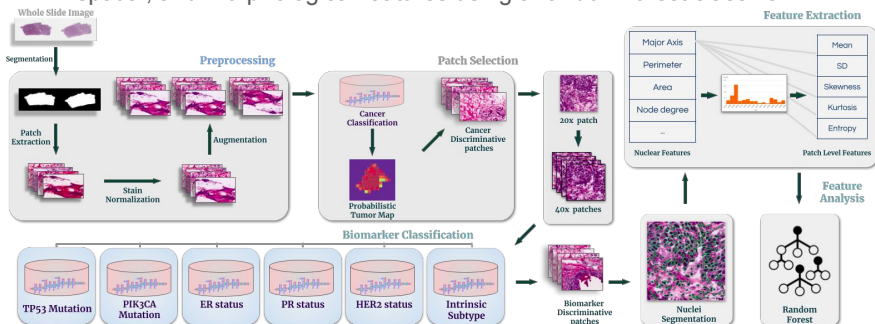
RESULTS

Prevalence of lymphocytic nuclei in TP53 mutated patches and presence of Karyorrhexis in PIK3CA mutated patches [green box].

Tumor Nuclei | Stroma Nuclei | Lymphocyte Nuclei
Macrophage Nuclei | Red Blood Cells | Karyorrhexis

Classification Results of Genomic Biomarkers

AUROC	Level	TP53	PIK3CA	ER	PR	HER2	Subtype
Ours	patch	0.829	0.721	0.866	0.820	0.798	0.877
	slide	**0.875**	**0.765**	**0.910**	**0.839**	**0.811**	**0.909**
Baseline	patch	0.677	0.565	0.665	0.614	0.666	0.703
	slide	0.643	0.541	0.632	0.578	0.622	0.685
Best	slide	0.75†	0.63†	0.89‡	0.81‡	0.79‡	0.826§

CONCLUSION

- DL applied for, predicting biomarkers not currently in pathologist's workflow
- Using discriminative patches for biomarker classification improved performance
- TP53 mutation showed correlation with lymphocytes and PIK3CA mutation showed correlation with karyorrhexis

REFERENCE

Ruchi Chauhan, PK Vinod and CV Jawahar. "Exploring Genetic-histologic Relationships in Breast Cancer" 2021 IEEE 18th International Symposium on Biomedical Imaging (ISBI). IEEE 2021.

Authors: Ruchi Chauhan, CV Jawahar, PK Vinod   Research Center Name: CVIT, CCNSB