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Radiogenomics: Combining SNP Studies with Pancreatic Ductal Adenocarcinoma Medical Images

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Abstract

Radiogenomics is a fusion of two methods, radiomics, and genomics. Radiomics is the process in which features are extracted from medical images such as MRIs, CT scans, and PET scans. Genomics on the other hand is studying an organism's genome and, in this study, their respective gene mutations. By combining both methods, radiogenomics can help find biomarkers for various cancers to help diagnose and treat patients more efficiently. In this study, Radiogenomics was used to find an association between genomic profiles and imaging features of patients with pancreatic ductal adenocarcinoma (PDAC). Out of a total of 117 patients available, only 29 patients were selected for the study. The study required that the patients must have a complete genetic profile available, a preoperative CT scan, PDAC, and are participating in the Rapid Autopsy Program (RAP).

Methods

- DNA was sequenced from 29 RAP patients and a SNP study was performed
- The entire Pancreatic cancer mass was contoured using Velocity AI software
- 3D Slicer software was used to extract all features with a voxel size 2³, loG kernel size 3³, and wavelet-based features
- A statistical correlation test is then ran between the SNP study and extracted features

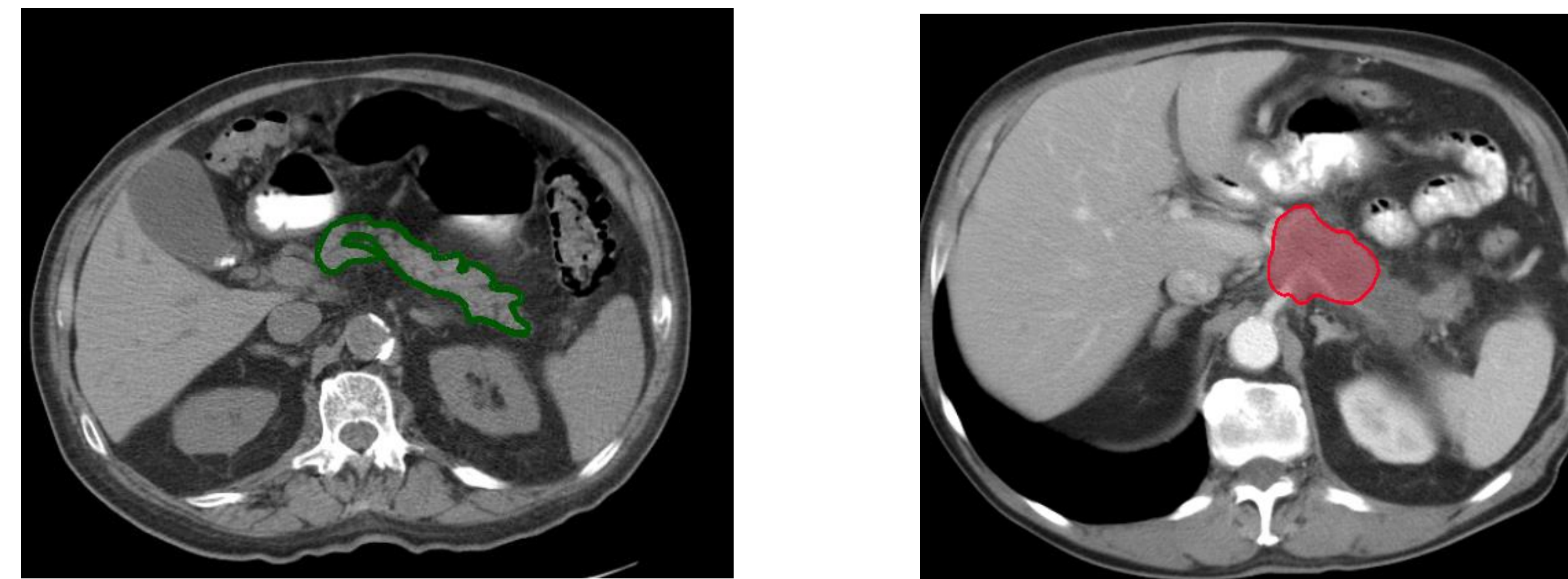


Figure 2. Health Pancreas contour (green) on the left and PDAC contour (Red) on the right

Results Pt 2

- Each RAP patient has a total of 982 features extracted with the glm parameter having the most features at 25%.
- Radiomics features are textural information that can be analyzed using advanced mathematics and AI.
- Wavelet and LoG are not features by themselves, but instead are filters used to obtain features. LoG filters use an “edge detection filter” to help enhance edges for AI’s. Wavelets are similar but detect intensity changes in the three directions (XYZ).

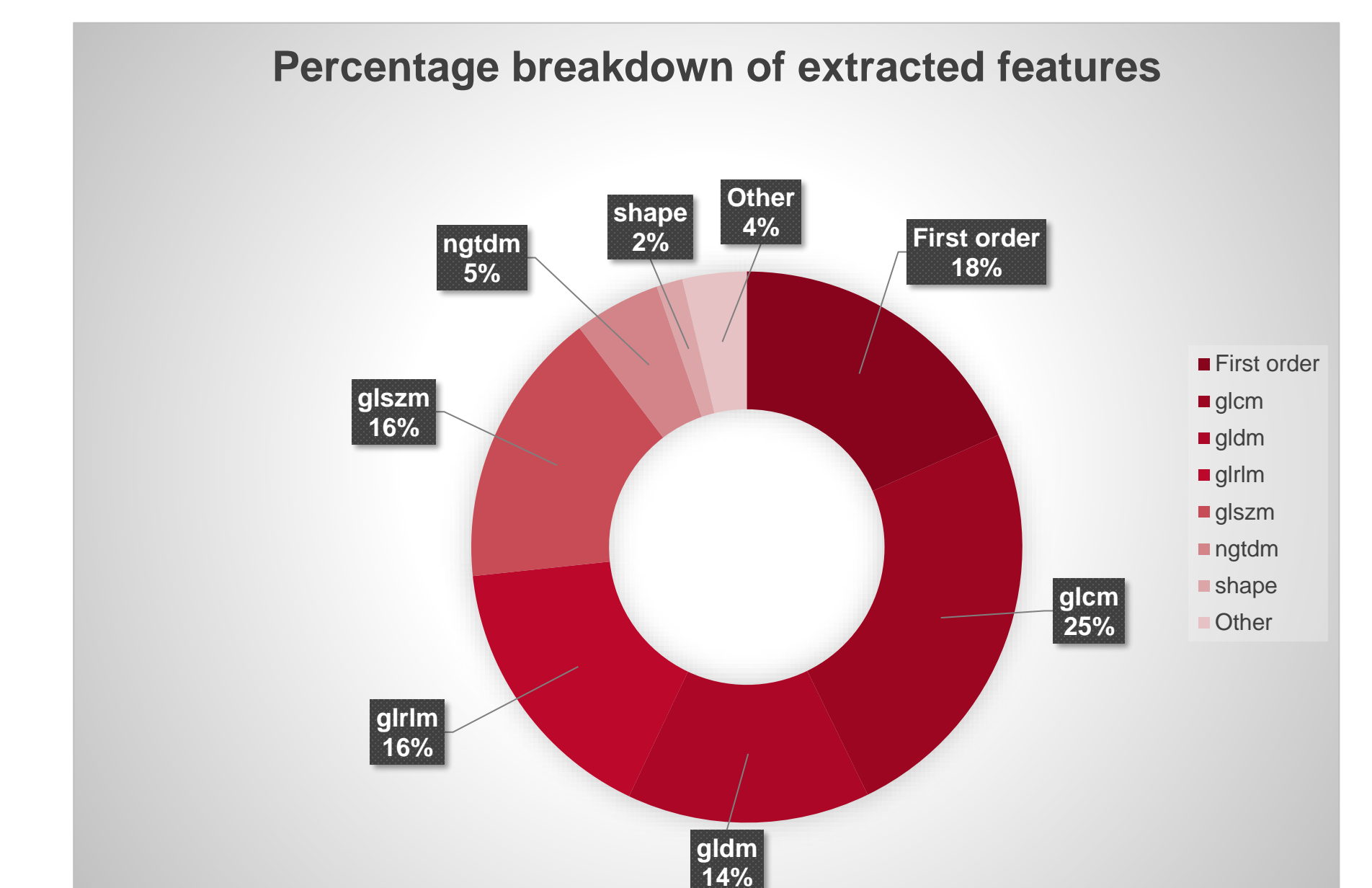


Figure 5. Pie chart of all features extracted in percentages.

Introduction

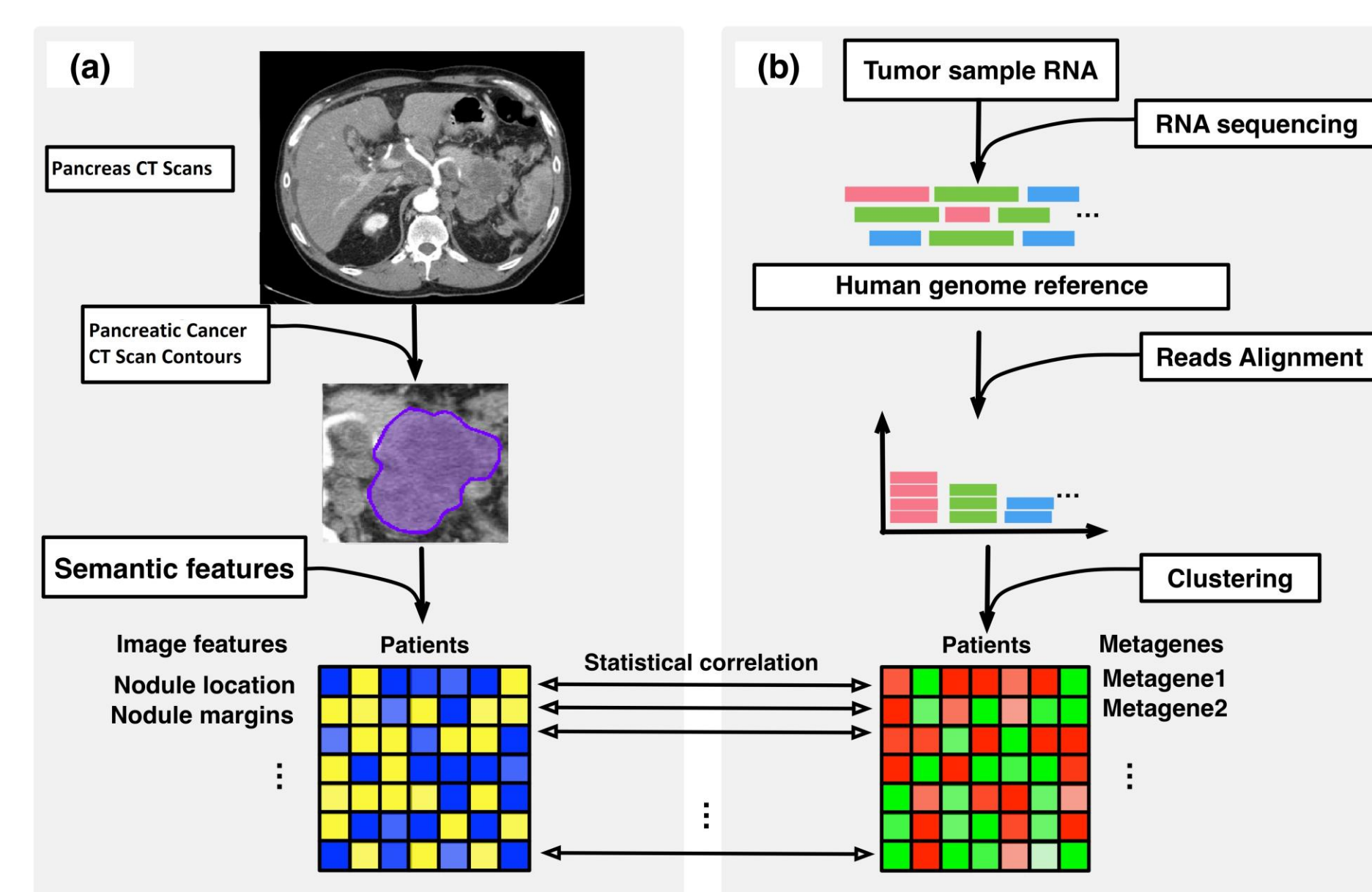


Figure 1. Radiogenomics workflow shows both processes, with radiomics listed as “(a)” and genomics as “(b)” respectively.¹

Pancreatic ductal adenocarcinoma :

- PDAC is a very lethal cancer, considering the survival rate of 5 years is just over 10%.
- Most patients are found between the ages of 55 - 84
- Other cancer types such as breast, prostate, and skin cancer have reduced lethality rates in the last 2 decades whereas PDAC has remained stagnant
- The lethality of this cancer is relatively high due to late diagnosis, resulting in metastasized tumors unable to be removed surgically.

Results

- Of the 29 RAP patients sequenced, there was a total of 14530 different single base-pair mutations. Looking at figure 1, Cytosine→Guanine was the most common base substitution with 2878 mutations, respectively. On the other hand, Thymine→Guanine appeared the least with a total of 415 mutations.
- Looking at figure 2, there are 26 different mutation type categories. The most common mutation type were intron variants with a total of 3328 appearances. The least common mutation types were stop_gained, start_lost, and protein_Protein_contact, appearing only three times

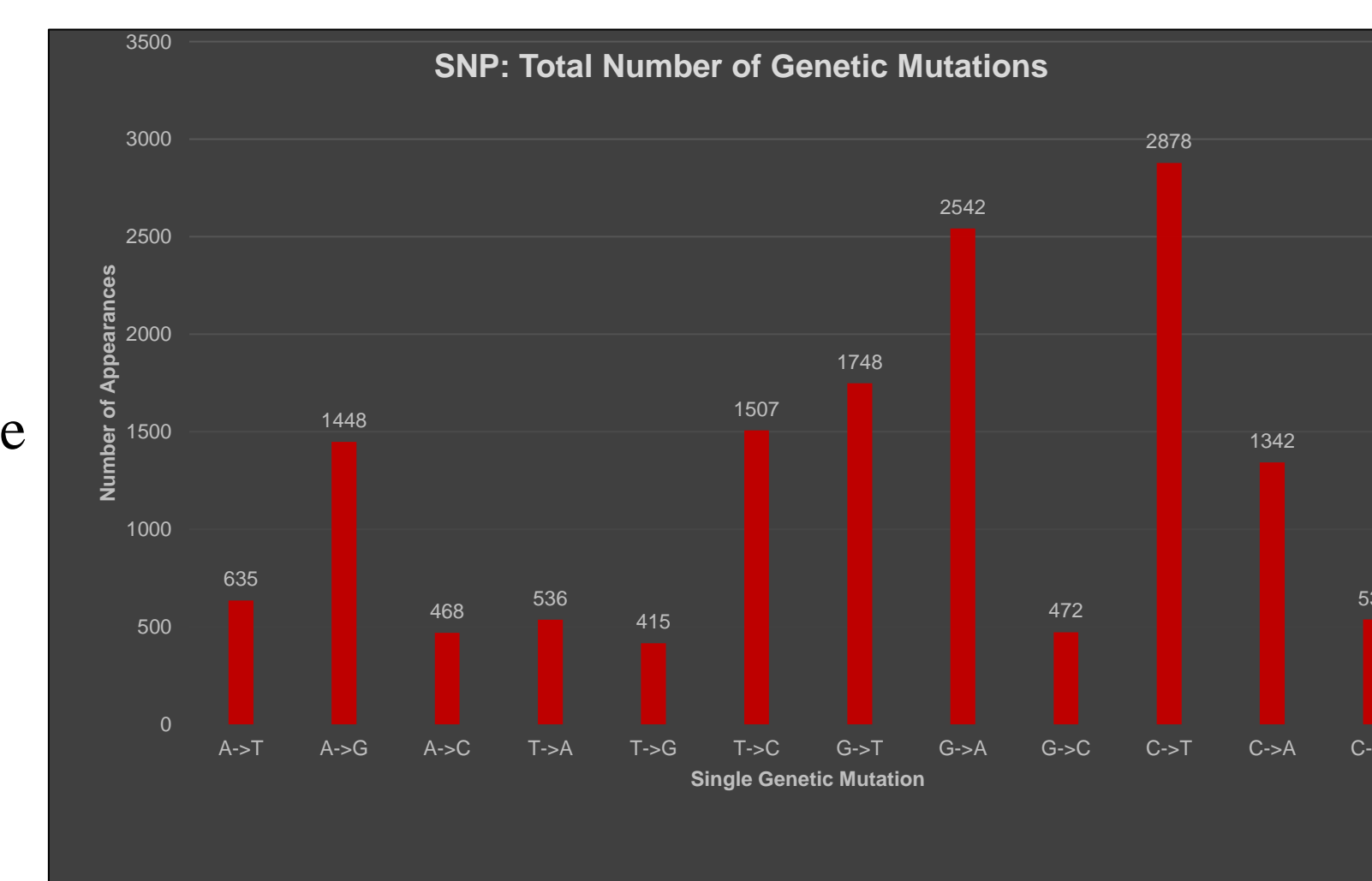


Figure 3. Total genetic mutations across all patients

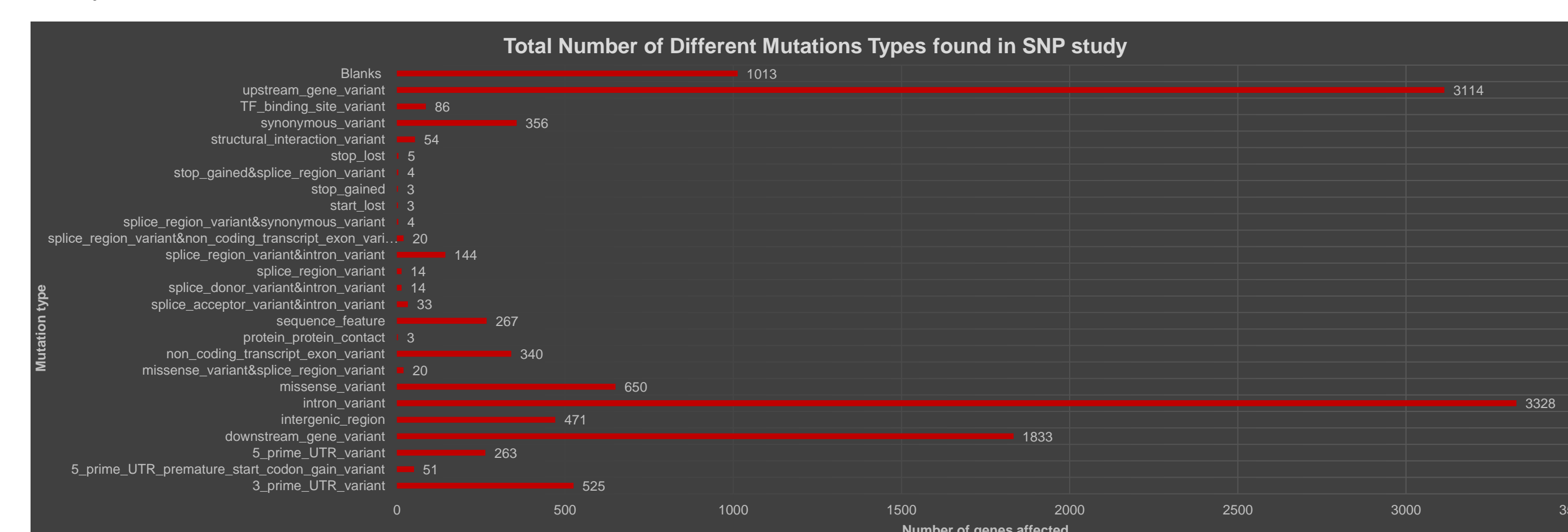


Figure 4. Total number of different SNP mutation types across all patients

Future Work

Currently the study is still being worked on. A statistical analysis needs to be completed between the radiomic and SNP study. If a high correlation is found between certain mutations and PDAC, this would positively impact the life expectancy of patients with PDAC. Physicians will be able to better diagnose PDAC sooner and allow those affected more personalized medicine options.

References

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