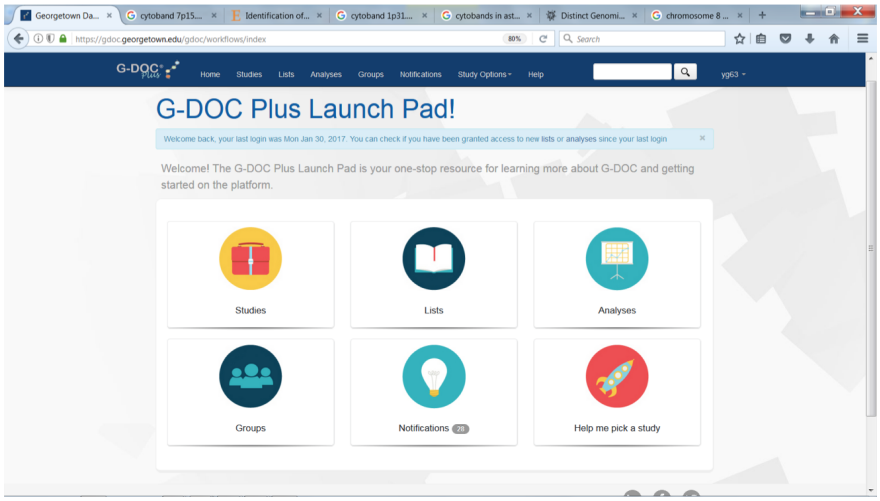
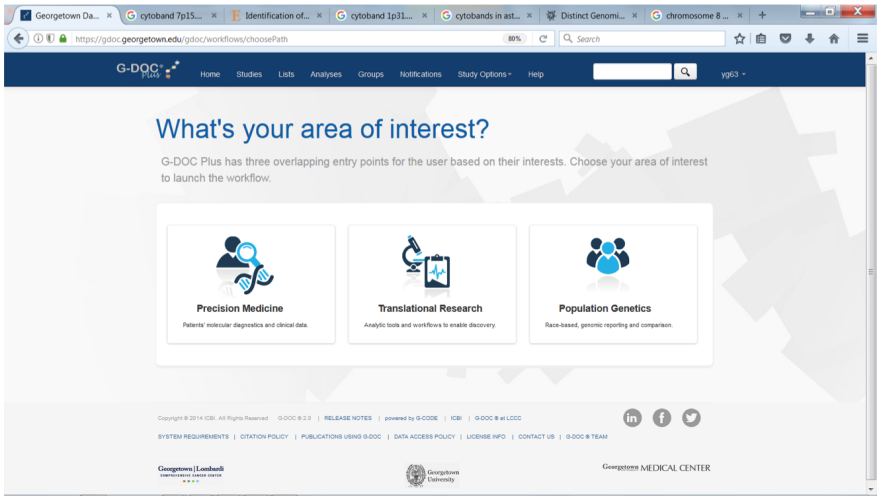
Genome Sequencing Exercise

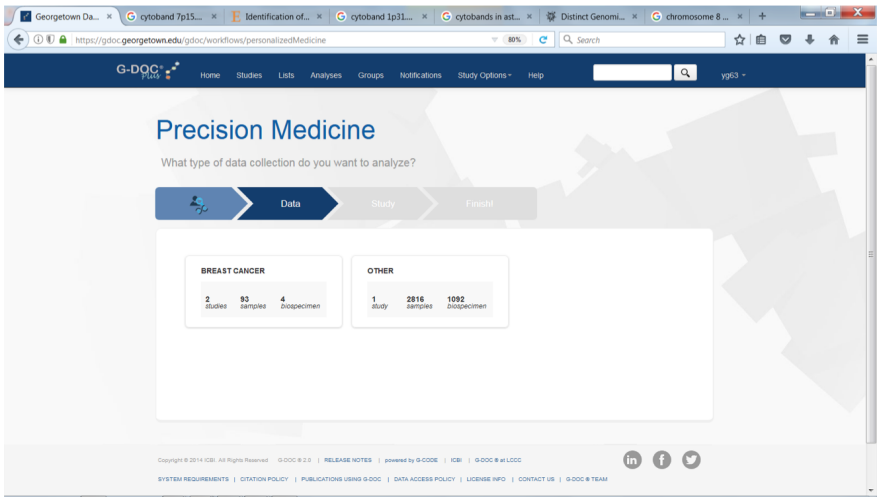
1. Log in to G-DOC (https://gdoc.georgetown.edu) and select the module "Help me pick a study."



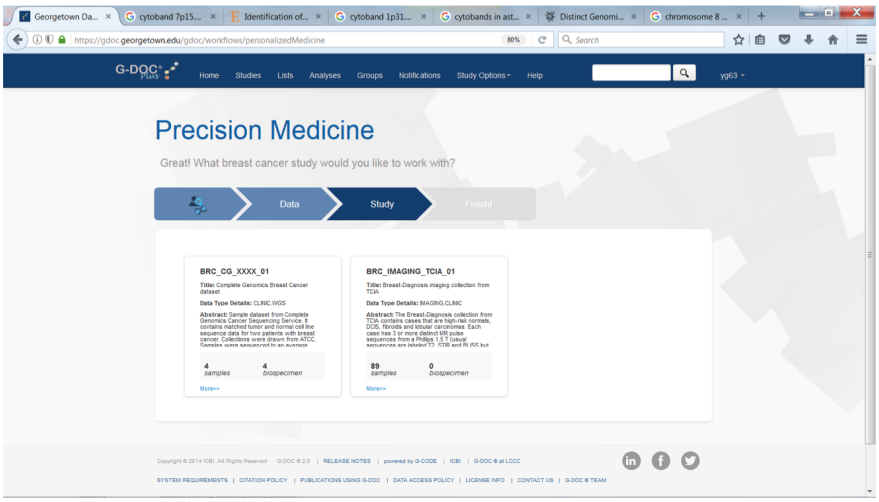
2. Click on the "Precision Medicine" module.



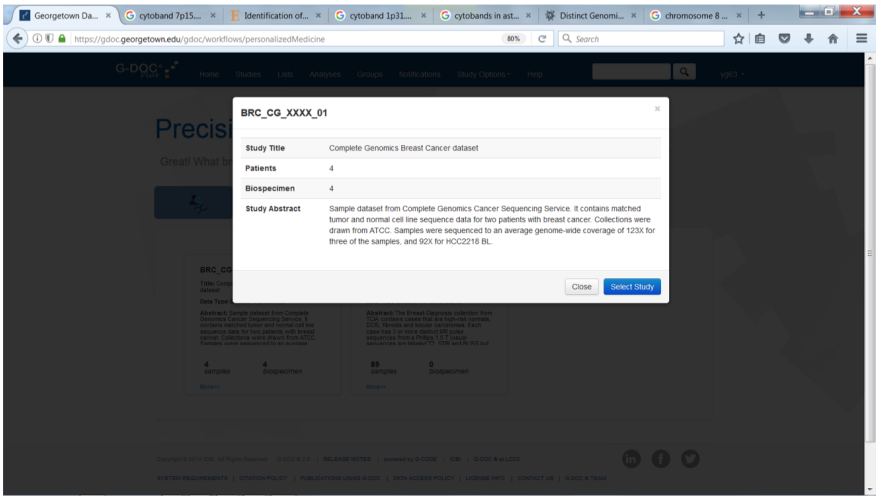
3. Click on the "breast cancer" study.



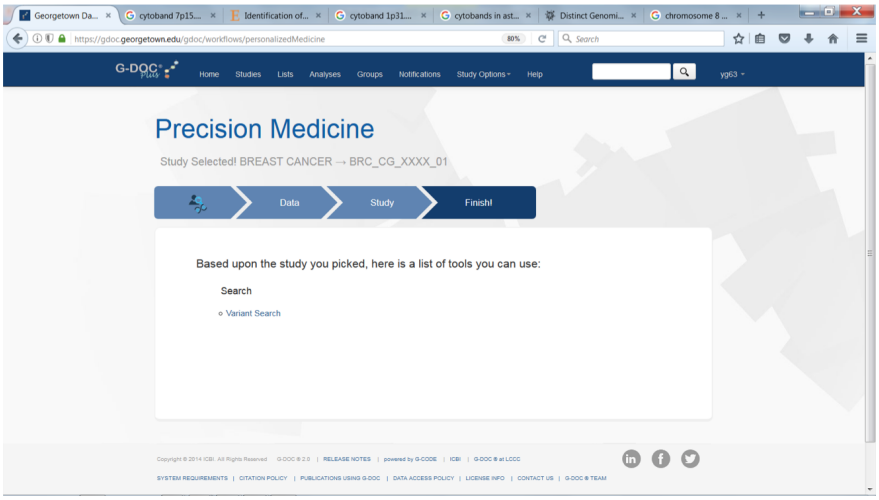
4. Open the study by clicking on "more."



5. Click the button "select study."

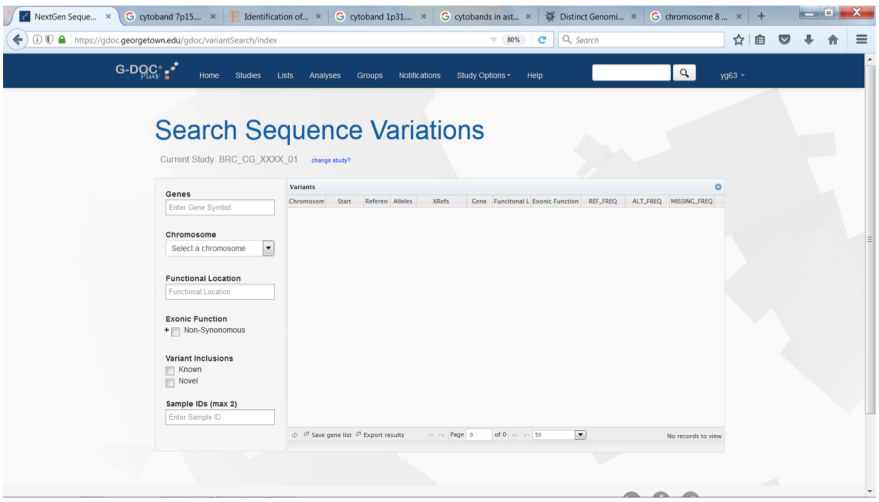


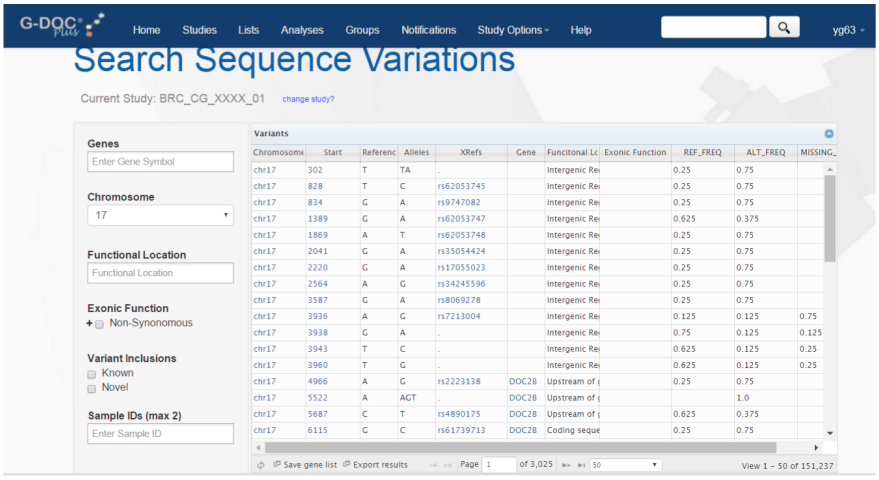
6. Select the exploratory analysis tool "variant search."



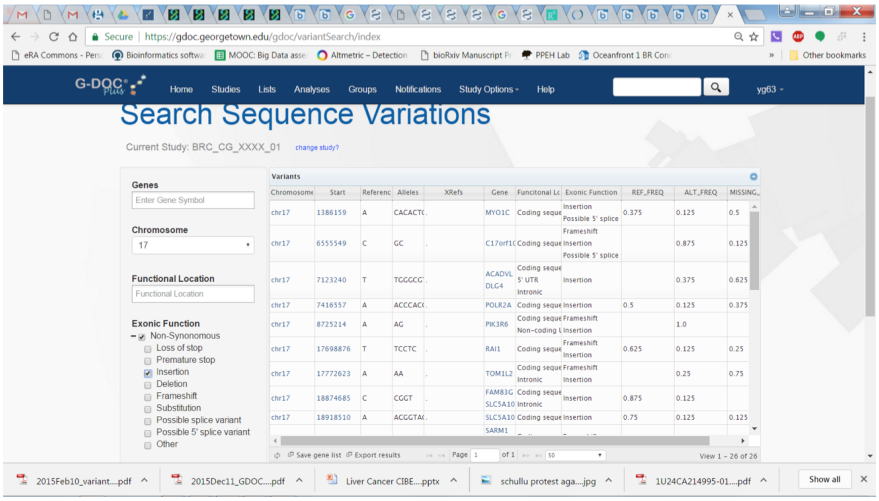
7. Select Chromosome 17 from the drop-down menu.

8. Explore the resulting table; specifically, check the total number of variants found.

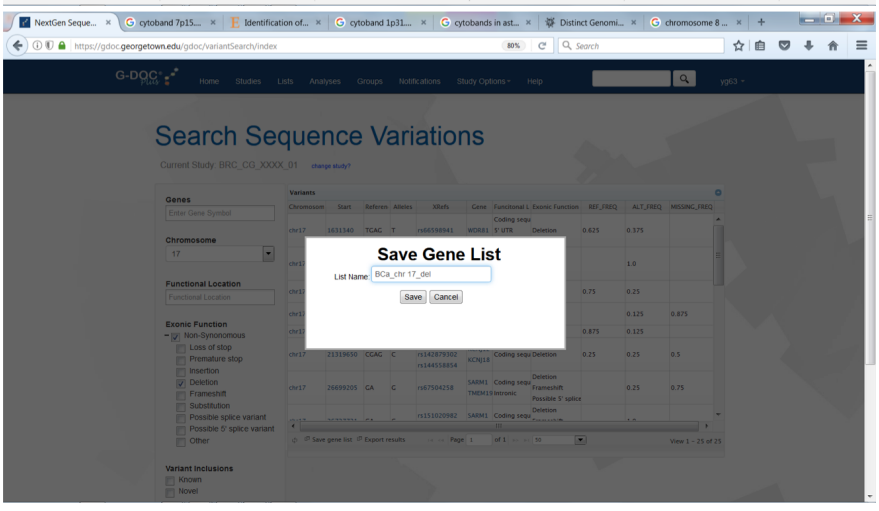




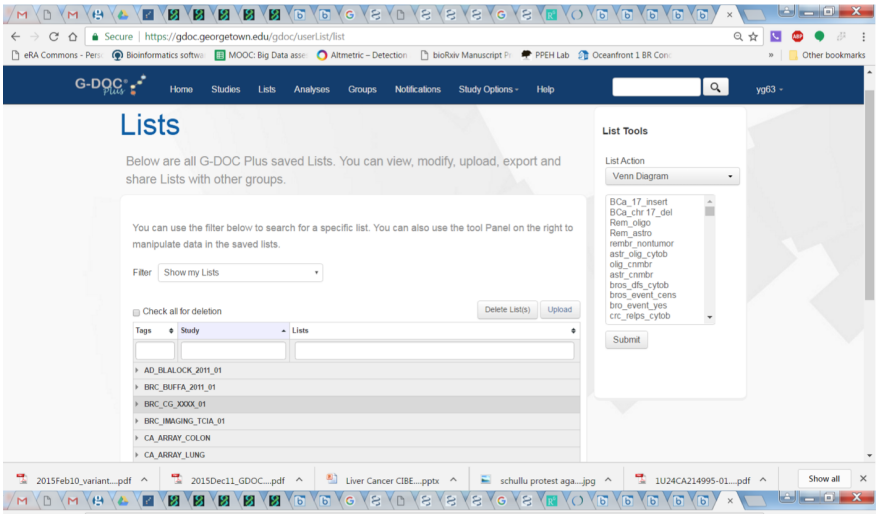
9. Select "insertion" from the menu of exonic function. After the filtering is complete, explore the resulting table. Check the total number of variants, noticing the drastic reduction of the total number of remaining variants.



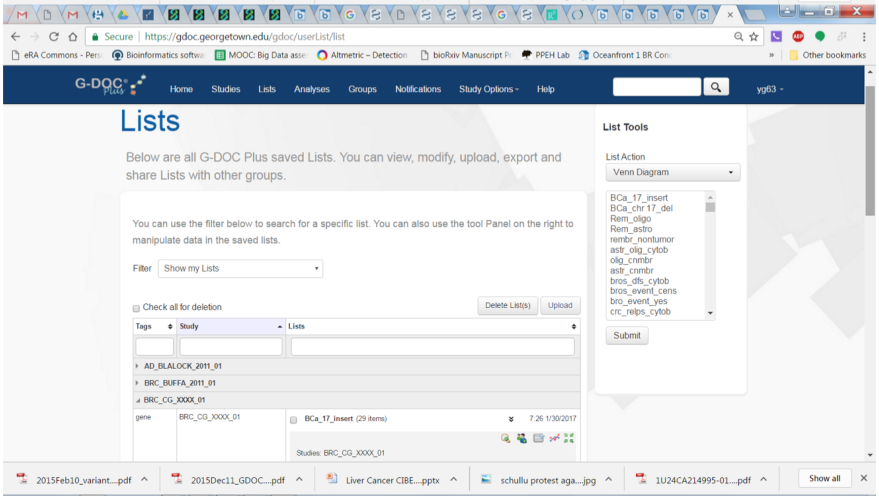
10. Select "gene list" from the filtered table. Type in the name of the list and click "save."



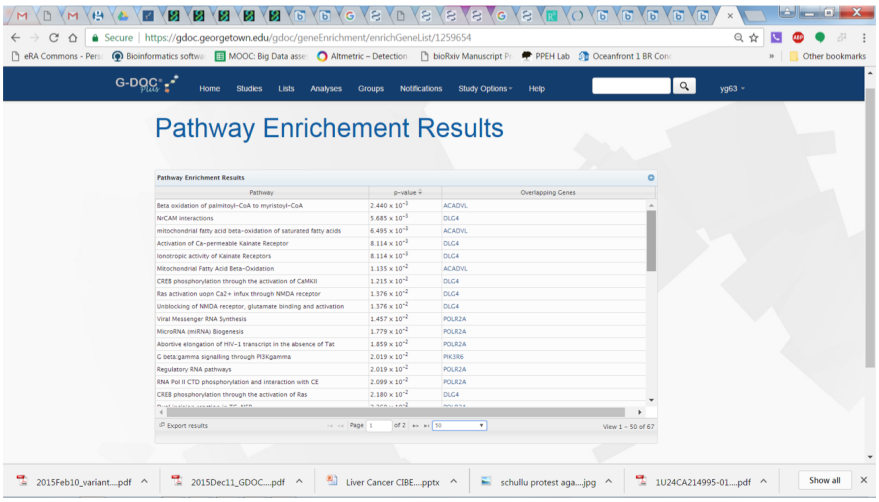
11. In the Perform Group Comparison Analysis window, open the Lists page from the main menu. Then find the study BRC_CG_XXX_01 and expand the list by clicking on the arrow.



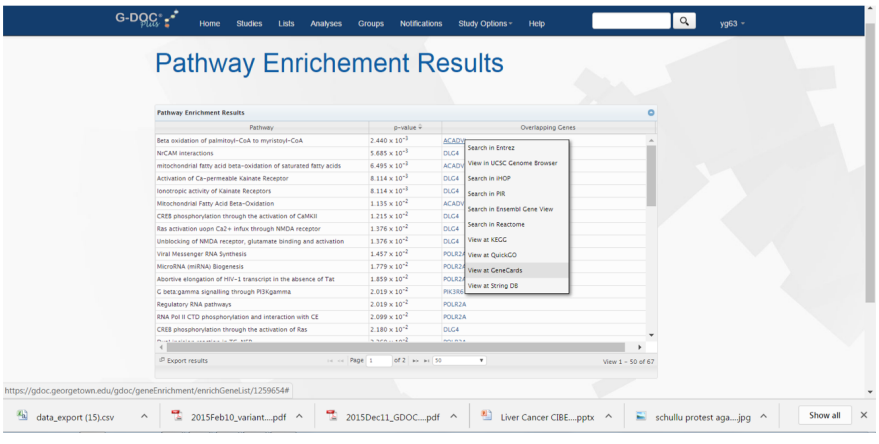
12. Find the list of gene names that you have saved and click on four green arrows (icon) to run the pathway enrichment analysis based on Reactome pathway collection.



13. Explore table with Pathway Analysis results. Export results.



14. Explore the table with pathways that are enriched with genes found in our analysis. Record the name of the top pathway listed in the first row of the table as well as the total number of pathways in the table (these are the answers to the first two questions). Be sure also to record the gene name in the top row that was mapped to this top pathway (top row in column "overlapping genes"). Explore the function of this gene in GeneCards.



15. Explore the function of this gene in Gene Cards at [this link]. Importantly, the main function of this gene is relevant to breast cancer. The protein encoded by this gene is targeted to the inner mitochondrial membrane where it catalyzes the first step of the mitochondrial fatty acid beta-oxidation pathway.