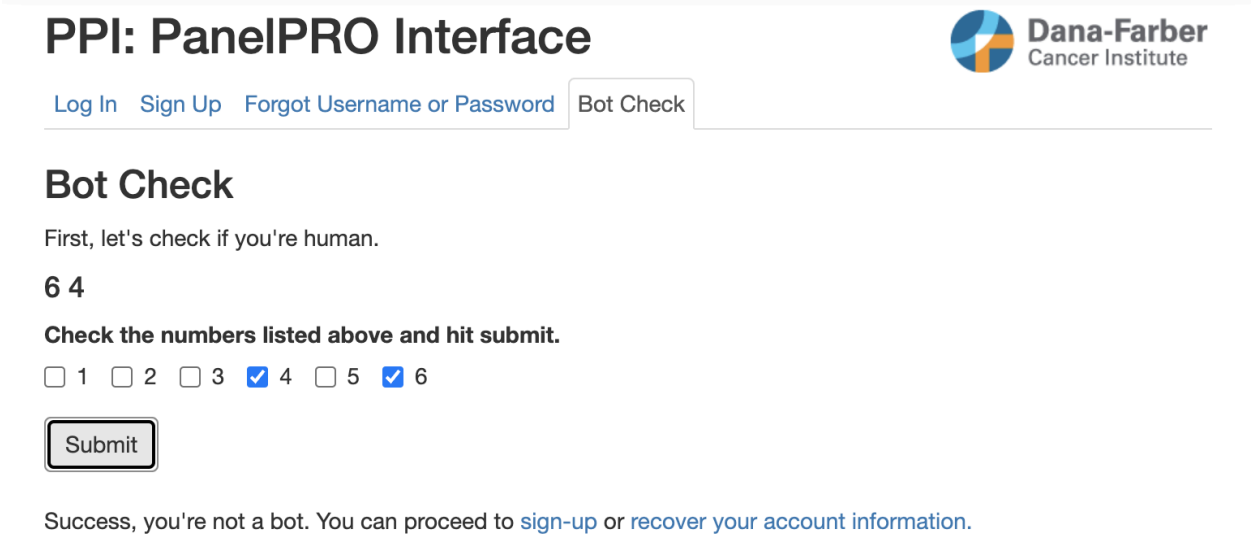
Fam3PRO Shiny App User Guide

Getting Started

1. Click either of the buttons to the sign-up page

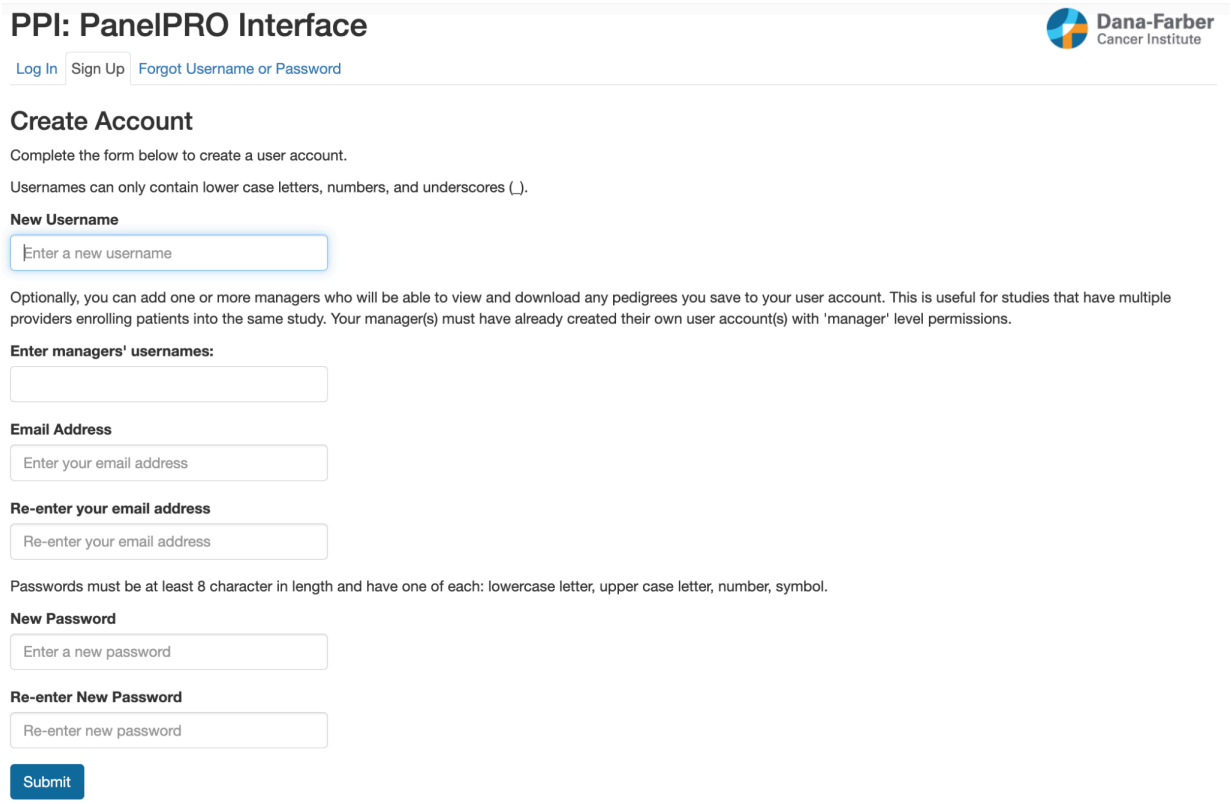
2. Click the numbers and hit “Submit” to pass the bot check



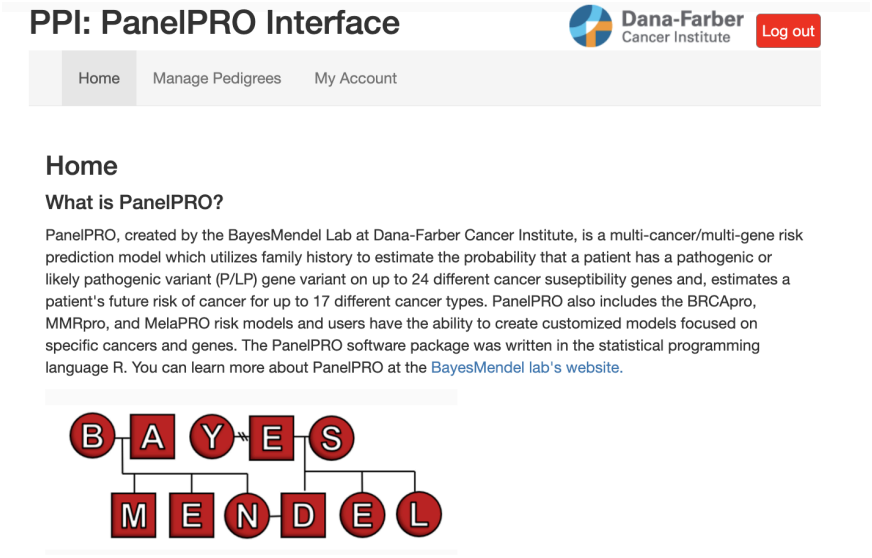
3. Enter information to finish creating a new account (“Enter manager username” is optional and can be left in blank)

   Username requirements:
   1. Usernames can only contain lowercase letters, numbers, and underscores (_)
   2. The length of your username should be less than 60 characters

   Password requirements:
   1. Your password must be at least 8 character in length
   2. Your password must contain one of each: lowercase letter (a-z), upper case letter (A-Z), number (0-9), symbol (example: !, @, #, $)



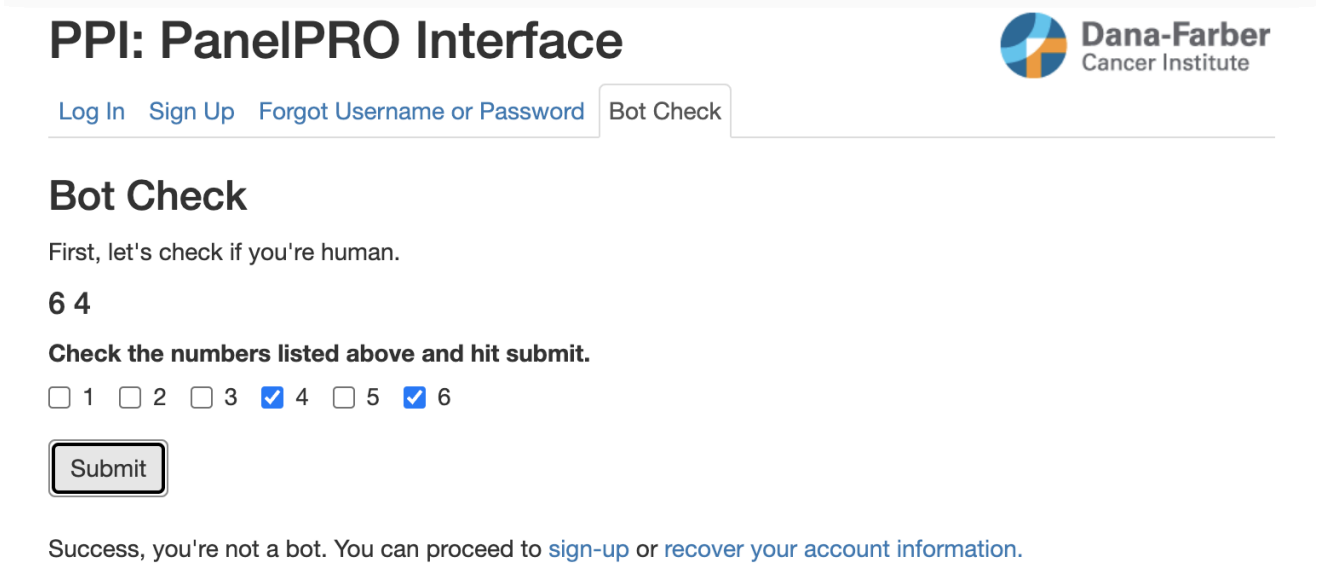
4. After the account is created, you will be guided back to the log-in page to enter your credentials. Once logging in, you will see the home page as shown below, and you are all set for using the application.



Forgot Username/Password

1. Click either of the buttons to the Username/Password recovery page

2. Click the numbers and hit “Submit” to pass the bot check



3. Enter the email address used for creating the account. If you only forgot your username, the username will be directly sent to the email address. If you forgot your password or both, you will be asked to enter a recovery code that will be sent to your email address. Once the recovery code is entered, you can reset your password.

Select which credentials you need to recover:

- ○ username
- ○ password
- ◉ both

Submit

Thank you. If the email address provided is in our system then you will receive one email with your username and a separate email with a password recovery code.

To reset your password, enter the recovery code below and submit it.

Recovery Code

Submit Recovery Code

If you don't receive the code after two minutes, click the button below.

Send New Code

Thank you. Please reset your password below.

Enter a new password

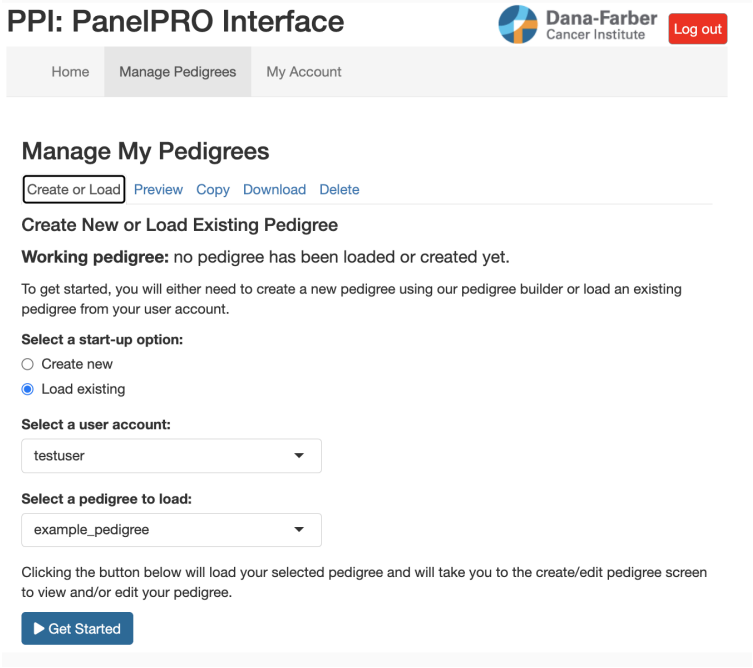
Re-enter the new password

Reset Password

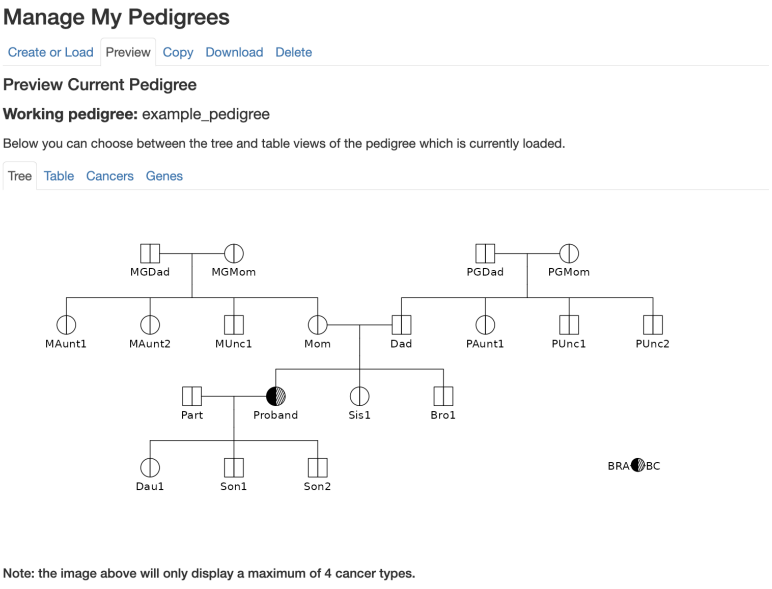
Step 1: Add/Load Pedigrees

1. Go to the "**Manage Pedigrees**" tab. You can either create a new pedigree from proband or use the default "Example Pedigree" as a template and make modifications on it later.

2. If you chose to load an existing pedigree, please wait for the Pedigree to load (which takes a few seconds) and click "Get Started" to proceed to the "Preview" page.



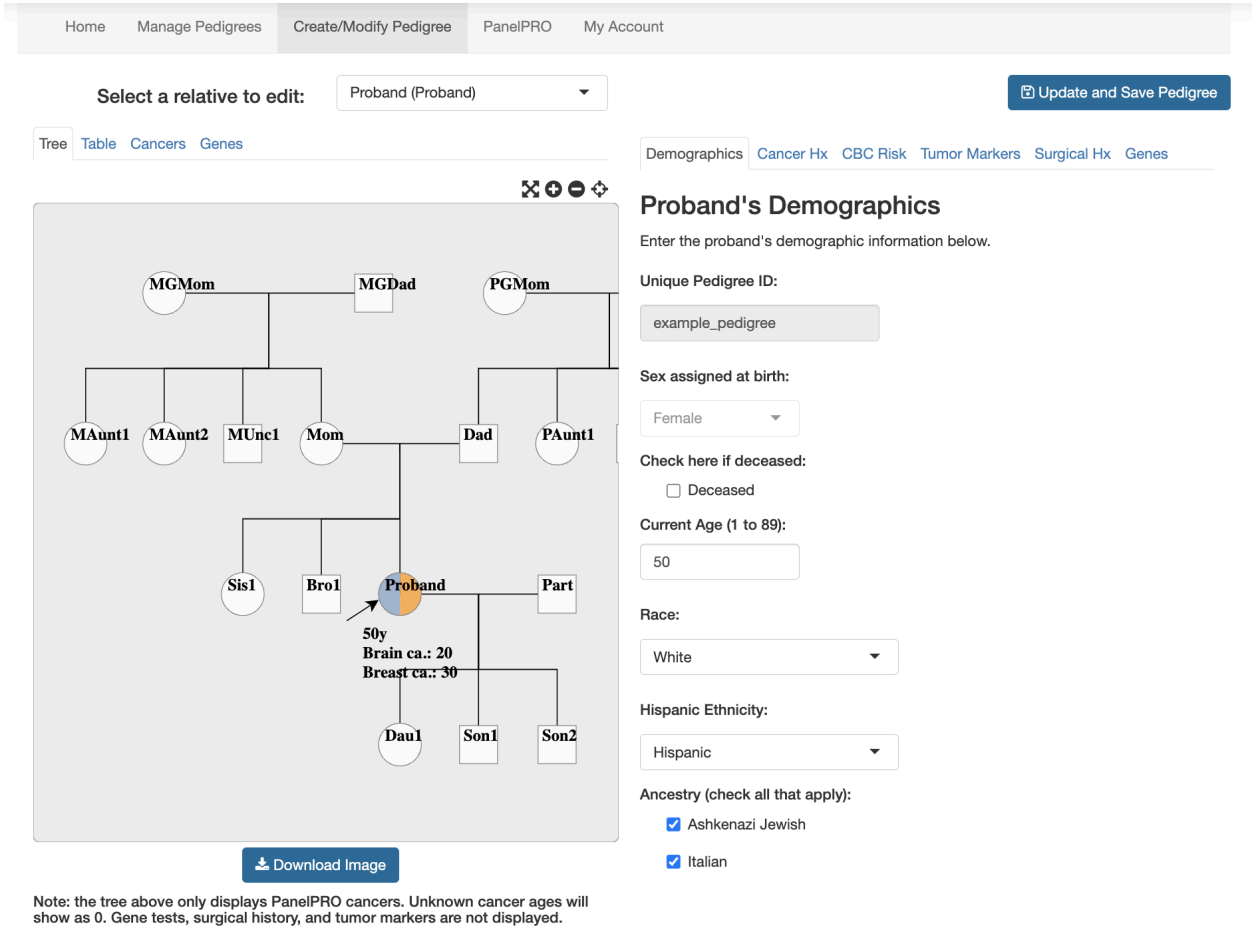
3. The "Preview" page allows you to look at the visualized pedigree.
   If you are a manager, you can load or use the "Copy" and "Delete" tab to copy/delete pedigrees from the associated accounts. The "Download" tab currently supports users downloading the pedigrees in either .csv or .rds format.



Note: the image above will only display a maximum of 4 cancer types.

Step 2: Create/Modify Pedigree

The "Create/Modify Pedigree" tab allows addition/deletion to elements in the working pedigree and display the pedigree in an interactive mode. Once you finish editing the pedigree, make sure to click **"Update and Save Pedigree"** to save your changes if storing pedigree information is allowed, or your changes will be lost. *Please double check your eligibility for saving the pedigrees with your organization!*



- **Left Panels:**

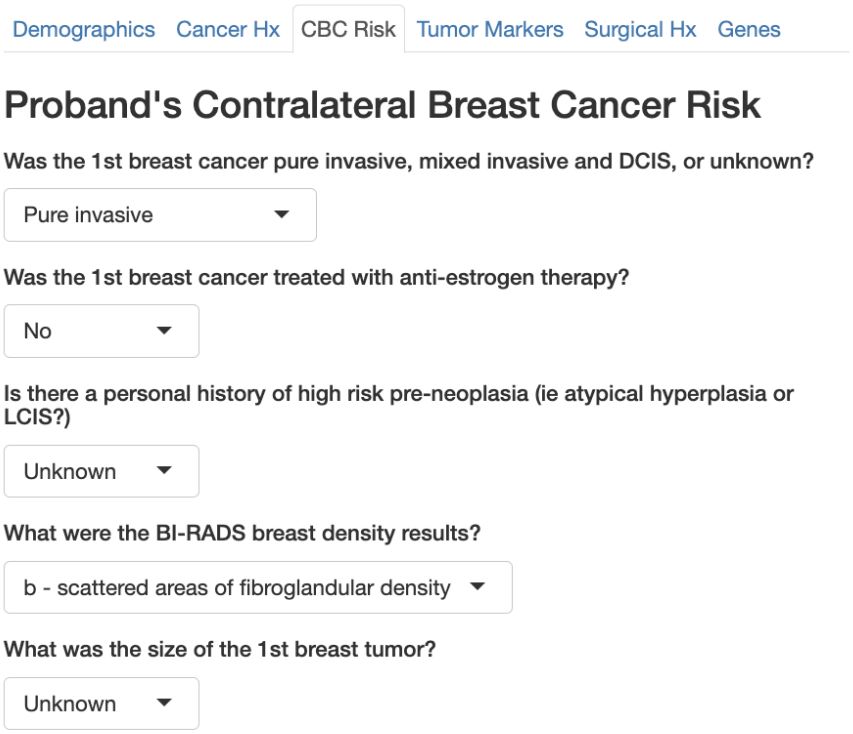
    1. Tree: an interactive tree that can be dragged around, where users can zoom in/out, show the perigree in full screen, manually adding/deleting nodes.
    2. Table: displaying the pedigree information as a dataframe, including all the basic information for all nodes in the pedigree.
    3. Cancers: displaying only the nodes with cancer diagnoses and relevant information (age of diagnosis, other cancer).
    4. Genes: showing the gene testing results for each gene variant and each node.

- **Right Panels:**

1. Demographics: panel for modifying demographic information for each node (to switch proband and relatives, use the drop-down tab above the panels: **“Select a relative to edit”**).

2. Cancer Hx: allows users to manually add cancer history for the proband and relatives, including cancer types diagnosis age (which can be left in blank if unknown).

3. CBC risk: allows users to specify the contralateral breast cancer risk for a node by choosing the answers for the questions below:



4. Tumor Markers: allows input for test results of any tumor markers for the related cancer of the node.

5. Surgical Hx: allows input for a node’s surgical history and the age at the specific surgery procedure.

6. Genes: allows users to enter and edit germline gene test results based on the instructions. Users can create/edit panels, edit panel results, and view results in a summary table.

Step 3: Run Fam3PRO

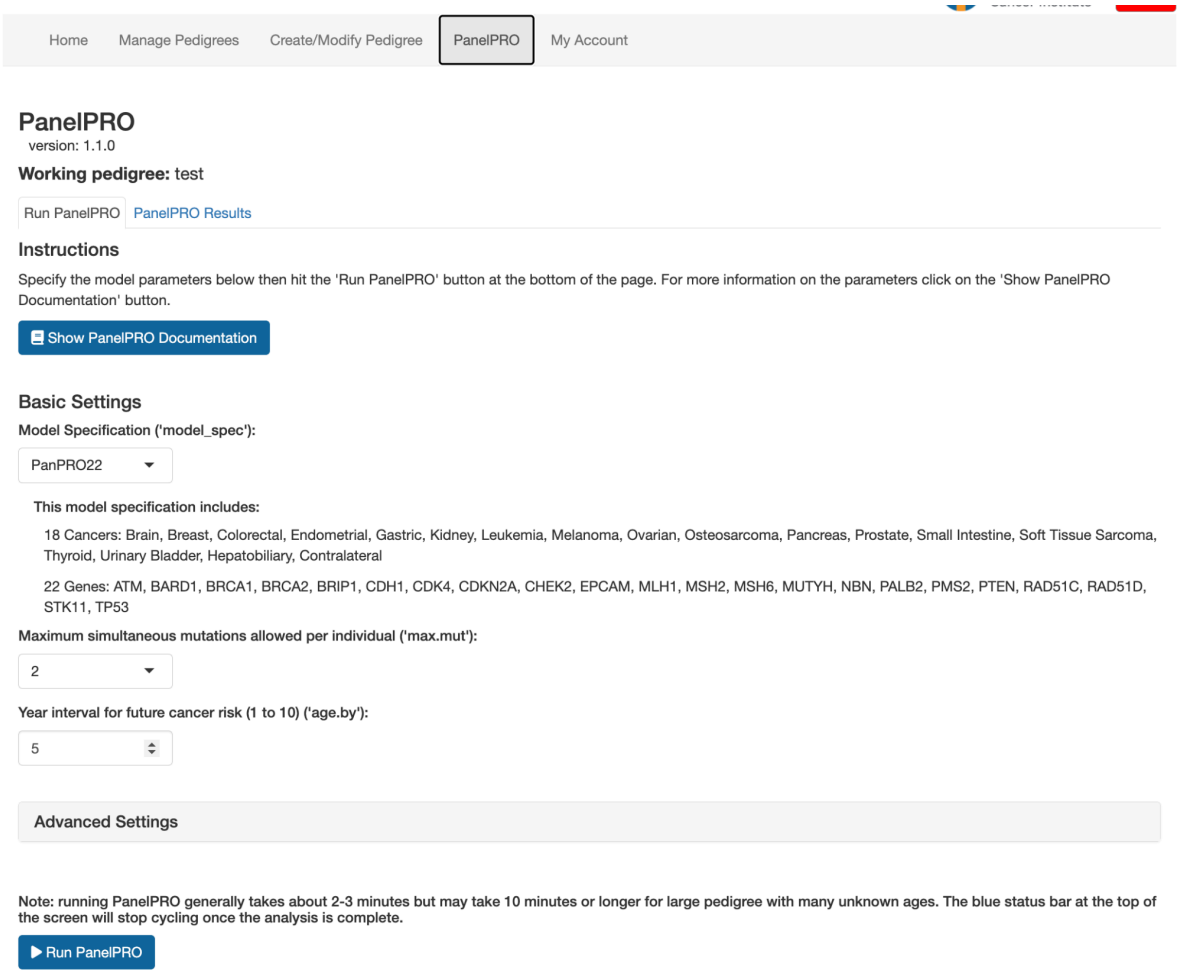
1. Hover to the “Fam3PRO” tab, you will see the current version of the Fam3PRO package running and the name of the working pedigree.

2. Click “Show Fam3PRO” Documentation if you want to learn further details of the Fam3PRO.

3. There are different options of model specifications available: PanPRO22 (default), BRCAPRO, BRCAPRO5, BRCAPRO6, MMRPRO, PanPRO11. Detailed cancer types and genes for each model are shown in respective specifications.

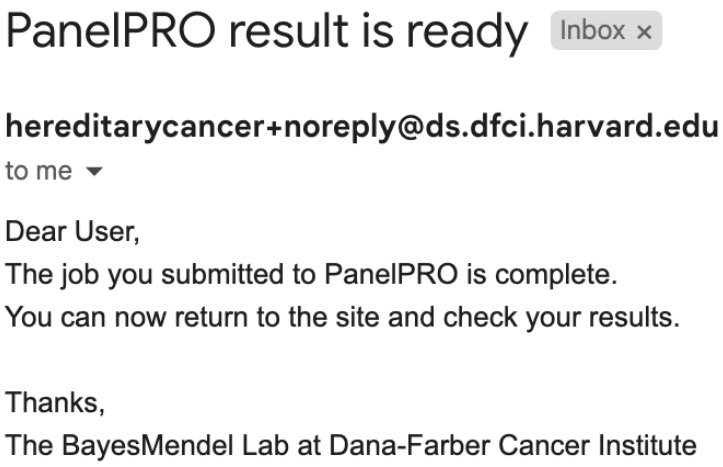
4. Adjust model parameters for maximum simultaneous mutations and year interval for future cancer risk.

5. Click “Run Fam3PRO”. You will see a notification below about the estimated time for completion based on the active processes running at the time **(please do not close the window)**. An email will be sent after the result is ready.



Step 4: Check Output Results

1. After running the Fam3PRO, you will receive an email notification when the process finishes. You will be automatically directed to the page with a cancer risk probability plot. If you have multiple processes running under one account, you will receive the email notification when the last process is complete.



PanelPRO result is ready Inbox ×

hereditarycancer+noreply@ds.dfci.harvard.edu

to me

Dear User,

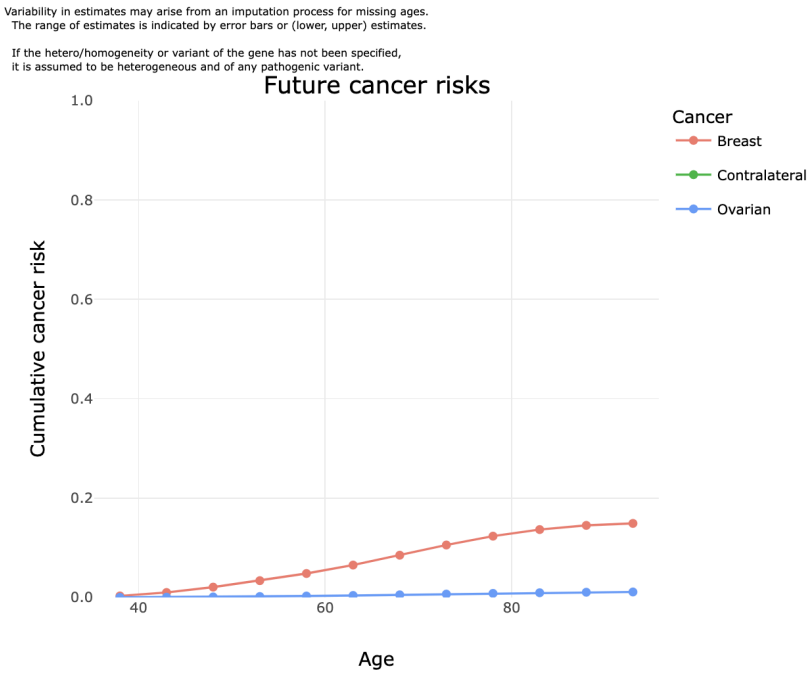
The job you submitted to PanelPRO is complete.

You can now return to the site and check your results.

Thanks,

The BayesMendel Lab at Dana-Farber Cancer Institute

2. You can switch between the tabs to look at the carrier probabilities and cancer risks in a table or a plot.



Thank you for using Fam3PRO!