



# **CancerGene Connect™ user guide**

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# GETTING STARTED

This guide explains the steps to collecting, assessing, and managing genetic family histories with CanceGene Connect's end-to-end software platform. You can navigate to and from information contained in this user guide by finding the topic in the table of contents, then clicking it to link directly to that page.

## KEY FEATURES

### ACCESSIBILITY

This tool is hosted in the Azure cloud—a computing service similar to the Amazon cloud. It is browser-based. The tool can be accessed from a HIPAA-compliant browser (i.e., a recently updated browser) and is mobile-friendly (i.e., it is designed for use on a smartphone or tablet).

### EASY-TO-ACCESS SIDEBAR MENU

At any time, you can click the sidebar menu to immediately navigate to any page in the questionnaire.

### USER-FRIENDLY RESPONSE FORMATS

Patient responses are collected in easy-to-use response formats. Inputting a response is as simple as clicking "yes" or "no" radio buttons, selecting answers from a drop-down menu, or entering text in a freeform box.

### USES BRANCHING LOGIC

A custom path through the questionnaire is created based on your patient's responses. Only those questions that are applicable to your patient will be presented to them.

### BUILT-IN DEFINITIONS AND PROMPTS

Throughout the questionnaire, blue information circles contain definitions and prompts for helping your patient to understand the health and medical terminology used in the questionnaire.

### BUILT-IN RISK MODELS

The platform has built in a series of risk models that conveniently run with the click of a button. Details are provided on the [Risk assessment page](#).

### AUTOSAVE FUNCTIONALITY

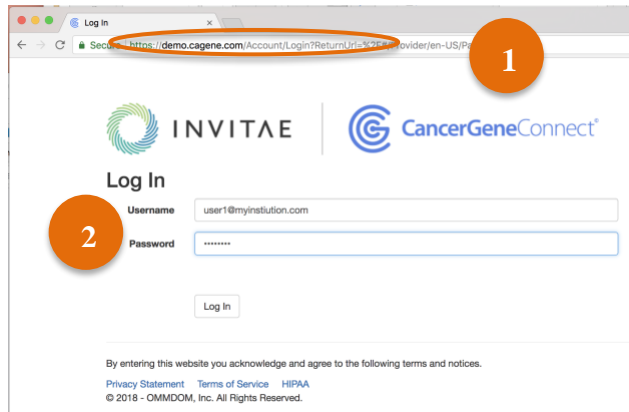
Your patient can log in to and out of the questionnaire as needed until they click **Submit** at the end of the questionnaire. Your patient's progress will be saved every time they click a "next" or "save" button.

### PRINTABLE QUESTIONNAIRES

Questionnaires are easy to print.

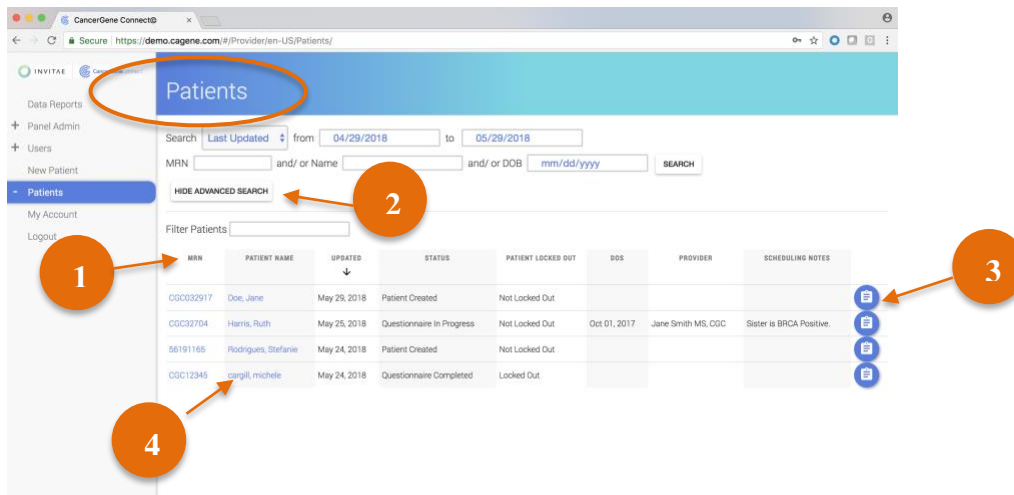
# HOW TO LOG INTO CANCERGENE CONNECT

1. Type your institution's custom CancerGene Connect web address into a HIPAA-compliant browser.
2. Enter in your login credentials (username and password).



# HOW TO NAVIGATE THE PATIENT DASHBOARD

1. When you log in as a provider, you are taken to the landing page of the Patients tab. This landing page provides you with a list of your patients, their MRNs, the dates their profiles were last updated, the status of their questionnaires, their current access to the questionnaire (i.e., whether or not they've been locked out), and the following optional fields: DOS (date of service), provider, and scheduling notes.
2. All patient information is searchable using the advanced search. Enter a date range, a partial MRN, a partial name, or a date of birth (DOB), then click **Search**.
3. Click the blue clipboard icon to access the [patient view](#) of the questionnaire.
4. Click a patient's name to access the [provider view](#) of the questionnaire.



## HOW TO USE THE SIDEBAR MENU

Upon logging in, you can access the sidebar menu on the left side of the screen. [Provider view](#) sidebars come with seven standard tabs. Show links to all the tab's pages by clicking the plus (+) symbol next to the tab's name. **NOTE:** Full directories of the provider-view and patient-view sidebar menus, as they relate to a selected questionnaire, are given in the [appendix](#). This user guide specifically highlights the [CancerGene Connect questionnaire](#).

### DATA REPORTS

This tab provides a comprehensive database of all your CancerGene Connect information. All reports can be downloaded into Excel. In addition to the five standard reports listed below, custom reports are available, by request, through our customer support team.

#### *ALLPOSITIVESREPORT*

This is a complete list of all patients who have a positive test result.

#### *GENEPOSITIVEREPORT*

Select a gene to return all patients with a positive test result for that selected gene.

#### *SPECIFICVARIANTREPORT*

Select a gene to return all variants for that selected gene.

#### *PSYCHCONCLUSIONS*

The report provides an overview of patient responses to four separate psycho-social assessments. The report produces patient scores and conclusions in addition to basic demographic data.

#### *MEGAREPORT*

Set a date range and receive output for all data points within CancerGene Connect.

### PANEL ADMIN

In this tab, you can build the custom genetic testing panels that are used by the "genetic testing tracking" feature on the Family page.

### USERS

Add, edit, and assign permission privileges using this tab. You can also view and track a history of CancerGene Connect site users. This tab is only available to providers for whom the "administrator" role has been enabled.

### NEW PATIENT

Use this tab to create a new patient in the system.

### PATIENTS

This tab includes the [patient dashboard landing page](#): Providers are automatically taken to this page upon logging in to the system. The Patient tab contains a number of pages that are specific

to the [questionnaire selection](#) which includes the ability for a provider to toggle between patient and provider views via the **Patient Side** subtab.

## MY ACCOUNT

In this tab, you can change your password or view this user guide.

## LOGOUT

Use this tab to log out of the CancerGene Connect platform.

**Expanded sidebar menu**

The screenshot shows the CancerGene Connect interface. On the left, a sidebar menu is expanded, showing options: Data Reports, Panel Admin, Users, New Patient, Patients (highlighted), My Account, and Logout. An orange arrow points from the 'Patients' menu item to the main content area. The main content area displays a 'Patients' table with columns: MRN, PATIENT NAME, UPDATED, STATUS, PATIENT LOCKED OUT, DOB, PROVIDER, and SCHEDULING NOTES. The table contains several rows of patient data. On the right, a detailed 'Expanded sidebar menu' is shown, listing various patient information categories: Data Reports, Panel Admin, Users, Patients (highlighted), Family, and others. An orange arrow points from the 'Patients' menu item in the sidebar to the 'Expanded sidebar menu'.

**Sidebar menu**

The screenshot shows the CancerGene Connect interface with the sidebar menu expanded. The sidebar menu includes: INVITAE, CancerGene Connect, Data Reports, Panel Admin, Users, New Patient, Patients, My Account, and Logout. An orange arrow points from the 'Patients' menu item to the 'Sidebar menu'.

# HOW TO CREATE A NEW PATIENT IN THE SYSTEM

1. Click the **New Patient** tab in the sidebar menu.
2. Enter a medical record number (MRN) or a patient identifier.
  - An MRN is required to move forward. It can be temporary.
  - The email, name, and date of service fields are optional.
3. Under **Questionnaire Options**, use the radio buttons to select the questionnaire(s) you wish to enable.
4. Click **Create**. The system will generate a username and password.

The screenshot shows the 'New Patient' form in the CancerGene Connect system. The interface includes a sidebar menu on the left with options: Users, Admin, New Patient (highlighted with callout 1), Patients, My Account, and Logout. The main form area has a title 'New Patient' and several input fields: MRN (with callout 2 pointing to it, containing 'OGC032917'), Email (optional), First Name (optional, containing 'Jane'), Last Name (optional, containing 'Doe'), and Date of Service (Optional) (month-day-year). Below these is a 'Questionnaire Options' section (with callout 3 pointing to it) containing a table with radio buttons for enabling or disabling various questionnaires. The 'CancerGene Connect® Questionnaire' is selected (callout 4 points to the 'CREATE' button). The 'CREATE' button is located at the bottom right of the form.

Questionnaire Options	Enabled	Disabled
Breast Health Questionnaire	<input type="radio"/>	<input checked="" type="radio"/>
CancerGene Connect® Questionnaire	<input checked="" type="radio"/>	<input type="radio"/>
Pedigree-Only Cancer Questionnaire	<input type="radio"/>	<input type="radio"/>
CancerGene Connect® Risk Ready Questionnaire	<input type="radio"/>	<input type="radio"/>



## HOW TO PROVIDE THE PATIENT WITH LOGIN CREDENTIALS

In the previous step, the system generated a unique username and password for the patient. These are the patient's login credentials. The patient will be able to access the questionnaire by visiting the institution's CancerGene Connect web address and signing in with the login credentials. You can get the login credentials to the patient using any of the three methods below.

1. **If you entered a valid email address:** If, in the previous step, you entered a valid patient email address, and *if the email feature has been activated in CancerGene Connect through customer support*, you can email the credentials directly to the patient by clicking **Generate and email**. The patient will receive a personalized email from the CancerGene Connect platform with unique login credentials and a link to the questionnaire. **NOTE:** The provider who is logged in to the system when the email is sent will show up as the email's sender.

Once the email is sent, the patient's status will change to "Login provided" on the **Patients** tab. Once the patient changes the temporary password, the status will update to "Questionnaire in progress."

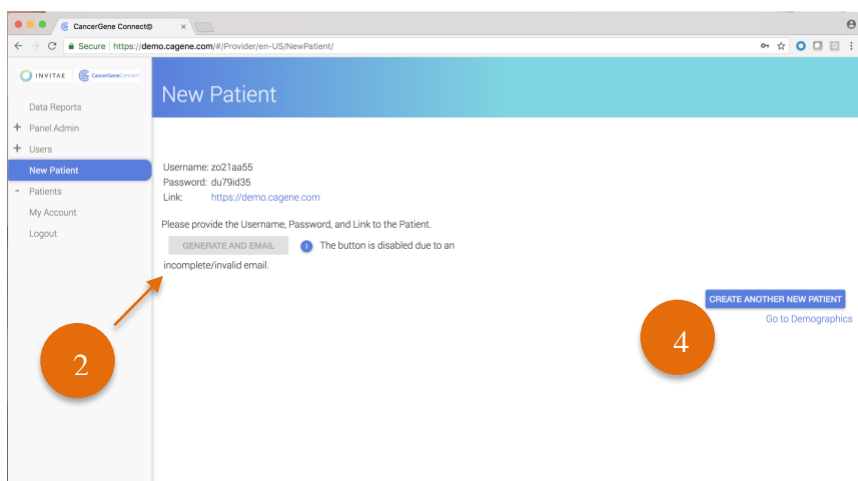
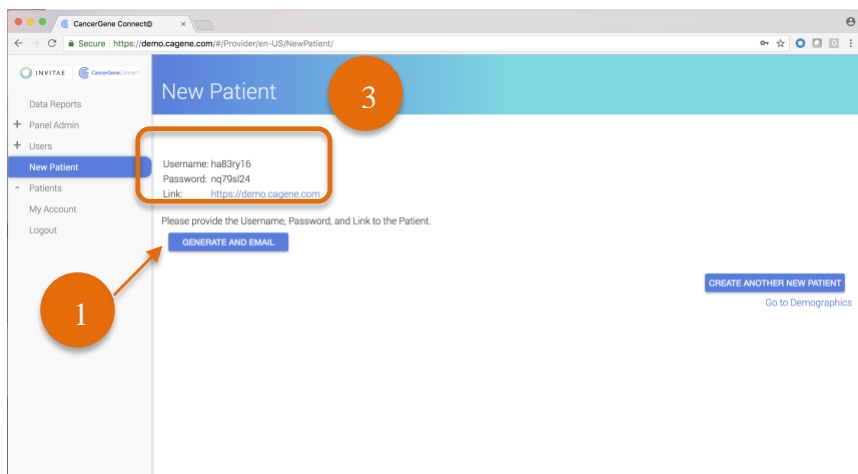
2. If an invalid email or no email is entered after clicking the **Create** button you will be taken to the screen with the Username, Password, and Link as usual. You will then see a greyed out "Generate and Email" button with a warning message regarding the invalid email.
  - You can alternatively copy and paste this information into an email you generate via your institutions webmail service.
  - You can distribute via a phone call.
  - You can navigate to the Patient Info tab and update the email field with a valid email address. Then send the credentials by clicking **Generate and email**.

Once the email is sent, the patient's status will change to "Login provided" on the **Patients** tab. Once the patient changes the temporary password, the status will update to "Questionnaire in progress."

3. **If there is no email address:** Make note of the username, password, and link URL, and distribute it in a phone call to the patient.

*As a HIPAA compliance measure, upon receipt of their login credentials, patients must reset their password to complete their questionnaires.*

Once you have disseminated a new patient's login credentials, you can create another patient by selecting **Create another new patient**, or you can begin entering your patient's demographic information in the Demographics page by clicking **Go to Demographics**.



## QUESTIONNAIRE SELECTION

CancerGene Connect includes four standard questionnaires, described below. See the [appendix](#) for a complete list of the pages and sections included in each questionnaire, for each view ([patient](#) [table 1] and [provider](#) [table 2]). NOTE: Because it contains nearly all available pages and sections (including those found in the other three questionnaires), the CancerGene Connect questionnaire is highlighted in this user guide and is depicted in the instructional images.

### BREAST HEALTH QUESTIONNAIRE

This questionnaire is very specific to breast health management and is used primarily by breast surgeons and professionals working within the breast specialty. The Breast Health questionnaire includes specific questions about breast disease and breast management.

### CANCERGENE CONNECT QUESTIONNAIRE

This is an exhaustive genetic counseling questionnaire that includes detailed questions about personal, health, and family history. It includes questions on colon disease and management. The

family history is used to generate a pedigree, and related information from the questionnaire feeds into the built-in risk models.

### PEDIGREE-ONLY CANCER QUESTIONNAIRE

This questionnaire generates a pedigree using in-depth family history questions in addition to a few demographic questions.

### CANCERGENE CONNECT RISK READY QUESTIONNAIRE

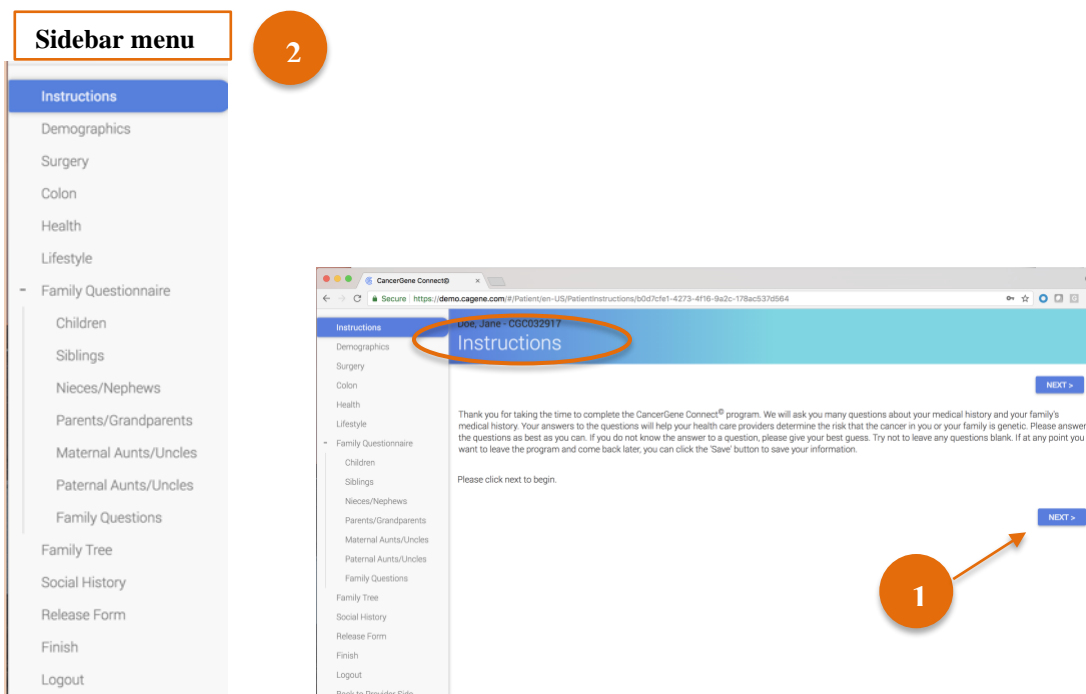
This questionnaire includes specific questions whose answers are necessary for the risk models that run in the background.

## PATIENT VIEW OF THE (CANCERGENE CONNECT) QUESTIONNAIRE

### INSTRUCTIONS PAGE

Upon logging in with their self-created password, patients are taken to the Instructions page. This landing page provides information on the types of questions that will be asked and how to complete the questionnaire. It also explains that patients can log in and out as needed and that their progress is saved every time they click a "next" or "save" button.

1. Click **Next** to begin filling out the questionnaire.
2. Directly access any page in the questionnaire by using the **sidebar menu**. Click the name of the desired page to navigate to it.



## DEMOGRAPHICS PAGE

The Demographics page includes questions related to a variety of demographic characteristics.

**NOTE:** To accurately run the risk models, some questions are required; patients will not be able to proceed until these questions are answered. On the Demographics page, questions that require answers include the Gender and Birth Date fields.

Demographics

First Name: Ruth, Middle Name: Kagan, Last Name: Harris

Gender (sex): ☒ Male ☒ Female

Birth Date (month day year): October 26 1989

Street Address: 1206 Glassy Way

City: New York, State: New York, Zip: 10058

Home Phone: 212-965-8875, Work Phone: 202-659-8544, Mobile Phone: 917-368-8745, Email Address: ruth.harris@yahoo.com

Were you adopted? ☐ Yes ☒ No

Are you Ashkenazi Jewish? ☒ Yes ☐ No

Healthcare Coverage: Aetna

Are you Black or African American? ☐ Yes ☒ No

Are you Hispanic or Latino? ☐ Yes ☒ No

## OBGYN AND HORMONAL HISTORY PAGES

The OBGYN and Hormonal History pages include questions on female reproductive health.

**NOTE:** These pages only populate for female patients, and only those questions that are material to the patient are included.

OBGYN

Have you ever had a period? ☒ Yes ☐ No

Your age when periods started: 12

Are you currently pregnant? ☒ Yes ☐ No

Your due date? (month day year)

Which of the following best describes you? I am having periods off and on

Your total number of live births: 2

Your age at first live birth: 22

Your age at last live birth: 25

Your total number of stillborns, miscarriages, or abortions: 0

Your total number of pregnancies: 2

CancerGene Connect®

Secure <https://demo.cagene.com/#/Patient/en-US/Hormonal/7d330bad-8641-4008-8460-201c27bdf5ea>

Harris, Ruth - CGC32704

## Hormonal History

< PREVIOUS SAVE NEXT >

Do you use, or have you ever used, birth control pills, shots or patches? (Do not include other types)

☒ Yes ☐ No

How old were you when you first started using birth control pills, shots or patches?

18

Are you still using birth control pills, shots or patches?

☐ Yes ☒ No

How old were you when you stopped using birth control pills, shots or patches?

21

Over your whole life, how many years have you used birth control pills, shots or patches?

3

Do you use, or have you ever used, hormone replacements?

☒ Yes ☐ No

What hormone replacements have you used?

Estrogen and progestin

How old were you when you started taking hormone replacements?

43

## SURGERY PAGE

The Surgery page includes questions on breast- and reproductive organ-related surgeries and screenings.

CancerGene Connect®

Secure <https://demo.cagene.com/#/Patient/en-US/Surgery/7d330bad-8641-4008-8460-201c27bdf5ea>

Harris, Ruth - CGC32704

## Surgery

< PREVIOUS SAVE NEXT >

Have you ever had a mammogram?

☒ Yes ☐ No

How old were you when you had your first mammogram?

40

When did you have your most recent mammogram? (month year)

January 2016

Where did you have your most recent mammogram?

Beth Israel Medical Center

Have you ever had a breast biopsy?

☐ Yes ☒ No ☐ Do Not Know

Has a doctor ever told you that you had breast cancer?

☐ Yes ☒ No

Has a doctor ever told you that you have changes in your breast that may lead to cancer (precancerous)?

☐ Yes ☒ No

Has a doctor ever told you that you have benign breast disease?

☐ Yes ☒ No

## COLON PAGE

The Colon page includes questions on colon-related procedures and disease management.

Harris, Ruth - CGC32704  
Colon

Have you ever had a colonoscopy? <sup>1</sup> During a colonoscopy, a doctor uses long flexible tube to check your colon (intestines) and remove polyps if necessary. Your doctor gives instructions before the test for a liquid diet the day before the exam and you will typically take strong laxatives or other medications to completely clear your bowels before the exam. You are usually given a relaxing medicine or are put to sleep for this procedure. You go home from the appointment the same day.  
☒ Yes ☐ No

When did you have your most recent colonoscopy?  
More than 1 but less than 5 years ago

Has a doctor ever told you that you had colon polyps? <sup>1</sup>  
☐ Yes ☒ No

Has a doctor ever told you that you had colon (bowel) or rectal cancer?  
☐ Yes ☒ No

Have you ever had colon surgery?  
☐ Yes ☒ No

Have you ever had an upper endoscopy? <sup>1</sup>  
☐ Yes ☒ No

How often do you take anti-inflammatory medications? (For example: Advil, Aleve, ibuprofen, Motrin, Naprosyn, Indocin)  
Occasionally/As needed

## HEALTH PAGE

The Health page includes questions on disease-management therapies, medication management, and supplement management. Because the information collected on this page is not material to the risk models, inclusion of the Health page is optional.

Harris, Ruth - CGC32704  
Health

Have you ever had radiation therapy? ☒ Yes ☐ No

Have you ever had chemotherapy? ☒ Yes ☐ No

Have you ever had a bone marrow transplant? ☐ Yes ☒ No

Are you taking any medications for pain or inflammation (such as ibuprofen, codeine, vicodin, cortisone)? ☐ Yes ☒ No

Are you taking any medications for preventing seizures (such as carbamazepine, diazepam, phenytoin, valproic acid)? ☐ Yes ☒ No

Are you taking any medications for cancer treatment (such as allopurinol, carboplatin, docetaxel, paclitaxel)? ☐ Yes ☒ No

Are you taking any medications for Parkinson's treatment (such as biperiden, levodopa, carbidopa)? ☐ Yes ☒ No

Are you taking any medications for preventing blood clotting (such as heparin, warfarin)? ☐ Yes ☒ No

Are you taking any medications for heart conditions or high blood pressure (such as verapamil, digoxin, amlodipine, hydrochlorothiazide)? ☒ Yes ☐ No

Are you taking any medications for high cholesterol (such as simvastatin, crestor, zetia)? ☐ Yes ☒ No

Are you taking any medications for kidney disease (such as furosemide, mannitol)? ☐ Yes ☒ No

## LIFESTYLE PAGE

The Lifestyle page gathers information on risk factors such as alcohol and tobacco use, caffeine consumption, and exercise. The height and weight entries generate the body mass index (BMI), which is available for viewing in the provider view. For risk modeling purposes, answers to the height and weight questions are required.

The screenshot shows the 'Lifestyle' page in the CancerGene Connect interface. The patient's name 'Harris, Ruth - OGC32704' is at the top. The left sidebar lists various medical history sections, with 'Lifestyle' currently selected. The main content area contains a form with the following fields: 'About how tall are you?' (5 ft, 5 in), 'About how much do you weigh?' (148 lbs), 'Marital status' (Married/Living with partner), 'What is your highest level of education?' (Postgraduate), 'Are you currently working for pay?' (Yes, full time), 'Occupation' (Lawyer), 'How often do you drink alcohol?' (One drink a day), 'Do you drink beer?' (No), 'Do you drink wine?' (No), 'Do you drink hard liquor?' (No), and 'Have you ever used tobacco or nicotine products?' (No).

## FAMILY QUESTIONNAIRE

This section includes questions about the cancer histories of your patient and your patient's family. Answers to the questions in this section generate the pedigree. ***These pages cannot be altered.***

1. Your patient enters their own cancer history and age at diagnosis.
  - Click the **Add cancer** button to list a cancer. Select the type of cancer from the dropdown menu; if the applicable cancer is not listed, choose "Other" and type in the information. Patients who have never been diagnosed with cancer do not need to click the **Add cancer** button; alternatively, they may choose "No Cancer" from the dropdown menu.
  - To delete a line entry, click the **Remove** button.
2. Your patient enters the cancer histories and ages at diagnoses of all living and deceased family members, including half-siblings, half-aunts, and half-uncles, but excluding non-blood relatives.
  - Click the **Add** button to add a family member.
  - Enter the family member's name. This will be helpful to the patient for keeping track of family members while filling out the family questionnaire. From a provider viewpoint, being able to refer to the patient's family members by their names helps you to create a personal connection with your patient. ***If names are not entered, the platform defaults to a naming convention that indicates the type(s) and number of relatives entered (e.g., Daughter 1, Son 2, Brother 3).***
  - To delete a line entry, click the **Remove [name]** button.



## FAMILY QUESTIONNAIRE LANDING PAGE

**Family Questionnaire**

Harris, Ruth - CGC32704

1

Instructions

The next few pages ask about all of your relatives in order to draw your family tree. **Please include ALL living AND deceased family members.** If you do not know all of your family members, provide as much information as possible. If you are not sure about the exact answers, please give your best guess. Please include half-siblings and half-aunts/uncles. Do not include non-blood relatives.

You will be asked the following details about your family:

- Current age or age at death
- Type of cancer, if applicable
- Age at cancer diagnosis
- To save you time, it is acceptable to only include your cousins that have cancer

You may include names of family members in the corresponding text box. This is optional and may help you when using your family tree later.

Your History

If you have been diagnosed with cancer, click the **add cancer** button to start. If you have not had cancer, click next to start filling out your family history information.

Cancer: Ovarian Cancer Age Diagnosed: 43

**ADD CANCER** **REMOVE**

**Dropdown menu**

- Liomyoma
- Leukemia
- Leukopenia
- Liver Cancer
- Lung Cancer
- Lymphoma, Non-Hodgkin's
- Mediastinal Carcinoma
- Medulloblastoma
- Melanoma
- Menigioma
- Myelodysplastic Syndrome (MDS)
- Myeloma
- Nephroblastoma
- Neuroblastoma
- Neurofibroma
- Oncocytoma
- Osteosarcoma
- Ovarian Cancer**
- Ovarian Fibroma
- Pancreatic Cancer
- Pancreatic Cancer, Adenocarcinoma
- Pancreatic Cancer, Islet Cell
- Pancreatic Cancer, Neuroendocrine
- Pancytopenia
- Paraganglioma
- Parathyroid Cancer
- Peritoneal Cancer
- Pheochromocytoma
- Pinealoblastoma
- Pituitary Tumor
- Pleuropulmonary Blastoma
- Polyps, Adenomatous
- Polyps, Hamartomatous
- Polyps, Hyperplastic

## CHILDREN PAGE

**Children**

Harris, Ruth - CGC32704

2

Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives.

**ADD Daughters**

Name: Susan Still Alive? ☒ Yes ☐ No ☐ Do Not Know Age Now or At Death: 26 **REMOVE SUSAN**

**ADD CANCER**

Name: Sloan Still Alive? ☒ Yes ☐ No ☐ Do Not Know Age Now or At Death: 1 **REMOVE SLOAN**

**Family member name**

**ADD CANCER**

**ADD Sons**

Name: James Still Alive? ☒ Yes ☐ No ☐ Do Not Know Age Now or At Death: 23 **REMOVE JAMES**

**ADD CANCER**



## SIBLINGS PAGE

Harris, Ruth - CGC32704  
Siblings

Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives.

**ADD** Sisters

Name	Still Alive?	Age Now or At Death	REMOVE
Sarah	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	51	REMOVE SARAH
<b>ADD CANCER</b>			
Susie	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	55	REMOVE SUSIE
Cancer	Age Diagnosed		
Cervical Cancer	45		REMOVE
<b>ADD CANCER</b>			
Maggie	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	46	REMOVE MAGGIE
<b>ADD CANCER</b>			

**ADD** Brothers

Name	Still Alive?	Age Now or At Death	REMOVE
Danny	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	--	REMOVE DANNY

## NIECES/NEPHEWS PAGE

Harris, Ruth - CGC32704  
Nieces/Nephews

Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives.

**ADD** Sarah's Daughters (Your Nieces)

Name	Still Alive?	Age Now or At Death	REMOVE
Callie	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	27	REMOVE CALLIE
Cancer	Age Diagnosed		
Basal Cell Carcinoma	22		REMOVE
<b>ADD CANCER</b>			
Charlie	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	25	REMOVE CHARLIE
<b>ADD CANCER</b>			

**ADD** Sarah's Sons (Your Nephews)

Name	Still Alive?	Age Now or At Death	REMOVE
Caleb	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	21	REMOVE CALEB
<b>ADD CANCER</b>			

**ADD** Susie's Daughters (Your Nieces)

## PARENTS/GRANDPARENTS PAGE

CancerGene Connect®

Secure | <https://demo.cagene.com/#/Patient/FamilyMember/en-US/Parents-Grandparents/7d330bad-8641-4008-8460-201c27bdfea>

Home, Ruth - UGU32704

### Parents/Grandparents

2

Instructions

Demographics

OB/GYN

Hormonal History

Surgery

Colon

Health

Lifestyle

Family Questionnaire

Children

Siblings

Nieces/Nephews

**Parents/Grandparents**

Maternal Aunts/Uncles

Paternal Aunts/Uncles

Family Questions

Maternal Cousins

Paternal Cousins

Family Tree

Social History

Release Form

Finish

Logout

Back to Provider Side

Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives.

< PREVIOUS SAVE NEXT >

**Your Mother**

Name: Mom

Still Alive? ☒ Yes ☐ No ☐ Do Not Know

Age Now or At Death: 72

Both ovaries removed? ☐ Yes ☐ No ☐ Do Not Know

Cancer: Breast Cancer Age Diagnosed: 58 REMOVE

Cancer: Pancreatic Cancer Age Diagnosed: 72 REMOVE

ADD CANCER

**Your Father**

Name: Dad

Still Alive? ☒ Yes ☐ No ☐ Do Not Know

Age Now or At Death: 78

ADD CANCER

**Your Mother's Mother (Maternal Grandmother)**

Name: MGMother

Still Alive? ☐ Yes ☒ No ☐ Do Not Know

Age Now or At Death: 79

Cancer: Polyps, Unknown Age Diagnosed: 60 REMOVE

ADD CANCER

**Your Mother's Father (Maternal Grandfather)**

Name: MGFather

Still Alive? ☐ Yes ☒ No ☐ Do Not Know

Age Now or At Death: 80

ADD CANCER

**Your Father's Mother (Paternal Grandmother)**

Name: PGMother

Still Alive? ☐ Yes ☒ No ☐ Do Not Know

Age Now or At Death: 62

Cancer: Breast Cancer Age Diagnosed: 61 REMOVE

ADD CANCER

**Your Father's Father (Paternal Grandfather)**

Name: PGFather

Still Alive? ☐ Yes ☒ No ☐ Do Not Know

Age Now or At Death: 65

Cancer: Lung Cancer Age Diagnosed: 65 REMOVE

ADD CANCER

## MATERNAL AUNTS/UNCLES PAGE

**Maternal Aunts/Uncles**

Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives.

**ADD Your Mother's Sisters (Maternal Aunts)**

Name	Still Alive?	Age Now or At Death	REMOVE
Sally	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	61	REMOVE SALLY
<b>ADD CANCER</b>			
Mary	<input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know		REMOVE MARY
<b>ADD CANCER</b>			

**ADD Your Mother's Brothers (Maternal Uncles)**

Name	Still Alive?	Age Now or At Death	REMOVE
Wilbur	<input type="radio"/> Yes <input checked="" type="radio"/> No <input type="radio"/> Do Not Know	25	REMOVE WILBUR
<b>ADD CANCER</b>			
Orville	<input type="radio"/> Yes <input checked="" type="radio"/> No <input type="radio"/> Do Not Know	77	REMOVE ORVILLE
<b>ADD CANCER</b>			

< PREVIOUS SAVE NEXT >

## PATERNAL AUNTS/UNCLES PAGE

**Paternal Aunts/Uncles**

Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives.

**ADD Your Father's Sisters (Paternal Aunts)**

Name	Still Alive?	Age Now or At Death	REMOVE
Lisa	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	50	REMOVE LISA
<b>ADD CANCER</b>			
Louise	<input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Do Not Know	50	REMOVE LOUISE
<b>ADD CANCER</b>			

**ADD Your Father's Brothers (Paternal Uncles)**

< PREVIOUS SAVE NEXT >

**NOTE:** In the provider view, on the [pedigree page](#), you will have the option to add in great grand relatives (e.g., great grandparents, great aunts, great uncles, grandchildren, grand nieces, grand nephews). You can also add and note extended family members (i.e., those family members with an unknown relationship to the patient). They will appear at the bottom of the pedigree.

## FAMILY QUESTIONS PAGE

After the patient completes their family histories, they will proceed to the Family Questions page. This page includes several yes/no questions on topics on which patients have historically provided inaccurate information. The answers to these questions will help provide accurate family and risk-assessment information.

1. This page includes questions on whether or not any family members have had their ovaries removed, there are any identical twins or half relatives in the family, or there are any affected cousins. If there are affected cousins, the [maternal cousins and paternal cousins pages](#) will populate and the patient can add information on any cousins with cancer.
2. If a patient answers "yes" to any of the questions on the Family Questions page, there will be a visual cue in the [Family page](#) on the provider side that reminds you to follow up with the patient and to enter in the details.

The screenshot shows the 'Family Questions' page in the CancerGene Connect interface. The page title is 'Family Questions' and the patient name is 'Harris, Ruth - CGC32704'. The page contains four questions with radio button options:

- Have any family members had their ovaries removed?  
☐ Yes ☒ No
- Are there any identical twins in your family?  
☒ Yes ☐ No
- Are any of your family members half relatives?  
☒ Yes ☐ No
- Are there any cousins with cancer in your family?  
☒ Yes ☐ No ☐ Unknown

A red circle with the number '1' is placed next to the first three questions. The page also includes a sidebar with navigation links: OBGYN, Hormonal History, Surgery, Colon, Health, Lifestyle, Family Questionnaire (with sub-links: Children, Siblings, Nieces/Nephews, Parents/Grandparents, Maternal Aunts/Uncles, Paternal Aunts/Uncles, Family Questions, Maternal Cousins, Paternal Cousins, Family Tree, Social History, Release Form, Finish, Logout), and a top navigation bar with '< PREVIOUS', 'SAVE', and 'NEXT >' buttons.

## MATERNAL COUSINS AND PATERNAL COUSINS PAGES

If, on the [Family Questions page](#), a patient indicates that there are any affected cousins, the Maternal Cousins and Paternal Cousins pages will populate so that the patient can add in any cousins with cancer. NOTE: If it's desired to flesh out the family pedigree, patients and providers can add in any non-affected cousins.

The screenshot shows the 'Maternal Cousins' page in the CancerGene Connect interface. The left sidebar contains a menu with options: Demographics, OBGYN, Hormonal History, Surgery, Colon, Health, Lifestyle, Family Questionnaire, Children, Siblings, Nieces/Nephews, Parents/Grandparents, Maternal Aunts/Uncles, Paternal Aunts/Uncles, Family Questions, **Maternal Cousins**, Paternal Cousins, Family Tree, Social History, Release Form, Finish, Logout, and Back to Provider Side. The main content area has a header 'Harris, Ruth - CGC32704' and a title 'Maternal Cousins' circled in orange. Below the title are navigation buttons: '< PREVIOUS', 'SAVE', and 'NEXT >'. A note states: 'Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives. For the purposes of this questionnaire, only include your cousins that have cancer.' There are five 'ADD' buttons for adding cousins: 'Sally's Daughters (Your Maternal Female Cousins)', 'Sally's Sons (Your Maternal Male Cousins)', 'Mary's Daughters (Your Maternal Female Cousins)', 'Mary's Sons (Your Maternal Male Cousins)', and 'Wilbur's Daughters (Your Maternal Female Cousins)'. The 'Orville's Daughters (Your Maternal Female Cousins)' button is highlighted with an orange box. Below these buttons is a form for adding a cousin. The 'Name' field contains 'Sheila', 'Still Alive?' has radio buttons for 'Yes' (selected), 'No', and 'Do Not Know', 'Age Now or At Death' is '45', 'Cancer' is 'Ovarian Cancer', and 'Age Diagnosed' is '35'. There are 'REMOVE SHEILA' and 'REMOVE' buttons. At the bottom is an 'ADD CANCER' button.

The screenshot shows the 'Paternal Cousins' page in the CancerGene Connect interface. The left sidebar contains a menu with options: OBGYN, Hormonal History, Surgery, Colon, Health, Lifestyle, Family Questionnaire, Children, Siblings, Nieces/Nephews, Parents/Grandparents, Maternal Aunts/Uncles, Paternal Aunts/Uncles, Family Questions, Maternal Cousins, **Paternal Cousins**, and Family Tree. The main content area has a header 'Harris, Ruth - CGC32704' and a title 'Paternal Cousins' circled in orange. Below the title are navigation buttons: '< PREVIOUS', 'SAVE', and 'NEXT >'. A note states: 'Please include living AND deceased family members. Please include half siblings and half aunts/uncles. Do not include non-blood relatives. For the purposes of this questionnaire, only include your cousins that have cancer.' There are four 'ADD' buttons for adding cousins: 'Lisa's Daughters (Your Paternal Female Cousins)', 'Lisa's Sons (Your Paternal Male Cousins)', 'Louise's Daughters (Your Paternal Female Cousins)', and 'Louise's Sons (Your Paternal Male Cousins)'. At the bottom are navigation buttons: '< PREVIOUS', 'SAVE', and 'NEXT >'.

## FAMILY TREE PAGE

The Family Tree page ("[Pedigree](#)" in the provider view) is generated from the patient's answers to the [family questionnaire](#). Patients can see their pedigrees and may access some of the Pedigree page's tools and features.

1. **Style** – Use the dropdown to toggle between *Primary Colors*, *Black*, and *White*.
2. **Size** – Set the margins for printing the family tree. Choose *Full Width* or *Landscape*. You can also select *Internet Explorer (IE) Copy/Paste* to ensure that the pedigree prints properly when using IE.
3. **Legend Location** – Use the dropdown menu to select the position of the legend.
4. **Hide** – Tick the boxes to exclude information from the family tree display.
5. The pedigree has both slide and zoom settings that are compatible with macOS and Windows applications.

The screenshot displays the "Family Tree" page for patient Ruth Harris (CGC32704). The page includes a sidebar with navigation options like Instructions, Demographics, OBGYN, Hormonal History, Surgery, Colon, Health, Lifestyle, and Family Questionnaire. The main area shows a pedigree chart for Ruth Harris (Oct 26, 1969) with a legend for various cancer types: Lung Cancer (red), Leukemia (green), Cervical Cancer (yellow), Breast Cancer (pink), Pancreatic Cancer (orange), Ovarian Cancer (light green), Basal Cell Carcinoma (purple), and Polyps, Unknown (cyan). The pedigree chart shows three generations of family members, with some individuals marked with cancer types. Below the chart, there are settings for Style (Primary Colors), Size (Full Width), Legend Location (Top-Left), and Hide options (Names, Patient Name/MRN, Ages, Cancers, etc.).

## SOCIAL HISTORY PAGE

The Social History page includes four informal psycho-social assessments. Questions gauge the patient's depression (CES-D), social support (ISEL), personal and family cancer history impact (IES-R), and cancer worries. See the [appendix](#) for details on how these assessments are scored and conclusions are drawn.

Because the information collected here is not material to the risk models, inclusion of the Social History page is optional. When this page is included in the patient's questionnaire, patient scores and conclusions are highlighted on the [Psych page](#) of the provider view.

Harris, Ruth - CGC32704

### Social History

< PREVIOUS SAVE NEXT >

During the past WEEK, how often did you feel or behave this way

	Rarely or none of the time ( < 1 day )	Some or a little of the time ( 1 - 2 days )	Occasionally or a moderate amount of the time ( 3 - 4 days )	Most or all of the time ( 5 - 7 days )
I was bothered by things that usually do not bother me	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I had trouble keeping my mind on what I was doing	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt depressed	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt that everything I did was an effort	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I felt hopeful about the future	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I felt fearful	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
My sleep was restless	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt happy	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt lonely	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I could not get 'going'	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>

Social Support

## RELEASE FORM PAGE

The Release Form page enables the patient to provide questionnaire-based medical reports and consultation letters to a member of their medical care team (e.g., their physician). The patient clicks **Add physician** and is prompted to include the care team member's name, address, and phone number. There is no limit on the number of physicians that can be added. If physicians are included, their names will carbon copy on any documentation generated on the provider side; they will also be visible on the Release Form page of the provider view.

Because the information collected here is not material to the risk models, inclusion of the Release Form page is optional.

**NOTE:** This page does not auto-release any information.

Harris, Ruth - CGC32704

### Release Form

< PREVIOUS SAVE NEXT >

To send medical reports to any of your physicians, please fill in their name, address, and phone numbers below.

Physician Name  
Dr. Bob Nussbaum

Address 1  
1400 18th Street

Address 2

City  
San Francisco

State  
California

Zip  
94103

Phone Number  
555-555-5555

Fax Number  
555-555-1234

DELETE

ADD PHYSICIAN

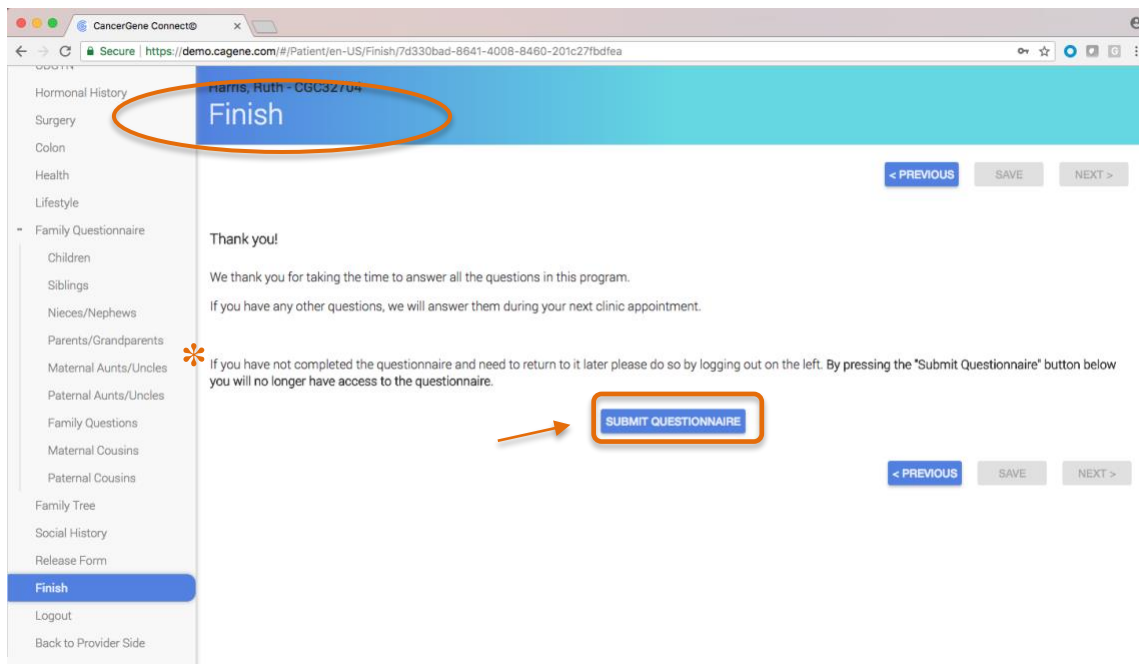
< PREVIOUS SAVE NEXT >



## FINISH PAGE

The Finish page is the last page of the patient questionnaire. On this page, the patient submits the questionnaire using the **Submit questionnaire** button. Once the questionnaire is submitted, the patient is locked out and will be unable to make any edits. Upon submission, the provider can review the patient's questionnaire.

There may be some instances in which the patient accidentally locks themselves out of the questionnaire or the provider wishes to turn their access on and off. Refer to the [provider view](#) section of this user guide for instructions on granting and revoking patient access to the questionnaire.



## PROVIDER VIEW OF THE QUESTIONNAIRE

This section describes the platform's **provider view**. Please refer to the beginning of this user guide for information about [logging in](#), [creating a new patient](#), [questionnaire selection](#), [patient dashboard](#), and [sidebar menu](#) navigation.

**NOTE:** Full directories of the sidebar menu as it relates to a selected questionnaire are given in the [appendix](#). This user guide specifically highlights the [CancerGene Connect questionnaire](#).

### IMPORTANT PROVIDER NOTES

- In the provider view, all the information seen on the **Demographics page** through the **Health page** is a mirror image of the patient view and reflects the answers that the patient has provided. For more information on these pages, refer to the [patient view](#) section of this user guide.
- Use the [sidebar menu](#) to switch between patient view and provider views.
- If a mistake has been made or a patient has updated information to add after submitting their questionnaire, a provider can update, edit, and re-save a patient's questionnaire in either the patient view or the provider view.

### PATIENT INFO PAGE

Patient Info is an administrative page on which the provider can view, add, and edit information about their patient or their patient's appointment. Information available on this page includes date of service, patient email address, the status of the questionnaire, a list of additional providers who may be working on the case, and scheduling notes. The provider can also use this page to enable and disable questionnaire options, add the patient type (new or follow-up), note whether or not the provider has entered data or if the patient was seen under a research setting, and update or change the patient's MRN. This page also covers three additional important provider functions:

1. **Patient access** — View and grant patient access to the questionnaire from the **Patient Locked Out** dropdown menu.
2. **Generate a new patient password** — If you have a valid email address for the patient, you can click **Generate and email** to generate the new password and send it to the patient. If you do not have a valid email address, click **Generate and display**. A new password will be generated, which you can copy and paste into a secure institutional email or give to your patient in person or over the phone. Upon logging in with the new credentials, patients are prompted to change their passwords.
3. **Delete a patient** — Click the red **Delete patient** button. You will be prompted to enter the patient's MRN; afterward, click **Confirm patient deletion**.

CancerGene Connect®

Secure https://demo.cagene.com/#/Provider/en-US/PatientInfo/7d330bad-8641-4008-8460-201c27bdf5ea

INVITAE CancerGene Connect®

Harris, Ruth - CGC32704

## Patient Info

SAVE

1

Date of Service (month day year)    Email  Status  Patient Locked Out ☐ Not Locked Out ☒ Locked Out

Provider 1  Provider 2

Scheduling Notes

Questionnaire Options

	Enabled	Disabled
Breast Health Questionnaire	<input type="radio"/>	<input checked="" type="radio"/>
CancerGene Connect® Questionnaire	<input checked="" type="radio"/>	<input type="radio"/>
Pedigree-Only Cancer Questionnaire	<input type="radio"/>	<input checked="" type="radio"/>
CancerGene Connect® Risk Ready Questionnaire	<input type="radio"/>	<input checked="" type="radio"/>

Patient Type ☐ New ☐ Follow Up

☐ Provider Entered Data ☐ Research

Change MRN (requires browser refresh)

2

Generate new password

New password for qq35pv65 is **hh48kc35**

Username

3

Confirm Patient MRN to delete:

SAVE

## UPDATES PAGE

The Updates page enables a provider to track patients after their appointments. Providers may use this page to determine which patients they might continue to see in a research, screening, or clinical trials setting.

The screenshot shows the 'Updates' page for patient 'Harris, Ruth - CGC32704'. The page is titled 'Updates' and has a 'SAVE' button in the top right corner. The left sidebar contains the following navigation options: Data Reports, Panel Admin, Users, New Patient, Patients (expanded), Patient Info, Updates (selected), Demographics, OBGYN, Hormonal History, Surgery, Colon, Health, Lifestyle, Psych, Pedigree, Family, Risk Assessment, Documentation, Patient Records, Release Form, Patient Side, My Account, and Logout. The main content area contains the following sections:

- This patient is deceased**  
☐ Yes ☐ No
- Other family tested**  
☐ Yes ☐ No
- New oral contraception**  
☐ Yes ☐ No
- Changed Diet**  
☐ Yes ☐ No
- Increased Exercise**  
☐ Yes ☐ No
- New Cancer Diagnosis (After Mutation Identified)**
  - Patient Diagnosed with Breast Cancer**  
☐ First primary ☐ Second primary ☐ N/A
  - Patient diagnosed with ovarian cancer**  
☐ Yes ☐ No
  - Patient diagnosed with endometrial cancer**  
☐ Yes ☐ No
  - Patient diagnosed with colon cancer**  
☐ Yes ☐ No
- Screening/Prevention**
  - Breast chemoprevention**  
☒ Yes ☐ No
  - Select**  
Tamoxifen

The Updates page, which is optional, is a popular addition.

## LIFESTYLE PAGE

For risk modeling purposes, answers to the height and weight questions are required.

The Lifestyle page gathers information on risk factors such as alcohol and tobacco use, caffeine consumption, and exercise. There is also a field for documenting the patient's head circumference.

The system uses the patient's height and weight values to generate the body mass index (BMI). Because the BMI (which is only viewable in the provider view) is material to risk modeling, the patient's height and weight values are required.

The screenshot shows the 'Lifestyle' page for patient 'Harris, Ruth - CGC32704'. The page is titled 'Lifestyle' and has a 'SAVE' button. The form includes the following fields:

- Height: 'About how tall are you? (feet/inches)' with input fields for feet (5) and inches (5).
- Weight: 'About how much do you weigh? (lbs.)' with input field 148.
- BMI: 'BMI' with input field 24.6.
- Head circumference: 'Head circumference (cm.)' with an empty input field.
- Marital status: 'Married/Living with partner' (dropdown menu).
- Education: 'What is your highest level of education?' with dropdown menu 'Postgraduate'.
- Occupation: 'Are you currently working for pay?' (Yes, full time) and 'Occupation' (Lawyer).
- Alcohol: 'How often do you drink alcohol?' (One drink a day).
- Drinking habits: 'Do you drink beer?' (No), 'Do you drink wine?' (Yes), 'Do you drink hard liquor?' (Yes).
- Tobacco: 'Have you ever used tobacco or nicotine products?' (Yes, sporadic).
- Cigarettes: 'Have you ever smoked cigarettes?' (No).
- Cigar: 'Have you ever smoked a cigar?' (Yes).
- Pipe: 'Have you ever smoked a pipe?' (Yes).
- Snuff: 'Have you ever used snuff?' (No).
- Coffee/tea/soda: 'Do you drink coffee/tea/soda?' (Yes) and 'How many cups per day?' (empty field).

## PSYCH PAGE

The Psych page, which is optional, is comparable to a distress protocol. It includes four informal psycho-social assessments that gauge a patient's depression (CES-D), social support (ISEL), personal and family cancer history impact (IES-R), and cancer worries. See the [appendix](#) for details on how these assessments are scored and conclusions are drawn.

This page mirrors the [Social History page](#) in the patient view. The difference in naming convention between the patient and provider view supports patient adherence to filling out the information. When the Social History page is included in the patient's questionnaire, the provider will see a list of scores on the Psych page, as well as a yes-or-no conclusion based on the patient's responses.

**Harris, Ruth - CGC3271-1**  
**Psych**

SAVE

Section	Score	Conclusion
Mood	15/30	Depression(CES-D)
Social Support	5/12	Social Support(SEL)
Thoughts About Cancer	12/24	Cancer Specific Stress
Cancer Worries	7/12	"Cancer Worries" Section Does Not Generate Conclusions

During the past WEEK, how often did you feel or behave this way

	Rarely or none of the time ( < 1 day)	Some or a little of the time (1 - 2 days)	Occasionally or a moderate amount of the time (3 - 4 days)	Most or all of the time (5 - 7 days)
I was bothered by things that usually do not bother me	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I had trouble keeping my mind on what I was doing	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt depressed	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt that everything I did was an effort	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I felt hopeful about the future	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
I felt fearful	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>
My sleep was restless	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt happy	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
I felt lonely	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>

## PEDIGREE PAGE

The Pedigree page generates a pedigree using either the answers your patient provided on their [Family Questionnaire](#) or information you've entered into the **Family** section (available on the provider view's sidebar).

### 1. Add family members and their histories.

- Click the **Add** button to add a family member.
- Enter the family member's name. This will be helpful for keeping track of family members throughout the questionnaire. From a provider viewpoint, being able to refer to the patient's family members by their names can help you to create a personal connection with your patient. ***If names are not entered, the platform defaults to a naming convention that indicates the type(s) and number of relatives entered (e.g., Daughter 1, Son 2, Brother 3).***
- Enter the cancer histories and ages at diagnoses of all the patient's living and deceased family members, including half-siblings, half-aunts, and half-uncles, but excluding non-blood relatives.
- To delete a line entry, click the **Remove [name]** button.

### 2. Add or edit additional information about family members.

- For each family member, you can provide additional information on ethnicity, ovary removal, twinning, half relatives, bilateral prophylactic mastectomies, bilateral breast cancer, pathology results, side indications of cancer results, any applicable test results, additional cancers, and clinical features. You also have the ability to add in a wide variety of notes, some of which can be displayed on the pedigree, others of which (i.e., "additional notes") that are for provider reference only.
- Clicking on a family member from inside the pedigree will open their [family member page](#) beneath the pedigree.

### 3. Set pedigree layout options and print the pedigree.

- Lay out the pedigree using these tools:
  - **Style** – Use the dropdown to toggle between *Primary Colors*, *Black*, and *White*.
  - **Size** – Set the margins for printing the family tree. Choose *Full Width* or *Landscape*. You can also select *Internet Explorer (IE) Copy/Paste* to ensure that the pedigree prints properly when using IE.
  - **Legend Location** – Use the dropdown menu to select the position of the legend.
  - **Hide** – Tick the boxes to exclude information from the family tree display.
  - The pedigree has both slide and zoom settings that are compatible with macOS and Windows applications.
- Print the pedigree by clicking **Print this page**.

CancerGene Connect®

Secure | https://demo.cagene.com/#/Provider/en-US/Pedigree/7d330bad-8641-4008-8460-201c27bfdea

INVITAE

**Pedigree**

Note: Click on a family member name below to make changes to their personal information.

PRINT THIS PAGE

Ruth Harris (GGC32704) Oct 26, 1969

Legend:

- Lung Cancer
- Leukemia
- Cervical Cancer
- Breast Cancer
- Pancreatic Cancer
- Ovarian Cancer
- Basal Cell Carcinoma
- Polyps, Unknown

ADD SON ADD DAUGHTER ADD BROTHER ADD SISTER

SAVE

**2**

Name: Mom

Relation: Mother

Still Alive? ☐ Yes ☒ No ☐ Do Not Know

Age Now or At Death (Give your best guess; enter 1 if younger than 1 year-old): 72

Ethnicity: Ashkenazi Jewish

Has she had both ovaries removed? ☐ Yes ☐ No ☐ Do Not Know

Identical Twin ID (Identify identical twins, triplets etc.. by setting them to the same number below):

Bilateral Prophylactic Mastectomy:

Additional Notes: Notes! Notes! Notes! Lots of notes!

Pedigree Notes: BRCA+

☐ For the purposes of risk models, has this person had "bilateral" breast cancer?

Cancer	Age Diagnosed	Pathology	Side	
Breast Cancer	58			REMOVE

Breast Marker Test

ER	PR	HER2-IHC	HER2-FISH	CK14	CK5,6

Pancreatic Cancer: 72

ADD CANCER

ADD CLINICAL FEATURE

Master Notes: This is a sample of notes to be displayed on the pedigree.

Style: Primary Colors

Size (Used for printing): Full Width

Legend Location: Top-Left

Hide

<input type="checkbox"/> Names	<input type="checkbox"/> Patient Name/MRN	<input type="checkbox"/> Ages	<input type="checkbox"/> Cancers
<input checked="" type="checkbox"/> "No Children"	<input type="checkbox"/> Individual cancers and features	<input type="checkbox"/> Legend	<input type="checkbox"/> Patient Arrow
<input type="checkbox"/> Ancestry	<input type="checkbox"/> Notes	<input checked="" type="checkbox"/> Consanguinity	

SAVE



## FAMILY PAGE

The Family page provides a tabular view of the patient's family. Cancers are highlighted with pink font. This page includes several important provider features and functions.

1. Fields for entering paternal and maternal ancestries are at the top of the page.
2. Consanguinity can be noted.
3. If a patient answered "yes" to questions on the [Family Questions page](#) about having had an ovary removed, being an identical twin, or having half relatives, the corresponding checkbox is highlighted. This is a visual cue to follow up with the patient and to personally enter those details on the appropriate [Family page](#) to ensure that the information there is accurate.
4. A highlighted *Ashkenazi Jewish* checkbox indicates that a patient noted their Ashkenazy Jewish ancestry on the [Demographics page](#). Follow up with the patient, then make that notation on their family member page (i.e., Proband page). ***Making the notation in the Proband page will ensure accurate risk modeling.***
5. Navigate into any [Family Member page](#) by clicking the family member's name in the Relation column. Make updates as needed.
6. Track genetic testing by clicking **Gene** in the Gene Testing column. This feature is also accessible from any family member's [Gene Testing page](#) using the sidebar menu.

**Family**

Family History

Paternal Ancestry:  Maternal Ancestry:  Consanguinity: ☒ Yes ☐ No

☐ Ovaries removed? ☒ Identical Twins? ☒ Half Relatives? ☒ Ashkenazi Jewish

Relation	Name	Age	Twin	Status	Ovary Removal	Breast	Ovarian	Other	Gene Testing
Proband	Ruth	48		Alive	Yes (42)	No	Yes (43)	No	Gene
Mother	Mom	N/A		Dead (72)		(58)	No	Pancreatic Cancer (72)	Gene
Maternal Grandmother	MGMother	N/A		Dead (79)			No	Polyps, Unknown (50)	Gene
Maternal Grandfather	MGFather	N/A		Dead (80)		No	No	No	Gene
Father	Dad	78		Alive		No	No	No	Gene
Paternal Grandmother	PGMother	N/A		Dead (52)		Yes (61)	No	No	Gene
Paternal Grandfather	PGFather	N/A		Dead (55)		No	No	Lung Cancer (55)	Gene
Daughter	Susan	26		Alive		No	No	No	Gene
Son	James	23		Alive		No	No	No	Gene
Sister	Sarah	51		Alive		No	No	No	Gene
Sister	Susie	55		Alive		No	No	Cervical Cancer (45)	Gene
Sister	Maggie	46		Alive		No	No	No	Gene

## FAMILY MEMBER PAGE

Every family member entered into the platform will have an individual Family Member page. This page allows the provider to add or edit information about that family member.

For each family member, you can provide additional information on ethnicity, ovary removal, twinning, half relatives, bilateral prophylactic mastectomies, bilateral breast cancer, pathology results, side indications of cancer results, any applicable test results, additional cancers, and clinical features. You also have the ability to add in a wide variety of notes, some of which can be displayed on the pedigree, others of which (i.e., "additional notes") that are for provider reference only.

You can also navigate to Family Member pages from the [Pedigree page](#).

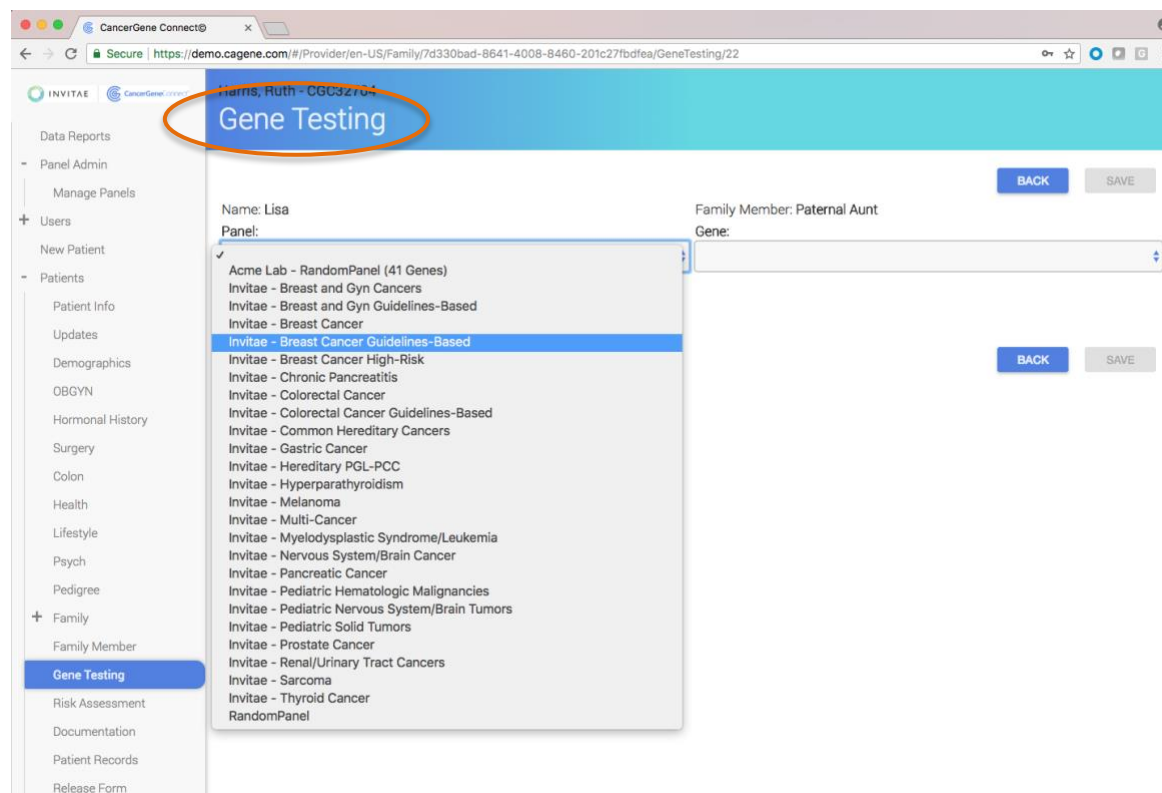
The screenshot shows the 'Family Member' page for a patient named Lisa. The page is part of the CancerGene Connect interface. The left sidebar contains a navigation menu with options like 'Data Reports', 'Panel Admin', 'Users', 'New Patient', 'Patients', 'Patient Info', 'Updates', 'Demographics', 'OB/GYN', 'Hormonal History', 'Surgery', 'Colon', 'Health', 'Lifestyle', 'Psych', 'Pedigree', 'Family', 'Gene Testing', 'Risk Assessment', 'Documentation', 'Patient Records', 'Release Form', 'Patient Side', 'My Account', and 'Logout'. The 'Family' section is expanded, and 'Family Member' is selected. The main content area is titled 'Family Member' and contains the following fields:

- Name:** Lisa
- Relation:** Paternal Aunt
- Still Alive?** Yes (selected), No, Do Not Know
- Age Now or At Death:** 50
- Ethnicity:** (dropdown menu)
- Has she had both ovaries removed?** Yes, No, Do Not Know
- Identical Twin ID:** 1
- Bilateral Prophylactic Mastectomy:** (dropdown menu)
- Is half sibling?** Yes
- Who is the shared parent?** Father
- Additional Notes:** (text area)
- Pedigree Notes:** (text area)
- For the purposes of risk models, has this person had "bilateral" breast cancer?** (checkbox)

Buttons for 'ADD CANCER' and 'ADD CLINICAL FEATURE' are located at the bottom of the form. 'BACK' and 'SAVE' buttons are also present at the top right and bottom right of the form area.

## GENE TESTING PAGE

Every [family member](#) entered into the platform will have an individual Gene Testing page. This page allows the provider to track genetic testing that is ordered through Invitae or another genetic testing lab. You can choose from a prebuilt and pre-populated Invitae curated panel, an individual gene, or (if you ordered genetic testing from another lab) a custom panel that you build yourself.



1. By default, a test is marked as *Pending*. Once results are received, you can choose from the following:
  - **Neg** – Negative or normal result
  - **POS/ABN** – Positive or abnormal result
  - **Var** – Variant result
  - **Pos/Var** – Variant result of unknown significance (VUS)
  - **Uninf** – Uninformative, Lynch-like result
  - **S. Site** – Single-site result

You can enter details of a specific mutation in the freeform box.

2. Remove any tests or panels by clicking **Remove panel**; remove individual genes by clicking **Remove**.

Harris, Ruth - CGC32704

## Gene Testing

BACK SAVE

Name: Lisa Family Member: Paternal Aunt

Panel: Gene:

Invitae - Breast Cancer Guidelines-Based was added.

### Invitae - Breast Cancer Guidelines-Based

	Pending	Neg	Pos/ABN	Var	Pos/Var	Uninf	S. Site	Specific Mutation	REMOVE PANEL
ATM	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
BRCA1	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
Is she/he the first family member to have BRCA1 or BRCA2 tested: <input type="radio"/> Yes <input type="radio"/> No									
BRCA2	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
Is she/he the first family member to have BRCA1 or BRCA2 tested: <input type="radio"/> Yes <input type="radio"/> No									
CDH1	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
CHEK2	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
NBN	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
NF1	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE
PALB2	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="checkbox"/>		REMOVE

The Gene Testing pages are accessible from the [Family page](#) by clicking **Gene** in the Gene Testing column.

### *BUILDING AND EDITING A GENETIC TEST PANEL*

You do not need to order from Invitae to use CancerGene Connect. If you routinely order from other genetic testing labs or have patients whose family members were tested elsewhere, you can build a genetic panel in Panel Admin and it will be added to the dropdown list on the Gene Testing page.

1. To add a new panel, click **Panel Admin** on the sidebar menu. Choose **Manage Panels**.
2. On the Manage Panels page, click **Add new panel**. This takes you to the Build Panel page. Enter a name for your panel, then search and choose from a directory of genes. Click **Save** when your panel is complete.
3. To edit an existing panel, click **Edit**. Search and choose from a directory of genes, or rename the panel. Click **Save** to save your updates.
4. To delete a panel, click **Delete**.

All panels are saved with a timestamp that include the date and time of creation or when it was last updated.

CancerGene Connect®

Secure | <https://demo.cagene.com/#/Provider/en-US/Admin/ManagePanels>

INVITAE | CancerGene Connect®

**1** Manage Panels

Panel Admin

Manage Panels

ADD NEW PANEL **2**

Panel Name	Edit Panel	Last Edited/Updated	Delete Panel
Acme Lab - RandomPanel (41 Genes)	<b>3</b> EDIT	2018-05-02 7:33:47 PM	DELETE
RandomPanel	EDIT	2018-02-16 3:19:04 PM	<b>4</b> DELETE

CancerGene Connect®

Secure | <https://demo.cagene.com/#/Provider/en-US/Admin/PanelBuilder/>

INVITAE | CancerGene Connect®

Build Panel **2**

< PREVIOUS SAVE BUILD NEW PANEL

Panel Name

Name your panel here

Search for a gene

SELECT ALL GENES DESELECT ALL GENES

AIP	AKT1	ALK	APC	ATM	ATR
AXIN2	BAP1	BARD1	BART	BLM	BMPR1A
BRAF	BRCA1	BRCA2	BRIP1	BUB1B	CASR
CBL	CDC73	CDH1	CDK4	CDKN1B	CDKN1C
CDKN2A	CEBPA	CEP57	CFTR	CHEK1	CHEK2
CTNNA1	CTRC	CYLD	DDB2	DICER1	DIS3L2
EGFR	ENG	EPCAM	ERCC2	ERCC3	ERCC4
ERCC5	ERCC6	ERCC8	ERRC4	EXT1	EXT2
EZH2	FAM175A	FAM175A/ABRAXAS	FANCA	FANCB	FANCC
FANCD2	FANCE	FANCF	FANCG	FANCI	FANCL
FANCM	FH	FLCN	GALNT12	GATA2	GEN1

## RISK ASSESSMENT PAGE

The Risk Assessment page manages all the risk modeling that the system performs based on patient responses to the questionnaire. The platform has a built-in set of risk models, described below. As applicable, the risk models run in CancerGene Connect are licensed to Invitae by their intuitional owners; they are updated as needed.

### *BAYES-MENDEL PACKAGE*

1. The **BRCAPRO model** will tell you the probability of a patient carrying a mutation in the BRCA1 or BRCA2 gene. The patient's 5-year and lifetime risks for breast and ovarian cancer are based on the calculated probability that they carry a BRCA1 or BRCA2 gene mutation. The model takes into account family history (affected and unaffected relatives, breast and ovarian cancer history, male breast cancer, and bilateral breast cancer) up to second-degree relatives, breast pathology, and oophorectomy status. The BRCAPRO model also considers ethnicity for each family member, which allows for differing mutation allele frequencies in families of mixed ethnicity. Nonhereditary risk factors and noninvasive breast cancer are not considered in the BRCAPRO model.

The BRCAPRO model uses Bayes' theorem and incorporates various facets of maternal and paternal family histories (including ages of affected and unaffected family members), published data on BRCA1 and BRCA2 mutation frequencies, and cancer penetrance in mutation carriers. The patient's breast and ovarian cancer probabilities are calculated based on the probability that the family has a mutation in either the BRCA1 or BRCA2 gene.

2. **MelaPRO** is a statistical model for assessing the probability that an individual carries a germline deleterious mutation of CDKN2A (p16) based on a family history of single primary and multiple primary melanomas. MelaPRO provides three separate prediction models using penetrances previously estimated from 1) high-risk melanoma families in high-baseline-incidence areas (Australia, the United States, and Sweden); 2) high-risk melanoma families in low-baseline-incidence areas (Europe, excluding Sweden); and 3) all melanoma families from Australia, the United States, and Italy. The model's results provide useful information on an individual's cutaneous melanoma risk, which can impact the individual's clinical decisions and enable researchers to enroll high-risk individuals in genetic studies.
3. **MMRpro** is a statistical model for assessing the probability that an individual carries a germline deleterious mutation of the MLH1, MSH2, and MSH6 MMR genes based on a family history of colorectal and endometrial cancer. The model's results provide useful information on an individual's colon cancer risk that the patient can use before deciding to undergo invasive screenings or expensive genetic testing.
4. **PancPRO** is a statistical model that estimates the probability that an individual carries a mutation in this major susceptibility gene (or gene effect) based on a family history of pancreatic cancer. The model further estimates the probability that an individual will develop pancreatic cancer in the future. The results will provide useful

information about a patient's pancreatic cancer risk before the individual decides to undergo invasive screenings. Researchers will be able to use the results to identify high-risk individuals for enrollment in screenings and genetic studies.

### *GAIL*

The Gail model estimates the probability that an individual develops breast cancer in the future. The Gail model is validated up to a lifetime age of 90 years.

### *CLAUS*

The Claus model estimates the probability that an individual develops breast cancer in the future. The Claus model is validated up to a lifetime age of 79 years.

### *TYRER-CUZICK v7.02*

Also known as the International Breast Cancer Intervention Study (IBIS), the Tyrer-Cuzick model assesses the probability that an individual carries a hereditary breast cancer predisposition gene, then further estimates the probability that an individual develops breast cancer in the future. The Tyrer-Cuzick v7.02 model is validated up to a lifetime age of 85 years.

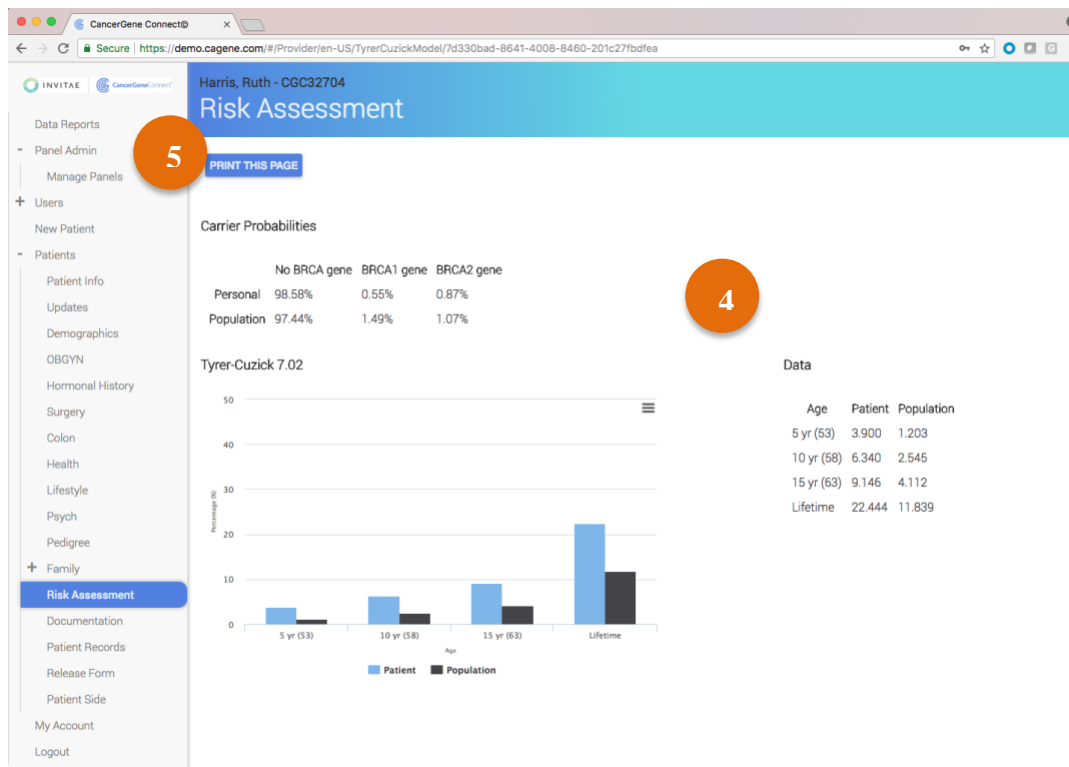
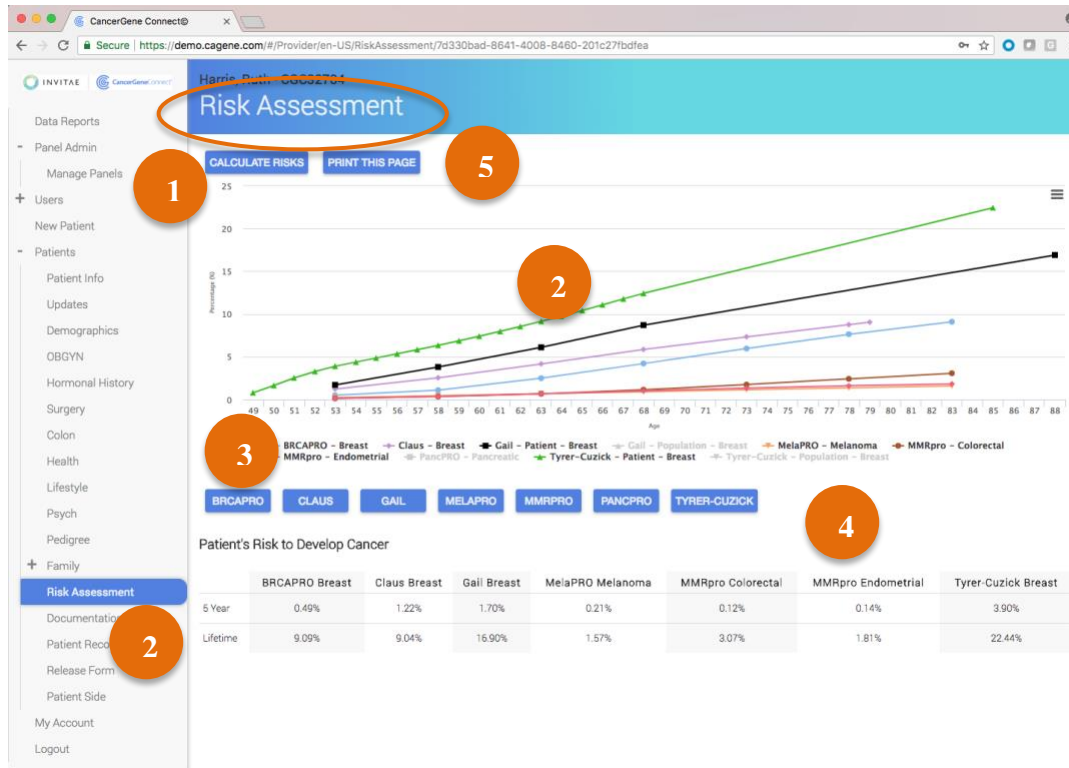
The Risk Assessment page has several important provider features and functions:

1. These models do not calculate automatically. To calculate risk, click the **Calculate risks** button. It may take up to a minute for the models to finish running. **NOTE: After making any relevant update to patient or family information, recalculate to ensure accurate modeling.**
2. Once the risks have been calculated, you will see a combined model line graph displaying every model with a 5-year incremental risk overview. This information is also displayed in a tabular format, beneath the line graph.
3. Toggle the models on and off by clicking each model's name in the legend, beneath the graph.
4. View details of a particular **model** by clicking on the blue model-designated buttons beneath the graph. This will open a separate page for that model with a bar graph and table that highlights 5-year incremental risk overview. If a model includes carrier probabilities, they will be displayed at the top of the individual model page. **NOTE:** To return to the line graph, click **Risk Assessment** in the sidebar menu.

If a model-designated button beneath the graph is grey instead of blue, that particular model is not validated for this particular patient; however, the button is still clickable and will open a separate page for this model. Red warning text noting the particular reason for the model being invalid will display above the graph. A 5-year incremental risk overview will display as if it were ignoring the model-invalidating data.



- Click the **Print this page** button to print the combined model graph and individual model pages.





## DOCUMENTATION PAGE

The Documentation page is designed to save time during general documentation creation. There are a number of templates built into the platform, including a variety of patient and family letters. All templates are customizable and editable within the platform. The templates pull in relevant patient information from the questionnaire and include freeform boxes and dropdowns for adding in patient information. The templates also incorporate macros that are designed to make providing recommendations or informing patients of a positive test result more efficient.

The Documentation page has several important provider features and functions:

1. Save, view, and edit documentation within an individual letter. Download, view/edit, delete, and upload on the main documentation page.
  - All generated documentation is saved with a timestamp that includes the date and time of creation or most recent update.
  - Letters can be downloaded as a Word document with your institution's logo in the header and relevant clinic information (address, phone, and fax) in the footer if these materials are provided at the time of site configuration.
2. The platform includes a number of standard templates for patient letters (specific documentation directed towards your patient) and best test letters (geared toward a patient family member who would provide the “better” genetic test).

The screenshot displays the 'Documentation' page in the CancerGene Connect interface. The page title is 'Harris, Ruth - CGC32704 Documentation'. The main content area is titled 'Generated Documents' and contains a table with the following data:

Document Name	Date/Time (UTC)	Comments	Download	View / Edit	Delete
Risk	2018-05-29 9:28:31 pm		DOWNLOAD	VIEW / EDIT	DELETE
Risk	2018-05-21 9:57:03 pm		DOWNLOAD	VIEW / EDIT	DELETE
BRCA Best Test Letter	2018-05-10 7:15:13 pm		DOWNLOAD	VIEW / EDIT	DELETE
True Negative	2018-05-04 9:38:55 pm		DOWNLOAD	VIEW / EDIT	DELETE
Negative	2018-04-18 4:36:09 pm		DOWNLOAD	VIEW / EDIT	DELETE
Positive	2017-11-21 5:37:08 pm		DOWNLOAD	VIEW / EDIT	DELETE

Below the table, there is a 'Choose File' button (No file chosen), a 'Document Type' dropdown, and an 'UPLOAD' button. The page also features sections for 'Patient Letters' and 'Best Test Letters', each with a list of templates. Callout 1 points to the 'Generated Documents' table, and callout 2 points to the 'Best Test Letters' section.

## RISK LETTER EXAMPLE

To send a risk letter using the template, select **Risk** from the Patient Letters section. The template will be populated with relevant patient information (e.g., personal information, health information, risk assessment). Use the text fields and dropdowns to include additional information in your letter, including recommendations, clinic names, and appointment time. Click **Generate document** to generate and save the letter. The letter will be listed on the [Documentation page](#), in the Generated Documents table, with a timestamp; click **View/edit** to view or edit the letter.

**Harris, Ruth - CGC32704**  
**Documentation**

**Risk**

**Cancer Genetics Risk Assessment**

June 8, 2018

Name: Harris, Ruth  
DOB: October 26, 1969  
MRN: CGC32704

Ruth Harris was evaluated on October 1, 2017 for the risk to develop cancer and/or to have an inherited genetic mutation for cancer. This information is based on her personal and family medical history using risk assessment algorithms. Ruth was referred by Dr. Sonia O'Connor for genetic evaluation due to

**Personal Medical History:**

Ruth is a 48 year old, G2P2A0 female who was (insert recent diagnosis/reason for referral)

Her medical and surgical history is significant for (insert significant info)

Ruth began menstruation at age 12 and is premenopausal. She had her first child at age 22, her last at age 25, and breastfed for 24 months. She has a 3 year history of OCP use. She has not used infertility treatments and has used HRT.

**Family History:**

Ruth's paternal ancestry is Hungarian. Her maternal ancestry is Ashkenazi Jewish. There is consanguinity reported in the family.

Ruth's pertinent family history is as follows:

- Mother was diagnosed with Breast Cancer at age 58, Pancreatic Cancer at age 72
- Maternal Grandmother was diagnosed with Polyps, Unknown at age 60
- Paternal Grandmother was diagnosed with Breast Cancer at age 61
- Paternal Grandfather was diagnosed with Lung Cancer at age 65
- Sister was diagnosed with Cervical Cancer at age 45
- Nephew was diagnosed with Leukemia at age 10
- Niece was diagnosed with Basal Cell Carcinoma at age 22
- Maternal Female Cousin was diagnosed with Ovarian Cancer at age 35

**Risk to Develop Cancer:**

Cancer Type	Patient's Lifetime Risk	Patient's 5 Year Risk	Population Lifetime Risk
Breast	22.44% (Tyrer-Cuzick)	3.90% (Tyrer-Cuzick)	12% women, < 1% men
Ovarian	N/A	N/A	1-2% women
Colon	3.07%	<1%	6% men and women
Pancreatic	2.35%	<1%	1% men and women
Endometrial	1.81%	<1%	3% women

**Risk for a Genetic Mutation for a Hereditary Cancer Syndrome:**

Hereditary Cancer Syndromes	Patient's Risk for a Genetic Mutation	Population Risk for Genetic Mutation
Hereditary Breast and Ovarian Cancer Syndrome (HBOC)	<1%	1/500 or 0.2% (1/40 or 2.5% in Ashkenazi Jewish population)
Hereditary Colon Cancer	<1%	1/500 or 0.2%
Hereditary Pancreatic Cancer	4.89%	1/1000 or 0.01%

**Environmental/Social Risks for Cancer:**

Body Mass Index (BMI)	24.63	Normal
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**NOTE:** The platform includes recommendations for all of Invitae's hereditary cancer panels included on the Gene Testing pages. When a panel is selected, the letter will populate with the genes in the panel, insurance information (if it is provided), a collection date (tied to the date of service entered in the platform), and expected turnaround time for test results. Text that can be used to make general recommendations (for colonoscopy, breast MRIs, and the like) is built in.

## POSITIVE LETTER EXAMPLE

To send a "positive letter" using the template, select **Positive** from the Patient Letters section. The template will be populated with relevant patient information. Use the text fields and dropdowns to include additional information in your letter, including relevant information about a positive test result. Click **Generate document** to generate and save the letter. The letter will be listed on the [Documentation page](#), in the Generated Documents table, with a timestamp; click **View/edit** to view or edit the letter.

The screenshot shows the 'Documentation' page for a patient named Ruth Harris (CGC32704). The page is titled 'Positive' and contains a form for generating a positive letter. The form includes fields for the date (June 8, 2018), patient name (Harris, Ruth), DOB (October 26, 1969), and MRN (CGC32704). It also has dropdowns for 'Ruth Harris underwent' and 'which was performed by'. A text field for 'A mutation was identified in the' gene is followed by a dropdown for 'This is a POSITIVE test result. This means that Ruth has (select gene to populate syndrome)'. Below this is a 'Plan:' section with five empty text boxes. At the bottom, there is a 'Positive Macro' dropdown menu with a list of gene names: None, ALK-Invitae, APC-Invitae, ATM-Invitae, AXIN2-Invitae, BAP1-Invitae, BARD1-Invitae, BLM-Invitae, BMPR1A-Invitae, BRCA1-Invitae (highlighted), BRCA2-Invitae, BRIP1-Invitae, CDC73-Invitae, CDH1-Invitae, CDK4-Invitae, CDKN2A-Invitae, CHEK2-Invitae, DICER1-Invitae, DIS3L2-Invitae, and ENG-Invitae. There are also fields for 'Ruth was informed on', 'Informed On', and a checkbox for 'Please do not concerns. Than'.

**NOTE:** The positive letter template includes positive macros for genes documented in studies on adult cancer. Macros are drawn from NCCN Guidelines and are maintained by Invitae's genetic counselors. When and if guidelines change, we are allowed 30 days to update the platform.

## DOWNLOADED WORD DOCUMENT EXAMPLE

To download a Word document from the Documentation page, click the corresponding **Download** button. A Word version of that document will be generated. If you provided your institution's logo and related clinic information (e.g., address, phone, fax) at the time of site configuration, then these items will also appear in the Word document generated by the system.

You can edit the document within Word, then upload it back into the platform. The timestamped, edited document will be accessible from the [Documentation page](#). **NOTE: You will not be able to open or otherwise view an uploaded document within the platform; you can only download it. View an edited document by downloading it, then opening it from your computer.**

The screenshot shows a Microsoft Word document with the following content:

**Logos:** INVITAE and CancerGeneConnect®

**Title:** Cancer Genetics Risk Assessment

**Date:** May 21, 2018

**Patient Information:**  
Name: Harris, Ruth  
DOB: October 26, 1969  
MRN: CGC32704

**Assessment Summary:**  
Ruth Harris was evaluated on October 1, 2017 for the risk to develop cancer and/or to have an inherited genetic mutation for cancer. This information is based on her personal and family medical history using risk assessment algorithms. Ruth was referred by Dr. Sonia O'Connor for genetic evaluation due to

**Personal Medical History:**  
Ruth is a 48 year old, G2P2A0 female who was . Her medical and surgical history is significant for . Ruth began menstruation at age 12 and is premenopausal. She had her first child at age 22, her last at age 25, and breastfed for 24 months. She has a 3 year history of OCP use. She has not used infertility treatments and has used HRT.

**Family History:**  
Ruth's paternal ancestry is Hungarian. Her maternal ancestry is Ashkenazi Jewish. There is consanguinity reported in the family.  
Ruth's pertinent family history is as follows:

- Mother was diagnosed with Breast Cancer at age 58, Pancreatic Cancer at age 72
- Maternal Grandmother was diagnosed with Polyps, Unknown at age 60
- Paternal Grandmother was diagnosed with Breast Cancer at age 61
- Paternal Grandfather was diagnosed with Lung Cancer at age 65
- Sister was diagnosed with Cervical Cancer at age 45
- Nephew was diagnosed with Leukemia at age 10
- Niece was diagnosed with Basal Cell Carcinoma at age 22
- Maternal Female Cousin was diagnosed with Ovarian Cancer at age 35

**Risk to Develop Cancer:**

Cancer Type	Patient's Lifetime Risk	Patient's 5 Year Risk	Population Lifetime Risk
Breast	22.44% (Tyrer-Cuzick)	3.90% (Tyrer-Cuzick)	12% women, < 1% men

«patient.first\_Name» «patient.last\_Name»  
INVITAE | CancerGeneConnect® | 1701 N. Market Street Ste. 435 Dallas, TX 75202 | www.cagene.com

**Word Interface:** The document is open in Microsoft Word, showing the ribbon (Home, Insert, Design, Layout, References, Mailings, Review, View) and the status bar (Page 1 of 3, 498 Words, English (US), 100% zoom).

## PATIENT RECORDS PAGE

The Patient Records page generates a printable version of the questionnaire. To exclude individual sections, click that section's **Exclude section from printed form** checkbox. Click the **Print this page** button to print the questionnaire.

**Harris, Ruth - CGC32704**

### Patient Records

[PRINT THIS PAGE](#) [SAVE](#)

**Demographics** ☐ Exclude section from printed form

First Name:  Middle Name:  Last Name:

Gender (sex): ☐ Male ☒ Female Birth Date (month day year):

Street Address:

City:  State:  Zip:

Home Phone:  Work Phone:  Mobile Phone:  Email Address:

Were you adopted? ☐ Yes ☒ No Are you Ashkenazi Jewish? ☒ Yes ☐ No Healthcare Coverage:

Are you Black or African American? ☐ Yes ☒ No

Are you Hispanic or Latino? ☐ Yes ☒ No

How do you describe yourself?

How did you find out about us?  Specify the person or organization name:

**OBGYN** ☐ Exclude section from printed form

Have you ever had a period? ☒ Yes ☐ No

## RELEASE FORM PAGE

The Release Form page is an optional page that enables a patient to release information to another member of their medical care team who may want documentation (e.g., medical reports, consultation letters). There is no limit on the number of physicians that can be added. If physicians are included, their names will carbon copy on any documentation generated. This Release Form page in the provider view is a mirror image of the [Release Form page](#) in the patient view.

You can select the referring provider from the list of physicians by checking the applicable **Referring Provider** box.

**NOTE:** This page does not auto-release any information.

The screenshot shows the 'Release Form' page in the CancerGene Connect provider view. The page title is 'Harris, Ruth - CGC32704 Release Form'. The left sidebar contains a menu with options: Data Reports, Panel Admin, Users, New Patient, Patients (expanded), Patient Info, Updates, Demographics, OBGYN, Hormonal History, Surgery, Colon, Health, Lifestyle, Psych, Pedigree, Family, Risk Assessment, Documentation, Patient Records, Release Form (selected), Patient Side, My Account, and Logout. The main content area has a 'SAVE' button at the top right. Below it, a message states: 'To send medical reports to any of your physicians, please fill in their name, address, and phone numbers below.' An orange arrow points to the 'Referring Provider' checkbox, which is checked. The form fields are: Physician Name (Dr. Bob Nussbaum), Address 1 (1400 16th Street), Address 2 (empty), City (San Francisco), State (California), Zip (94103), Phone Number (555-555-5555), and Fax Number (555-555-1234). There are 'DELETE' and 'ADD PHYSICIAN' buttons. A second 'SAVE' button is at the bottom right.

## APPENDIX

TABLE 1: DIRECTORY OF SIDEBAR MENUS, PATIENT VIEW

These are the pages included in the patient view of the questionnaires. Questionnaire pages marked with an asterisk (\*) are not referenced in this user guide.

Questionnaire pages	Expanded questionnaire sections	Questionnaire options			
		Breast health	CancerGene Connect	Pedigree-only cancer	CancerGene Connect risk ready
Instructions		X	X	X	X
Demographics		X	X	X	X
*Appointment Reason		X			
OBGYN		X	X		X
Hormonal history		X	X		X
Surgery		X	X		X
Colon			X		
Health		X	X		
Lifestyle		X	X		X
Family questionnaire	Children	X	X	X	X
	Siblings	X	X	X	X
	Nieces/nephews	X	X	X	X
	Parent/grandparents	X	X	X	X
	Maternal aunts/uncles	X	X	X	X
	Paternal aunts/uncles	X	X	X	X
	Family questions	X	X	X	X
	Maternal cousins	X	X	X	X
	Paternal cousins	X	X	X	X
Family tree		X	X	X	X
Social history		X	X		
Release form		X	X		X
Finish		X	X	X	X
My account		X	X	X	X
Logout		X	X	X	X

TABLE 2: DIRECTORY OF SIDEBAR MENUS, PROVIDER VIEW

These are the pages included in the provider view of the questionnaires. Questionnaire sections marked with an asterisk (\*) are not referenced in this user guide.

Questionnaire pages	Expanded questionnaire sections	Questionnaire options			
		Breast health	CancerGene Connect	Pedigree-only cancer	CancerGene Connect risk ready
Data reports		X	X	X	X
Panel admin	Manage panels	X	X	X	X
	Build panel	X	X	X	X
Users	New user	X	X	X	X
	Logs	X	X	X	X
Patients	Patient info	X	X	X	X
	Updates	X	X		X
	Demographics	X	X	X	X
	*Appointment Reason	X			
	OBGYN	X	X		X
	Hormonal history	X	X		X
	Surgery	X	X		X
	Health	X	X		
	Colon		X		
	Lifestyle	X	X		X
	Psych	X	X		
	Pedigree	X	X	X	X
	Family	X	X	X	X
	Children	X	X	X	X
	Siblings	X	X	X	X
	Nieces/nephews	X	X	X	X
	Parent/grandparents	X	X	X	X
	Maternal aunts/uncles	X	X	X	X
	Paternal aunts/uncles	X	X	X	X
	Maternal cousins	X	X	X	X
	Paternal cousins	X	X	X	X
	Grandchildren	X	X	X	X
	Extended family	X	X	X	X
	Great ancestors	X	X	X	X
	Family member	X	X	X	X
	Gene testing	X	X	X	X
	Risk assessment	X	X		X
	*Management	X			
	Documentation	X	X		X
	Patient records	X	X		X
	Release form	X	X		X
	Patient side	X	X	X	X
My account		X	X	X	X
Logout		X	X	X	X



## SOCIAL HISTORY REFERENCES

The **Social History** and **Psych** pages include four informal psycho-social assessments that include validated measures for depression (CES-D), social support (ISEL), and personal and family cancer history impact (IES-R), as well as a fourth component that gauges cancer worries, which is not validated and does not contain a cutoff.

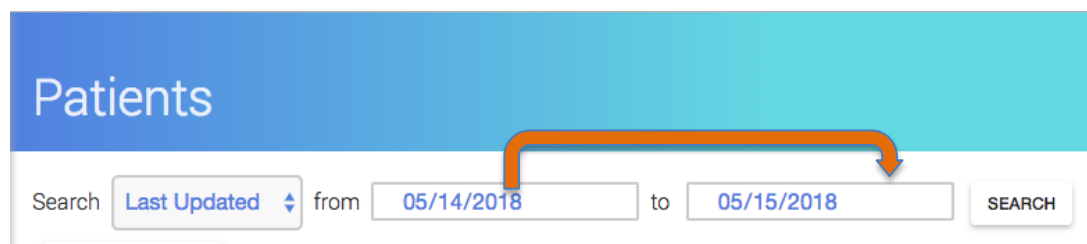
The questions included in CancerGene Connect were developed and reviewed by Dr. Heidi Hamann, a psychologist who is also a trained genetic counselor, and by Dr. Jeff Kendall, a psychologist who, at the time CancerGene Connect was developed, was practicing at University of Texas Southwestern and led the support services division. Explanations of each validated tool are below. NOTE that these assessments do not have a definitive clinical cut-off. If, for example, the patient scored a 9/30 on the Mood (CES-D) assessment, they may still warrant a referral.

1. **Mood (CES-D)** — Ten questions, three points each. Based on a paper from the *Journal of Aging and Health*. For the Common Depression Measure, a "yes" answer and an overall score greater than or equal to 10 indicate that the patient is at risk of depression. (The higher the score, the more severe the depression.) Items are totaled based on response, with the exception of the two "positive" items (questions 4 and 7), which are reverse-scored before summing.
2. **Social Support (ISEL)** — Four questions, three points each. This was adopted from Schonfeld's "[Dimensions of functional social support and psychological symptoms](#)" (1991). A score that is less than or equal to 6 indicates that a patient needs additional social support. The lower the score, the less social support there is. There are no clinical cutoffs, but, in general, scores lower than 6 should be flagged for follow-up, including a referral for support groups or other networks.
3. **Thoughts about Cancer (IES-R)** — Six questions, four points each. Based on Thoressen *et al.*, "[Brief measure of posttraumatic stress reactions: impact of Event Scale-6](#)" (2010). A score that is greater than or equal to 13 indicates that the patient has increased anxiety. The score is summed. There is no particular cutoff, but higher scores (>13) may indicate a need for follow-up or referral.

## DATE AND TIME INFORMATION

The CancerGene Connect platform uses coordinated universal time (UTC). This increases the platform's functionality outside the US and in international time zones.

**Patient edits made to CancerGene Connect after 6 pm CST will require a platform refresh. The search date must also be advanced by one day on the Patients page.**



The screenshot shows the 'Patients' section of the CancerGene Connect interface. It features a search bar with a dropdown menu set to 'Last Updated'. To the right of the dropdown is a date range filter with two input boxes: the first contains '05/14/2018' and the second contains '05/15/2018'. An orange arrow points from the first date box to the second, indicating a one-day advancement. A 'SEARCH' button is located to the right of the date range. The background is a light blue gradient.