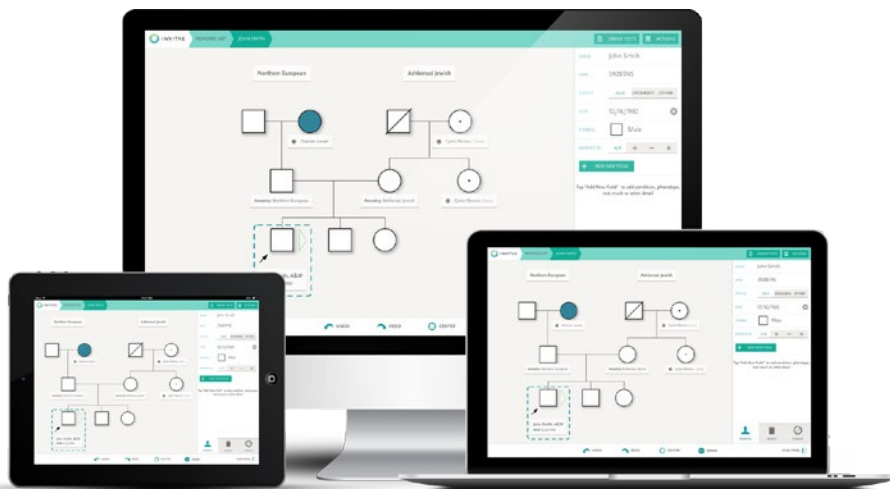




Family History Tool User Guide





Invitae Family History Tool User Guide

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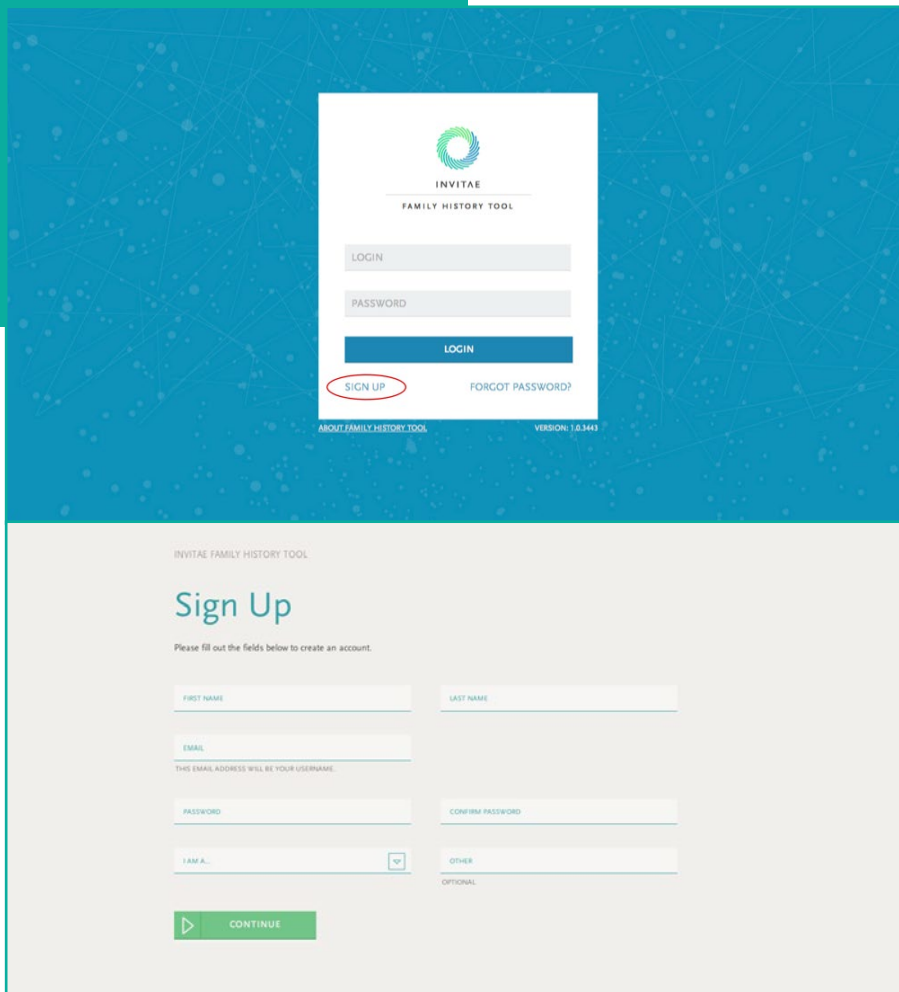
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This document was last updated on 9/23/14.

If you have any questions, please email familyhistory@invitae.com.

Creating an Account

You can create an account by selecting the “Sign Up” option on the Family History Tool welcome screen.



The screenshot shows the Family History Tool welcome screen with a blue background and a network pattern. A white login box is centered, containing the Invitae logo, the text "FAMILY HISTORY TOOL", and fields for "LOGIN" and "PASSWORD". A blue "LOGIN" button is below these fields. A red circle highlights the "SIGN UP" link, and a "FORGOT PASSWORD?" link is to its right. Below the login box, the text "ABOUT FAMILY HISTORY TOOL" and "VERSION: 1.0.3443" are visible.

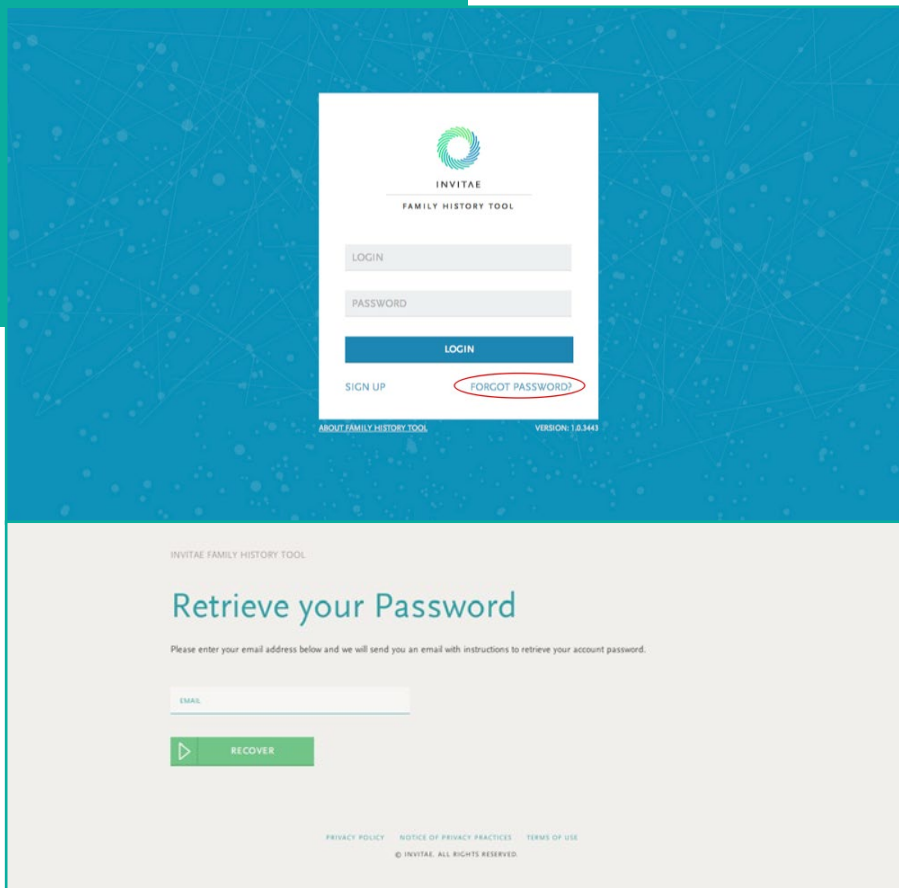
The "Sign Up" form is shown below the login box. It has a title "Sign Up" and a subtitle "Please fill out the fields below to create an account." The form includes the following fields:

- FIRST NAME
- LAST NAME
- EMAIL (with a note: "THIS EMAIL ADDRESS WILL BE YOUR USERNAME.")
- PASSWORD
- CONFIRM PASSWORD
- NAME (with a dropdown menu and a "GO" button)
- OTHER (with a note: "OPTIONAL")

A green "CONTINUE" button with a play icon is at the bottom of the form.

Retrieving an Account Password

You can retrieve an account password by selecting “Forgot Password” on the Family History Tool welcome screen.



The screenshot displays the Invitae Family History Tool interface. The top section is a blue header with a white login form. The form includes the Invitae logo, the text 'INVITAE FAMILY HISTORY TOOL', and input fields for 'LOGIN' and 'PASSWORD'. Below these fields are buttons for 'LOGIN', 'SIGN UP', and 'FORGOT PASSWORD?'. The 'FORGOT PASSWORD?' link is circled in red. Below the login form is a link to 'ABOUT FAMILY HISTORY TOOL' and a version number 'VERSION: 1.0.3443'.

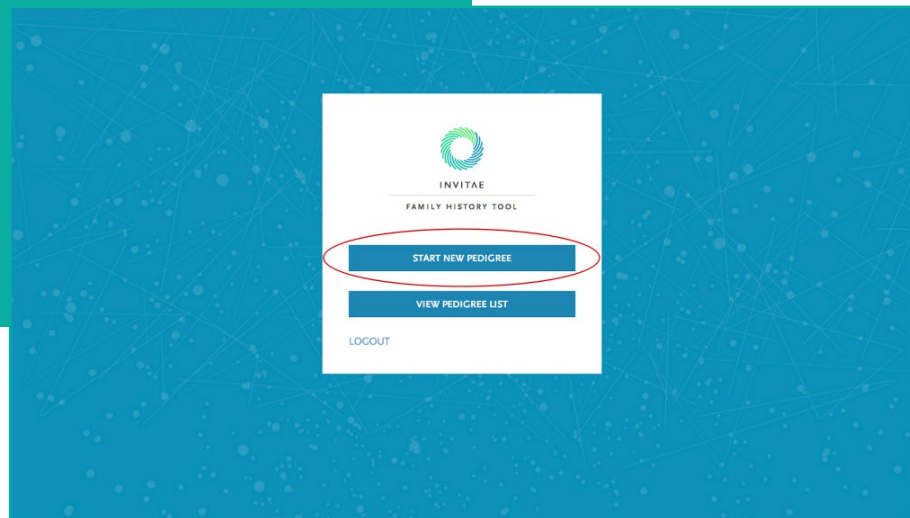
The bottom section is a light gray area with the heading 'Retrieve your Password'. Below the heading is a subheading 'Please enter your email address below and we will send you an email with instructions to retrieve your account password.' and an input field for 'EMAIL'. Below the input field is a green button with a right arrow and the text 'RECOVER'.

At the bottom of the page, there are links for 'PRIVACY POLICY', 'NOTICE OF PRIVACY PRACTICES', and 'TERMS OF USE', followed by the copyright notice '© INVITAE. ALL RIGHTS RESERVED.'

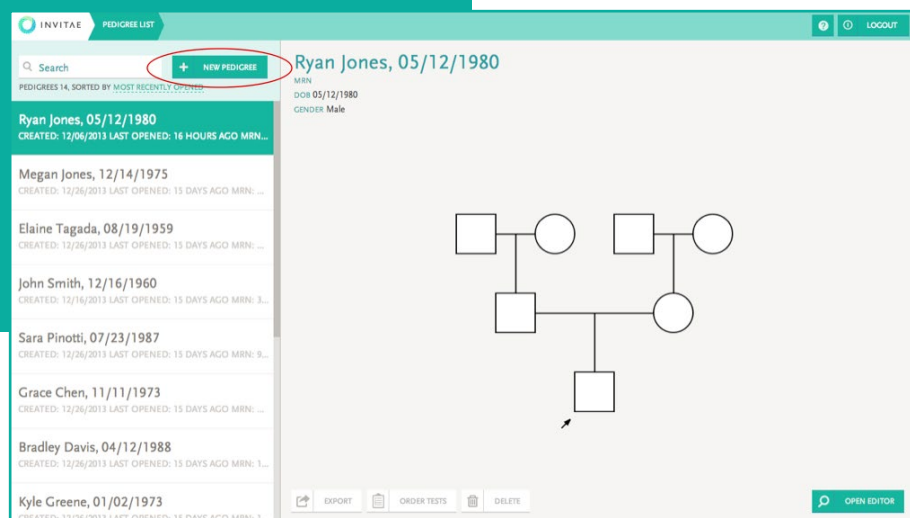
Starting a New Pedigree and Sending a Patient Questionnaire

There are three ways to start a new pedigree:

1. From the Privacy screen that appears after logging in, select “Start New Pedigree”:

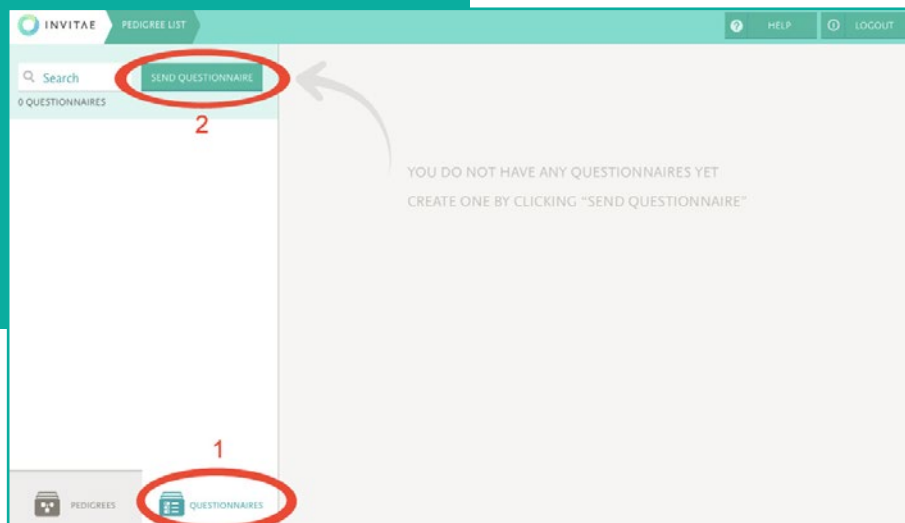


2. From the Pedigree List screen, select “New Pedigree” in the panel on the left:

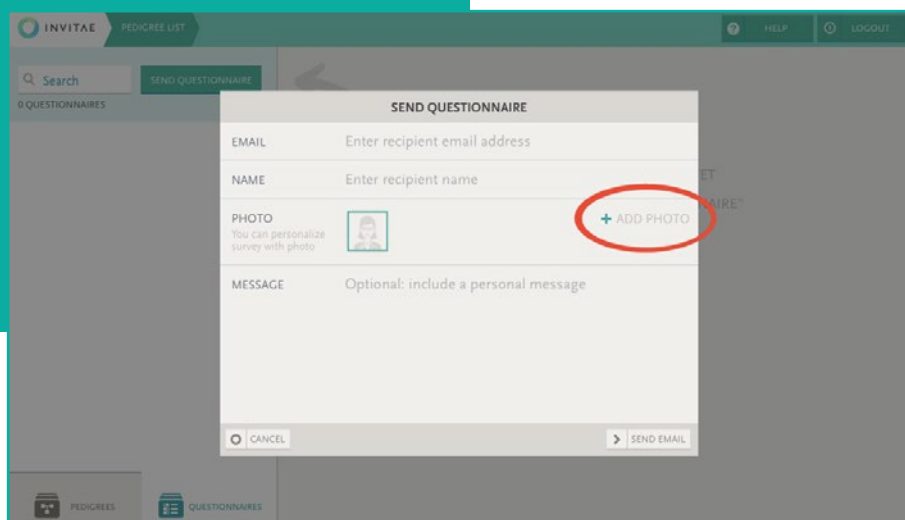


3. You can also start a new pedigree by sending a questionnaire directly to your patient.

From the Pedigree List screen, select “Questionnaires” at the bottom of the left panel, and then select “Send Questionnaire”:



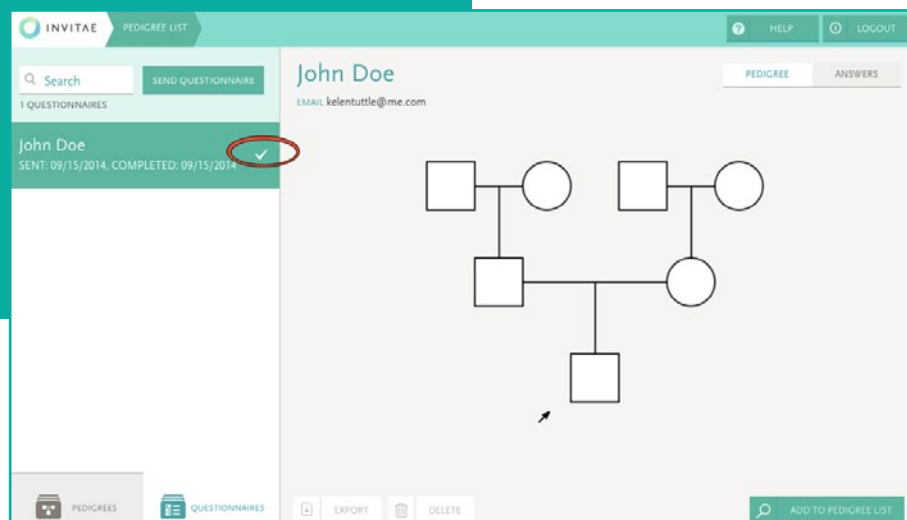
In the “Send Questionnaire” box, you can personalize the questionnaire with a photo of yourself or your clinic’s logo. This photo will appear at the top of your patient’s questionnaire screen. (Note: At this time, the photo option is only available when using the web app.)





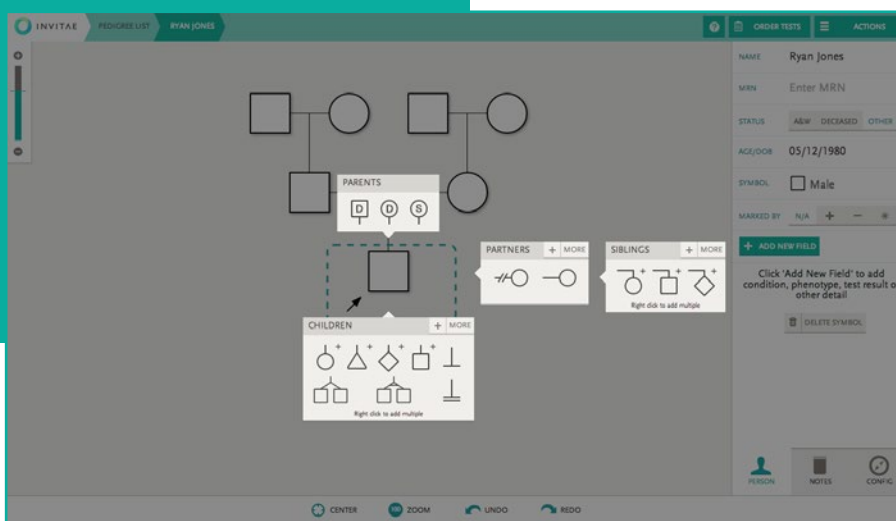
The questionnaire collects the following information: name, gender, DOB, height, weight, racial group, ancestry category, reproductive history, cancer history, previous genetic testing, preventative surgery, and immediate family. Once your patient completes the questionnaire, a digital pedigree will automatically be rendered.

Once your patient has completed the questionnaire, you will receive an email notification and a checkmark will appear next to that questionnaire in the app:



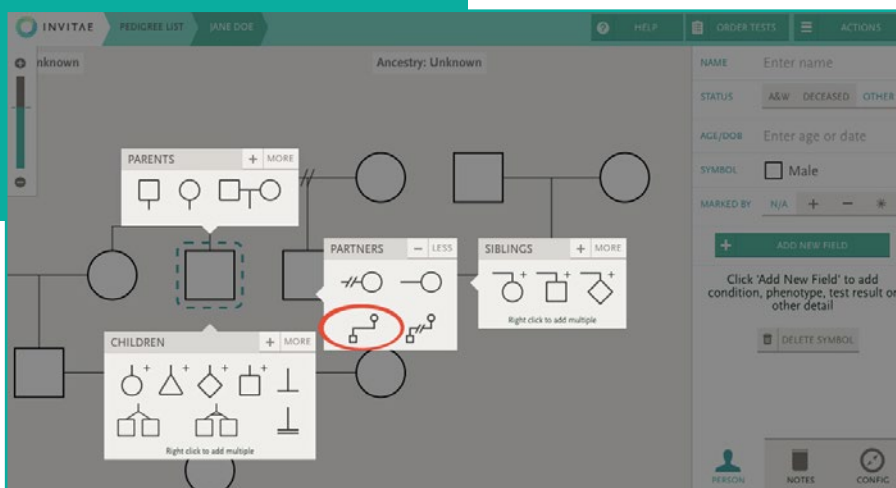
Adding a Family Member to a Pedigree

To add a family member to a pedigree, click on an individual in the pedigree and select the type of relation you would like to add.



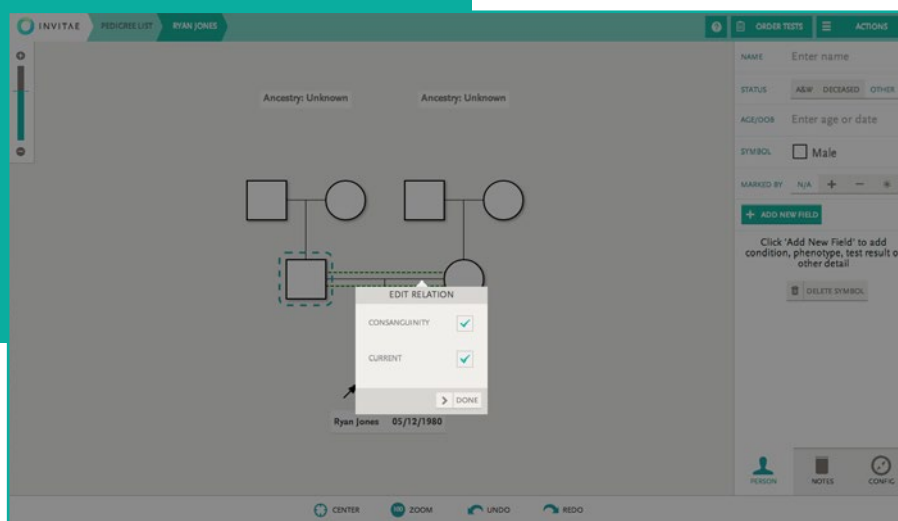
To add multiple people at once, right-click and the ability to add multiple individuals will display. (If using an iPad, tap and hold.)

To add a partner relationship between two individuals already in the pedigree, right click on one of the individuals and select “+More” button in the “Partners” section. Then select the icon circled in red below. The tool will allow you to select any other individual as a partner.

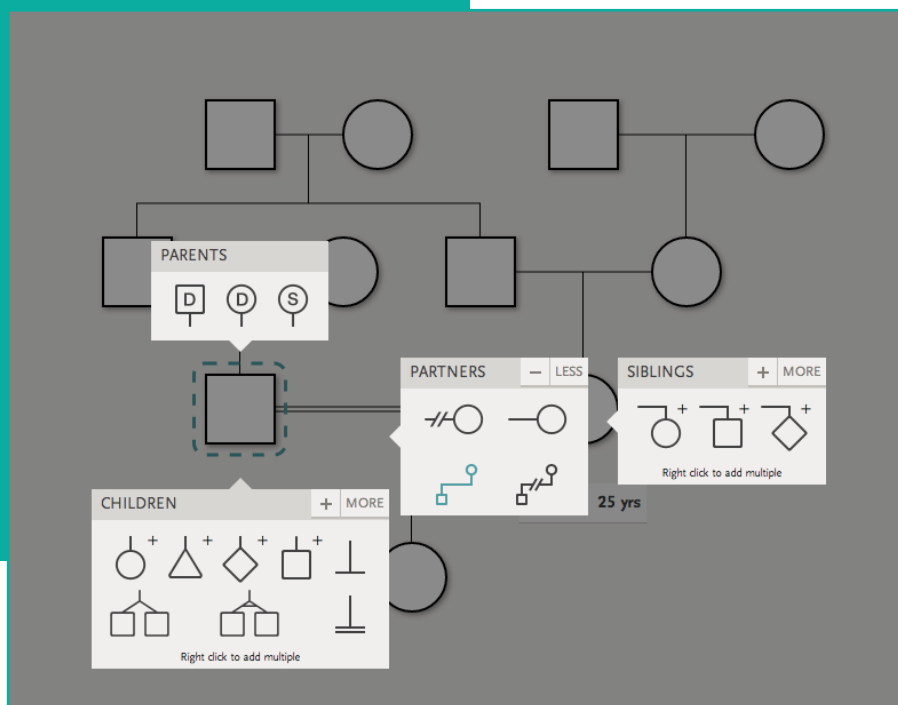


Editing a Relationship

To edit the relationship between individuals, click the line connecting them. From here you can indicate consanguinity and whether they are current or former partners.

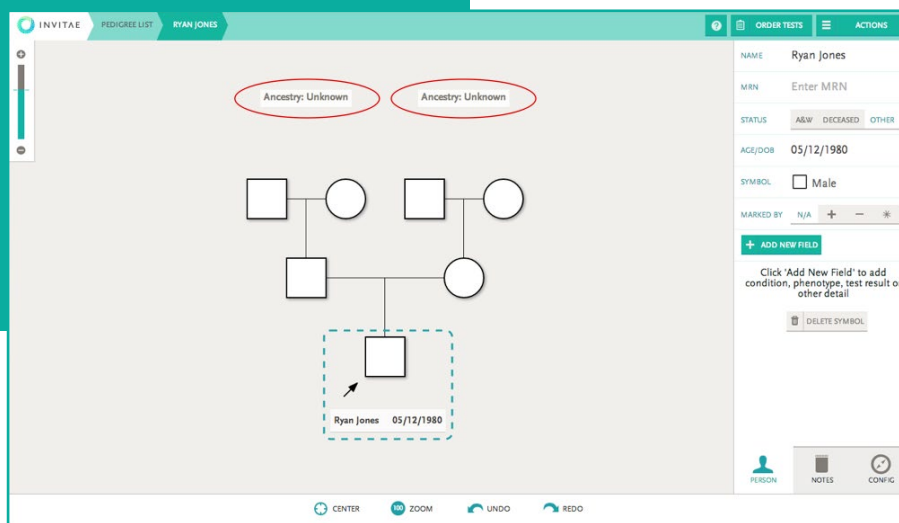


To add more complex consanguineous relationships, click on one of the two individuals on the pedigree screen and select the "+More" button on the "Partners" section. The icon at the bottom with two symbols connected by a line will allow you to select any other individual as a partner.

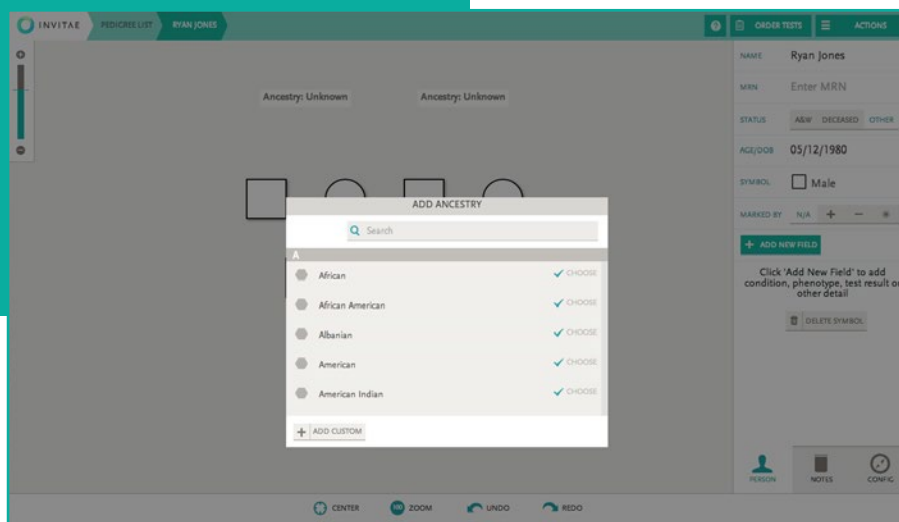


Adding Ancestry

You can add ancestry to the maternal and paternal branches of a family by clicking on the “Ancestry: Unknown” button at the top of the pedigree template.

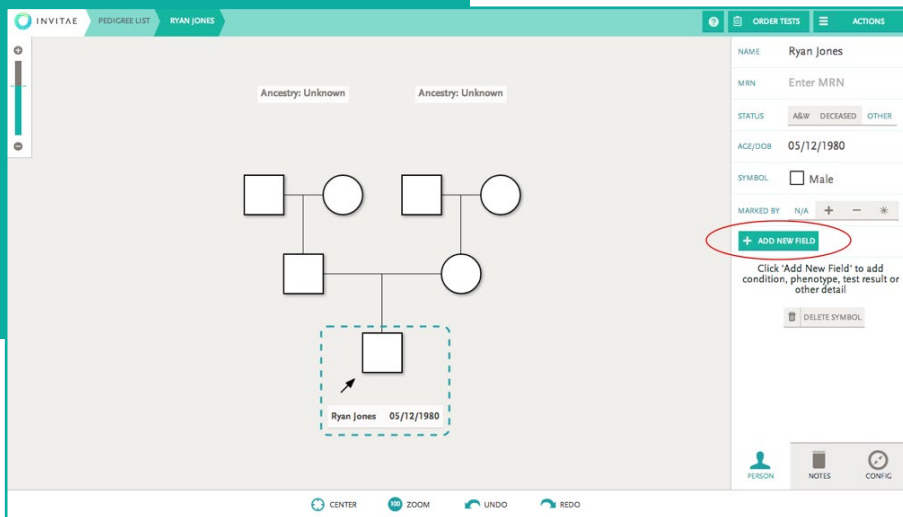


You can also add ancestry to a particular individual by clicking on the individual and selecting the “+ Add New Field” button in the panel on the right.

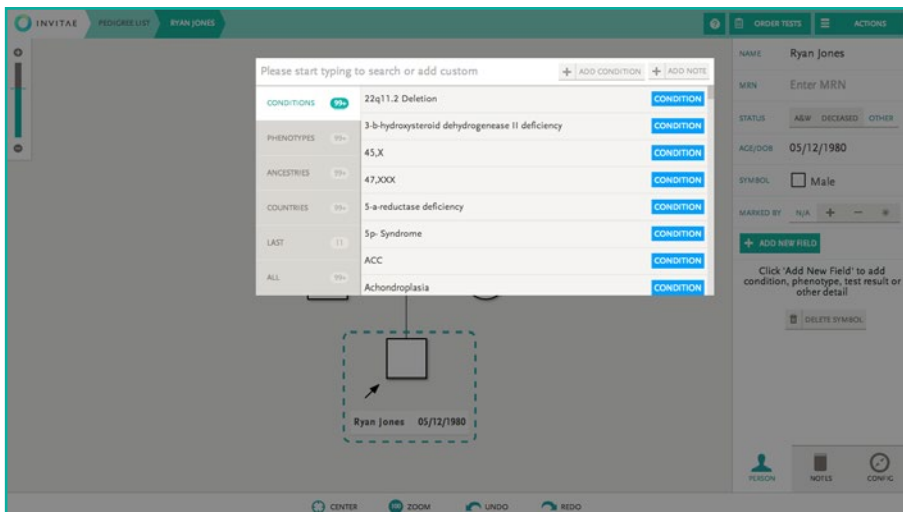


Adding a Condition or Phenotype to a Person

To add a condition, phenotype, ancestry or country to a particular individual, select the individual (indicated by the dotted box surrounding that individual's symbol) and click the "+ Add New Field" button in the panel on the right.



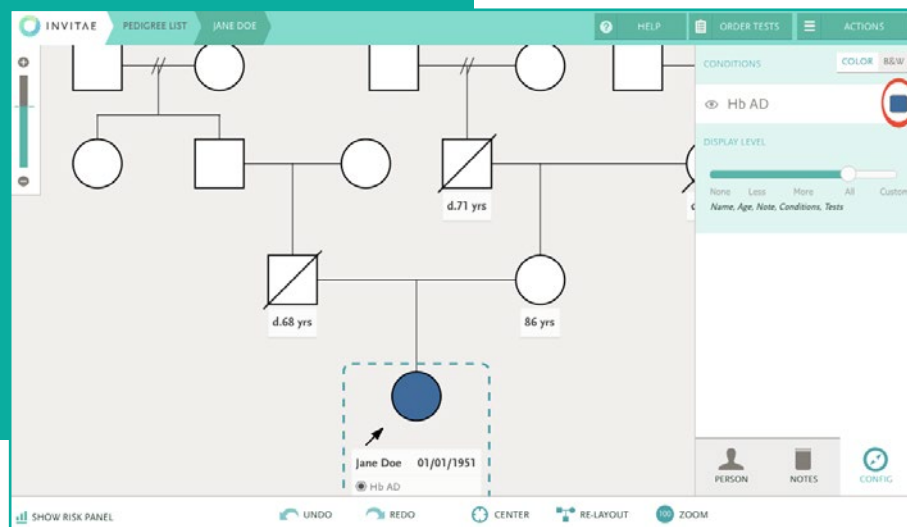
The screenshot shows the INVITAE Family History Tool interface. On the left, a pedigree chart displays a family tree. A dotted box highlights a specific individual, Ryan Jones, with a date of birth of 05/12/1980. On the right, a panel for the selected individual shows various fields: NAME (Ryan Jones), MRN (Enter MRN), STATUS (ABW, DECEASED, OTHER), AGE/DOB (05/12/1980), SYMBOL (Male), and MARKED BY (N/A). The "+ ADD NEW FIELD" button is highlighted with a red circle. Below this button, a tooltip reads: "Click 'Add New Field' to add condition, phenotype, test result or other detail". At the bottom of the panel is a "DELETE SYMBOL" button. The top of the interface shows tabs for "INVITAE", "PEDIGREE LIST", and "RYAN JONES". The bottom of the interface has navigation buttons: CENTER, ZOOM, UNDO, and REDO.



This screenshot shows the same interface as the previous one, but with a search overlay open. The overlay has a search bar with the text "Please start typing to search or add custom" and two buttons: "+ ADD CONDITION" and "+ ADD NOTE". Below the search bar, there are several categories of results: CONDITIONS (22q11.2 Deletion, 3-b-hydroxysteroid dehydrogenase II deficiency, 45,X, 47,XXX, 5-a-reductase deficiency, 5p- Syndrome, ACC, Achondroplasia), PHENOTYPES (45,X), ANCESTRIES (47,XXX), COUNTRIES (5-a-reductase deficiency), LAST (5p- Syndrome), and ALL (ACC, Achondroplasia). Each result has a "CONDITION" button next to it. The background pedigree chart and the right panel are dimmed. The bottom of the interface has navigation buttons: CENTER, ZOOM, UNDO, and REDO.

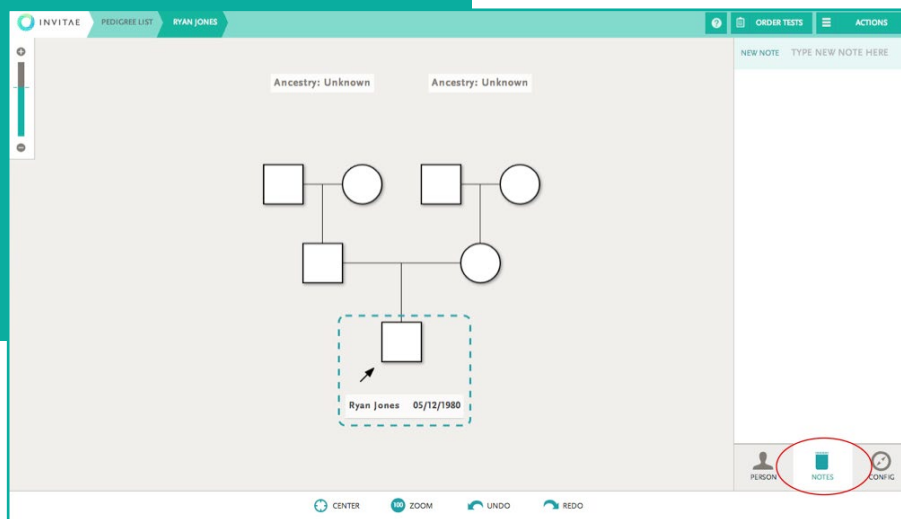


To change the color of individuals with a specific condition, select the “Config” tab in the bottom right corner, and then in the right panel click on the color you would like to change. This same menu also allows you to toggle between color and black & white. You can also change the condition color directly from the “Person” tab by clicking on the condition color box.

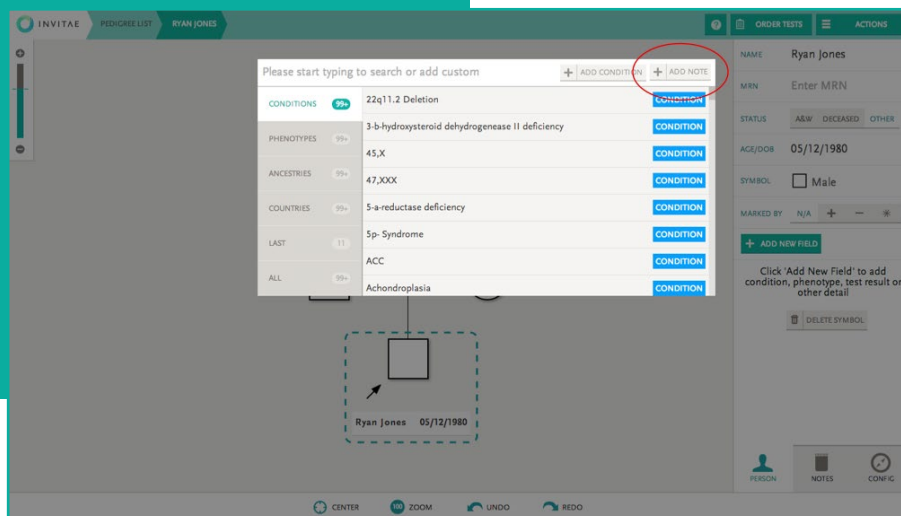


Adding a Note

You can apply a note to the entire pedigree by selecting the “Notes” tab in the panel on the right.

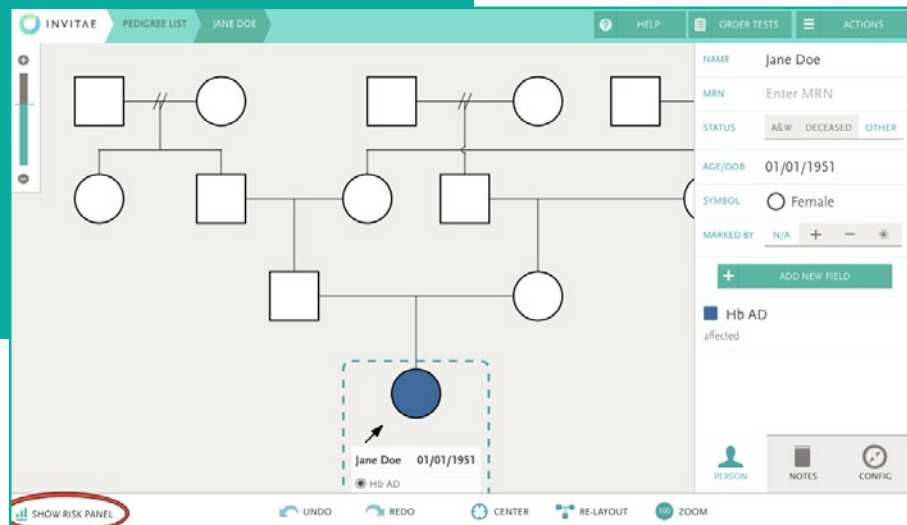


You can also apply a note to a specific individual by selecting the “+ Add New Field” button in the panel on the right and clicking the “+ Add Note” button.

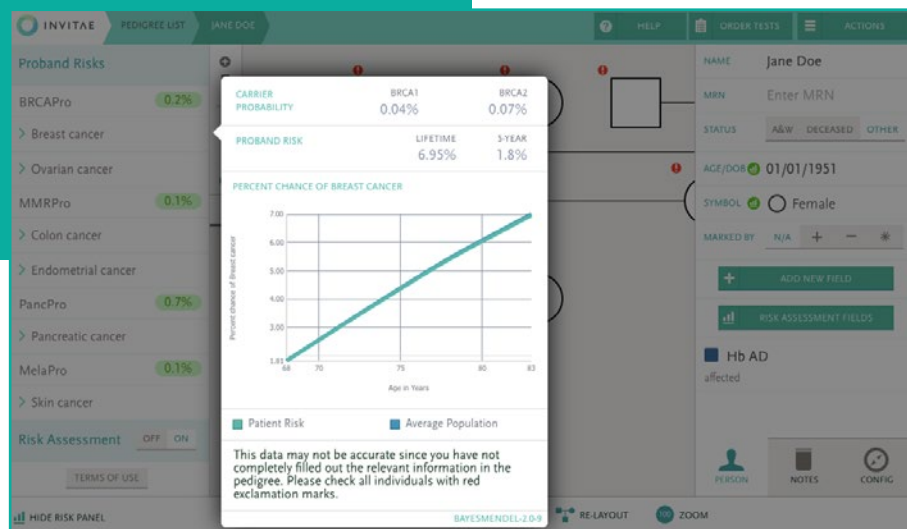


Conducting a Cancer-Focused Risk Assessment

You can conduct a cancer-focused risk assessment by selecting the “Show Risk Panel” button in the lower left corner of a pedigree:



After accepting the terms of use, clicking on each condition in the leftmost panel will display carrier probability (across all genes), lifetime and five-year risk, as well as a comparison to the average population. The risk model and version used to calculate risk are displayed in the bottom right corner of pop-up window. Note that the risks that appear in the panel are only for the proband:



Condition	Carrier Probability	Lifetime Risk	5-Year Risk
BRCA1	0.04%		
BRCA2	0.07%		
Breast cancer (Proband)		6.95%	1.8%

PERCENT CHANCE OF BREAST CANCER

Y-axis: Percent chance of breast cancer (0.00 to 7.00)
X-axis: Age in Years (65 to 85)

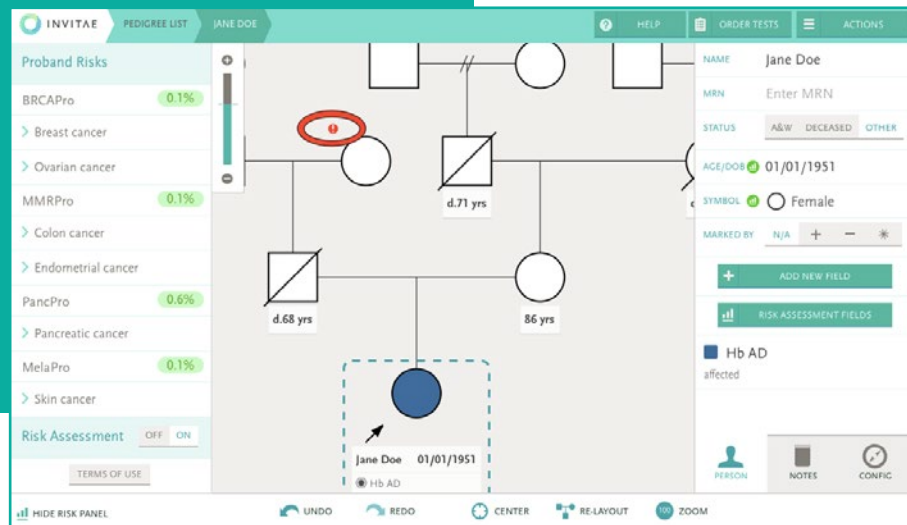
Legend: Patient Risk (red line), Average Population (blue line)

This data may not be accurate since you have not completely filled out the relevant information in the pedigree. Please check all individuals with red exclamation marks.



To improve risk assessment, fill out the pedigree as fully as possible. Anywhere that a red circle with an exclamation mark appears, more information (such as age and gender) would improve the assessment.

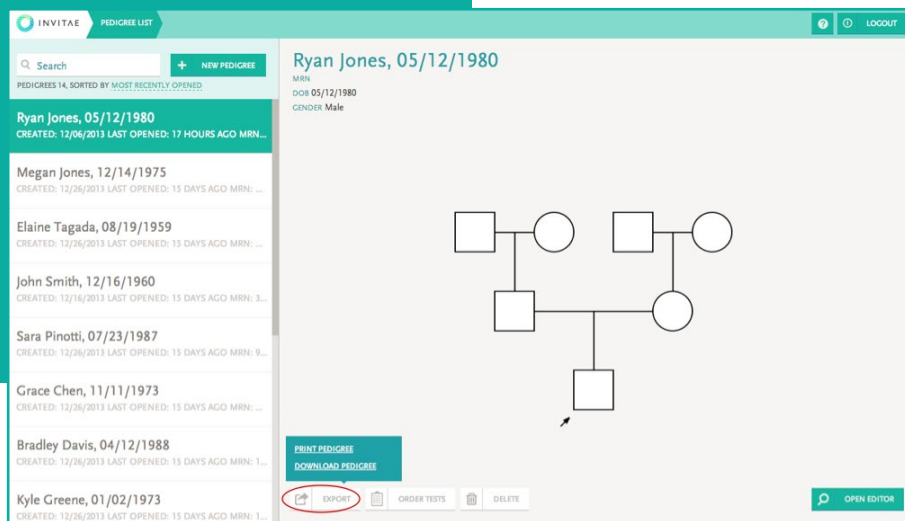
Each time you add additional information to the pedigree, the risk assessment is automatically updated.



Exporting a Pedigree

There are two ways to export a pedigree:

- 1) By clicking the “Export” button on the Pedigree List screen:



INVITAE PEDIGREE LIST

Search + NEW PEDIGREE

PEDIGREES 14, SORTED BY MOST RECENTLY OPENED

Ryan Jones, 05/12/1980
CREATED: 12/06/2013 LAST OPENED: 17 HOURS AGO MRN: ...

Megan Jones, 12/14/1975
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

Elaine Tagada, 08/19/1959
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

John Smith, 12/16/1960
CREATED: 12/16/2013 LAST OPENED: 15 DAYS AGO MRN: 3...

Sara Pinotti, 07/23/1987
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 9...

Grace Chen, 11/11/1973
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

Bradley Davis, 04/12/1988
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 1...

Kyle Greene, 01/02/1973
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 1...

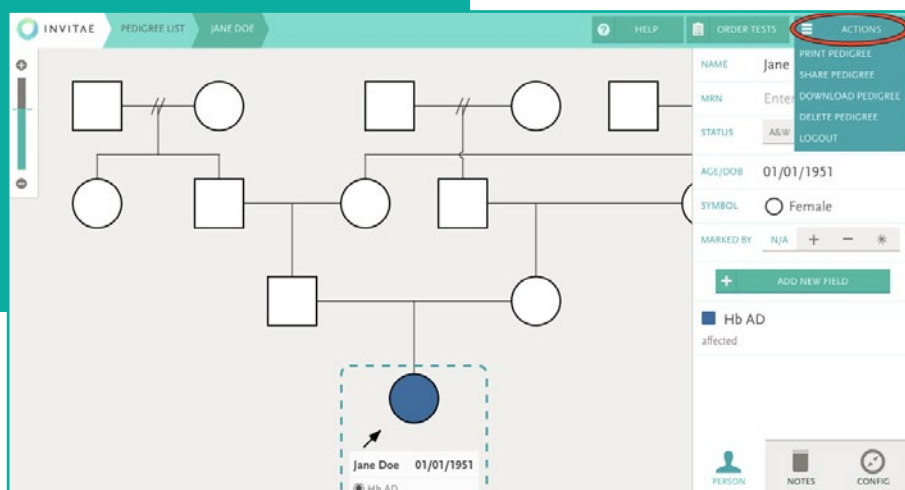
Ryan Jones, 05/12/1980
MRN: 05/12/1980
GENDER: Male

PRINT PEDIGREE
DOWNLOAD PEDIGREE

EXPORT ORDER TESTS DELETE

OPEN EDITOR

- 2) By clicking the “Actions” button from the Pedigree Template screen:



INVITAE PEDIGREE LIST JANE DOE

HELP ORDER TESTS ACTIONS

NAME Jane
MRN Ente
STATUS A&W
AGE/DOB 01/01/1951
SYMBOL Female
MARKED BY N/A + - *

+ ADD NEW FIELD

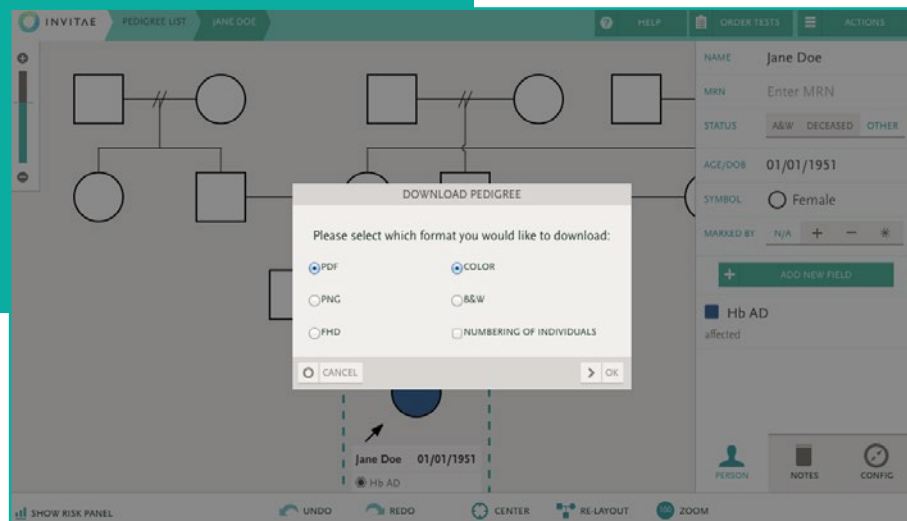
Hb AD
affected

Jane Doe 01/01/1951
Hb AD

PERSON NOTES CONFIG



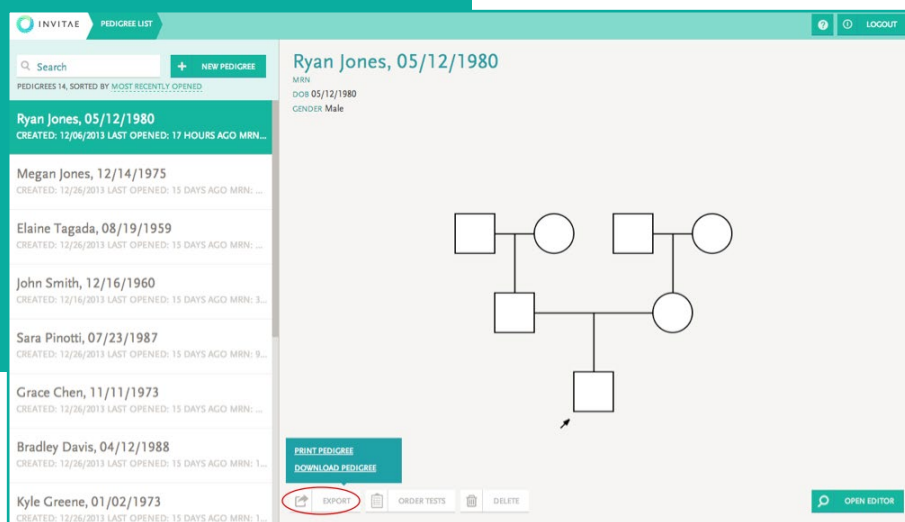
While the iPad version only offers emailing deidentified pedigrees, the web version offers the option of downloading the pedigree as a PDF, PNG or FHD. You can also choose to number generations and individuals within a generation by selecting the “Numbering of Individuals” option:



Printing a Pedigree

There are two ways to print a pedigree:

- 1) By clicking the “Export” button on the Pedigree List screen



INVITAE PEDIGREE LIST

Search + NEW PEDIGREE

PEDIGREES 14, SORTED BY MOST RECENTLY OPENED

Ryan Jones, 05/12/1980
CREATED: 12/06/2013 LAST OPENED: 17 HOURS AGO MRN...

Megan Jones, 12/14/1975
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

Elaine Tagada, 08/19/1959
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

John Smith, 12/16/1960
CREATED: 12/16/2013 LAST OPENED: 15 DAYS AGO MRN: 3...

Sara Pinotti, 07/23/1987
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 9...

Grace Chen, 11/11/1973
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

Bradley Davis, 04/12/1988
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 1...

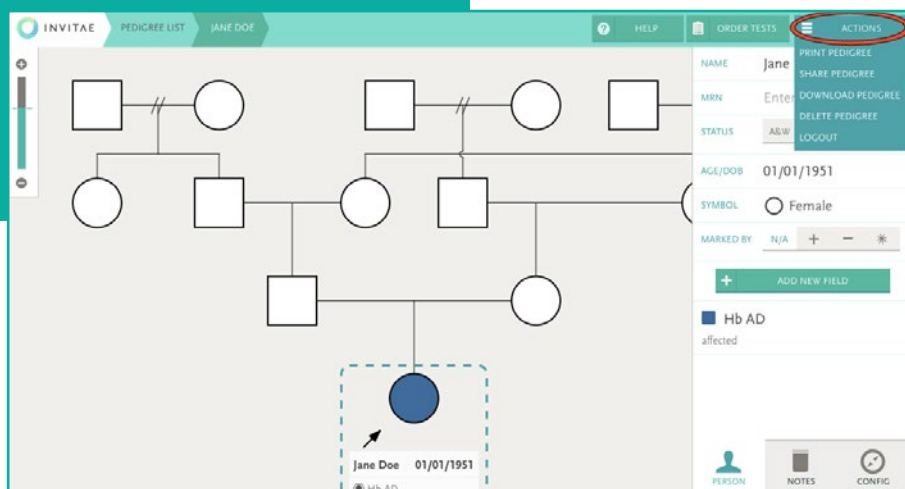
Kyle Greene, 01/02/1973
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 1...

Ryan Jones, 05/12/1980
MRN
DOB 05/12/1980
GENDER Male

PRINT PEDIGREE
DOWNLOAD PEDIGREE
EXPORT ORDER TESTS DELETE

OPEN EDITOR

- 2) By clicking the “Actions” button from the Pedigree Template screen:



INVITAE PEDIGREE LIST JANE DOE

HELP ORDER TESTS ACTIONS

NAME Jane
MRN Enter
STATUS A&W
AGE/DOB 01/01/1951
SYMBOL Female
MARKED BY N/A + - *

+ ADD NEW FIELD

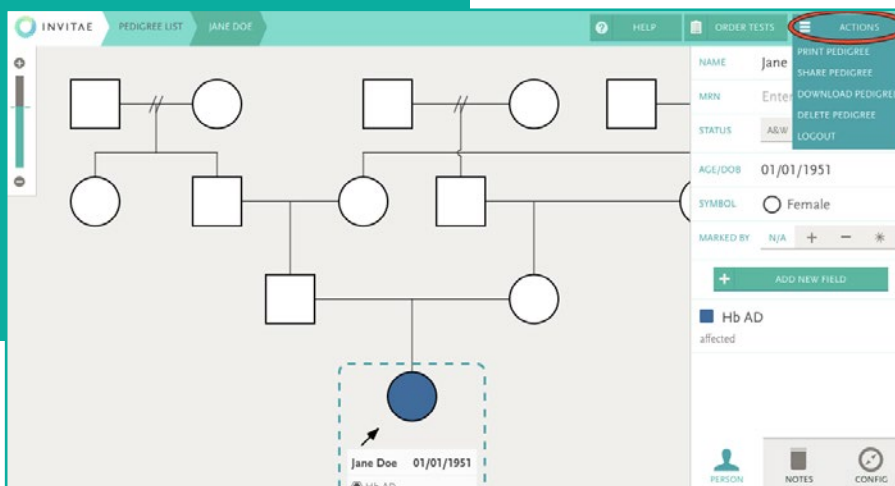
Hb AD
affected

PERSON NOTES CONFIG

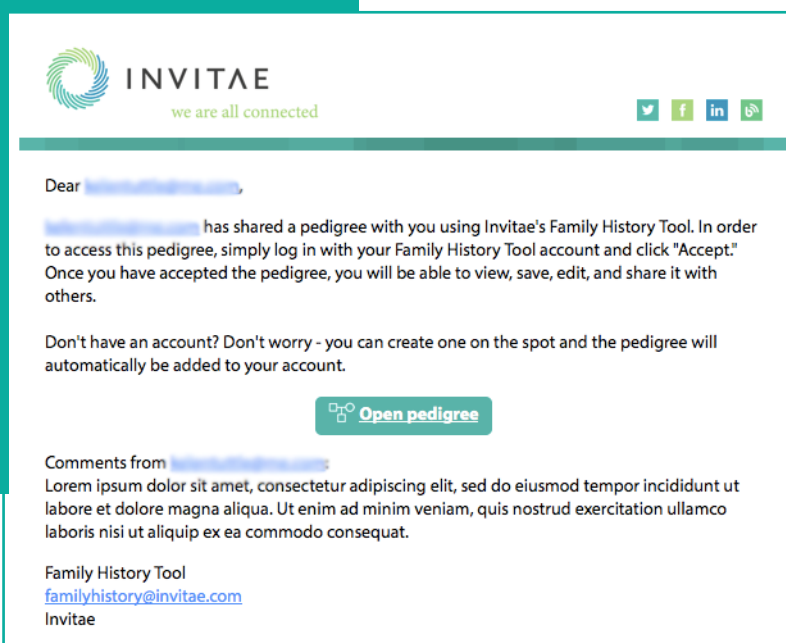
Jane Doe 01/01/1951
Hb AD

Sharing a Pedigree

To share a pedigree, click the “Actions” button from the Pedigree Template screen and select “Share Pedigree.”



You will be asked to enter the recipient’s email address as well as a personal message. This will generate an automated email like the one below. Note that if the recipient already has an Invitae account, you will need to share the pedigree with them at the email address they used to set up their Invitae account for them to receive the pedigree. If they do not yet have an account, they will be prompted to create one using the email address with which you shared the pedigree.



INVITAE
we are all connected

Dear [REDACTED],

[REDACTED] has shared a pedigree with you using Invitae's Family History Tool. In order to access this pedigree, simply log in with your Family History Tool account and click "Accept." Once you have accepted the pedigree, you will be able to view, save, edit, and share it with others.

Don't have an account? Don't worry - you can create one on the spot and the pedigree will automatically be added to your account.

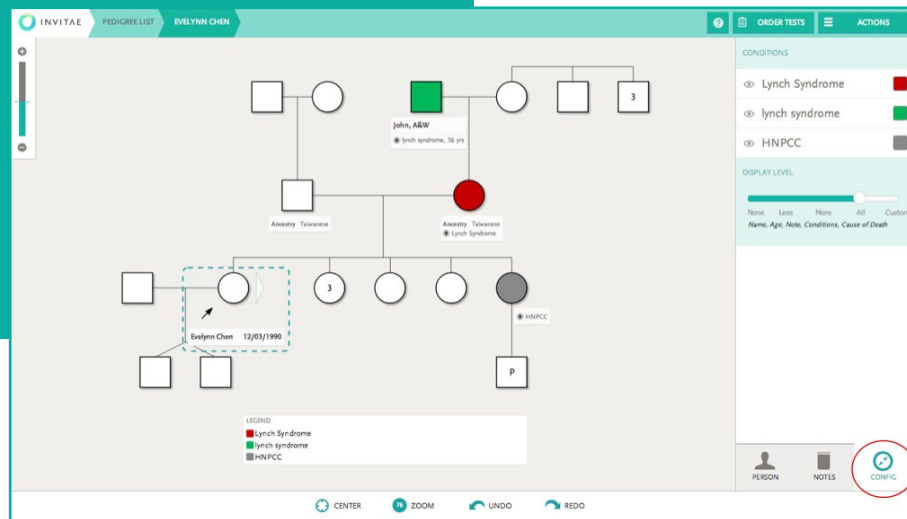
[Open pedigree](#)

Comments from [REDACTED]
Lorem ipsum dolor sit amet, consectetur adipiscing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat.

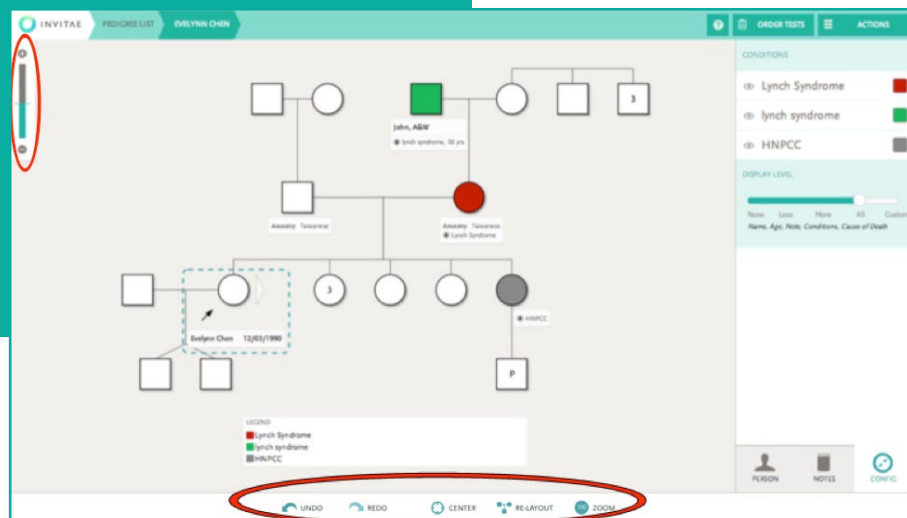
Family History Tool
familyhistory@invitae.com
Invitae

Configuring Display Options

You can modify the amount of information displayed on the Pedigree Template screen by clicking the “Config” tab in the panel on the right.

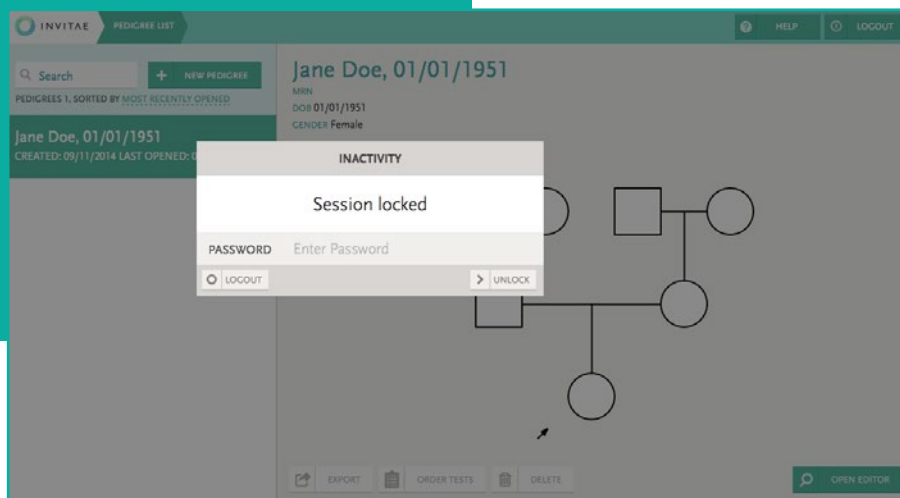


Zoom tools can be found in both the upper left corner and bottom center of the pedigree tool. The “re-layout” option moves individuals into the simplest organization. To override the “re-layout” function, simply click and drag an individual to another location.



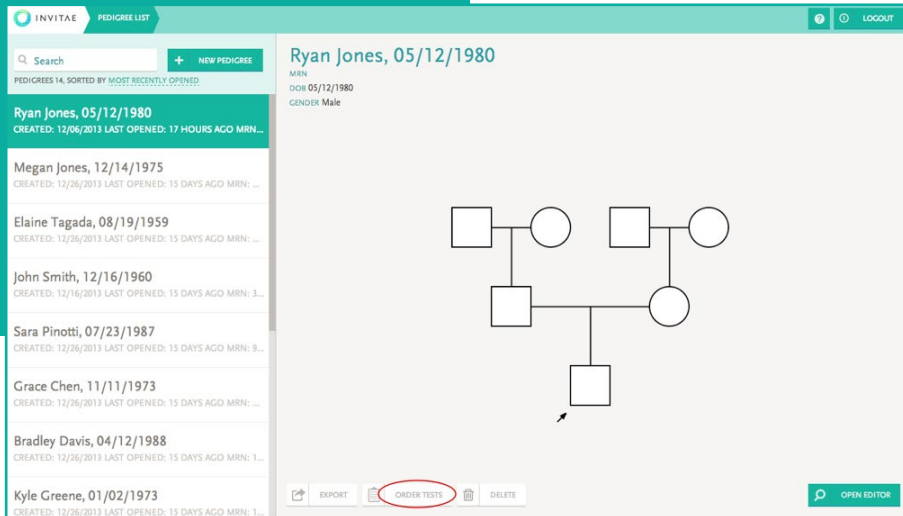
Automatic Lock Screen

To preserve confidentiality, the Family History Tool will automatically lock if inactive for 30 minutes in the web version or 10 minutes in the iPad application. Type in your password to unlock the screen.



Ordering a Test

You can order a test from Invitae by clicking the “Order Tests” button on either the Pedigree List or Pedigree Template screens:



INVITAE PEDIGREE LIST

Search + NEW PEDIGREE

PEDIGREES 14, SORTED BY MOST RECENTLY OPENED

Ryan Jones, 05/12/1980
CREATED: 12/06/2013 LAST OPENED: 17 HOURS AGO MRN: ...

Megan Jones, 12/14/1975
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

Elaine Tagada, 08/19/1959
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

John Smith, 12/16/1960
CREATED: 12/16/2013 LAST OPENED: 15 DAYS AGO MRN: 3...

Sara Pinotti, 07/23/1987
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 9...

Grace Chen, 11/11/1973
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: ...

Bradley Davis, 04/12/1988
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 1...

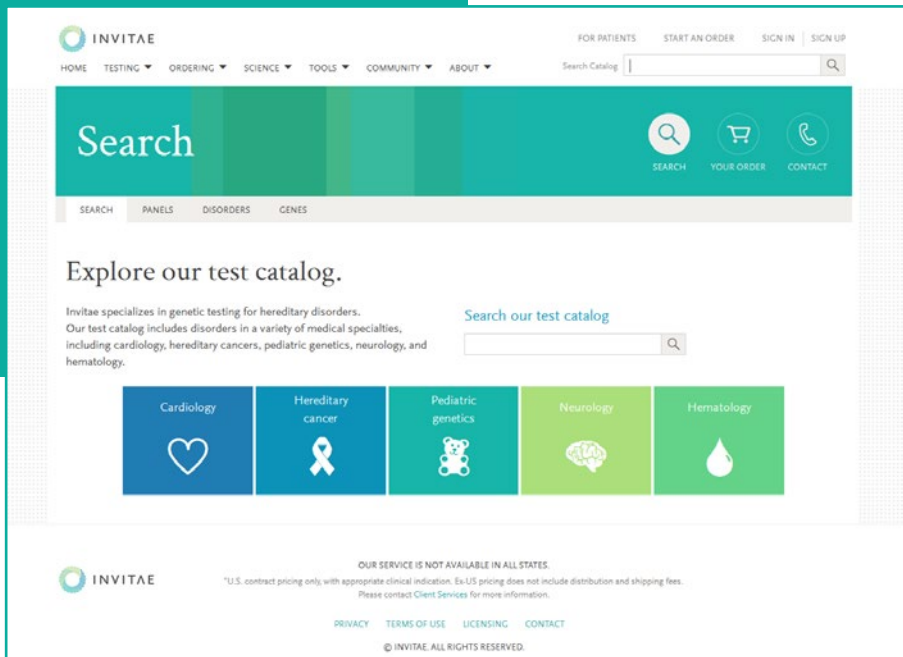
Kyle Greene, 01/02/1973
CREATED: 12/26/2013 LAST OPENED: 15 DAYS AGO MRN: 1...

Ryan Jones, 05/12/1980
MRN
DOB 05/12/1980
GENDER Male

EXPORT ORDER TESTS DELETE

OPEN EDITOR

This will redirect you to the Invitae catalog. You will be automatically logged in and your patient's information will already be entered.



INVITAE

FOR PATIENTS START AN ORDER SIGN IN SIGN UP

HOME TESTING ORDERING SCIENCE TOOLS COMMUNITY ABOUT

Search Catalog

Search

SEARCH PANELS DISORDERS GENES

Explore our test catalog.

Invitae specializes in genetic testing for hereditary disorders. Our test catalog includes disorders in a variety of medical specialties, including cardiology, hereditary cancers, pediatric genetics, neurology, and hematology.

Search our test catalog

Cardiology Hereditary cancer Pediatric genetics Neurology Hematology

OUR SERVICE IS NOT AVAILABLE IN ALL STATES.
*U.S. contract pricing only, with appropriate clinical indication. Ex-US pricing does not include distribution and shipping fees.
Please contact Client Services for more information.

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