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# GeneLeaf Documentation

*Release 1*

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**Nov 09, 2020**



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## Quick Start Guides:

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Pedigree diagrams are a standard visual format for family histories. They enable the representation of genetic information in a compact format which is universally understandable. GeneLeaf is a lightweight online application which enables users to quickly capture create and edit pedigree diagrams through an intuitive user interface, which supports rapid entry of information in a flexible format.

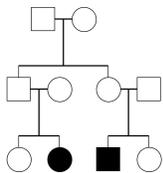
This site contains reference information on the application, including quick start guides designed to get you up and running as well as an in-depth manual to provide information on advanced topics.



GeneLeaf is a visual editor for genomics, allowing users to quickly and intuitively add and update information on family relationships in the form of Pedigree or Genogram diagrams. GeneLeaf is a SaaS application, which users can access securely via the web, but supports offline storage of pedigree files in a proprietary format (.glf) as well as interoperability with other common pedigree file formats as well as common image formats (.svg, .png and .svg).

GeneLeaf uses standard conventions to represent the family relationships included in Pedigrees, as in the example below. Pedigrees are visual representations of family relationships. These diagrams follow a specific set of formatting conventions to represent family information. These include the following key components:

- Squares/Circles: used to represent males & females
- Affected: color (including black) is used to indicate that an individual is affected by a condition.
- Spousal relationships: represented by horizontal bars between individuals
- Sibling relationships: represented by horizontal bars above individuals
- Generation relationships (parents/children): represented by vertical bars



In the figure above, two cousins members (1 male and 1 female) are affected.

## 1.1 Input Modes

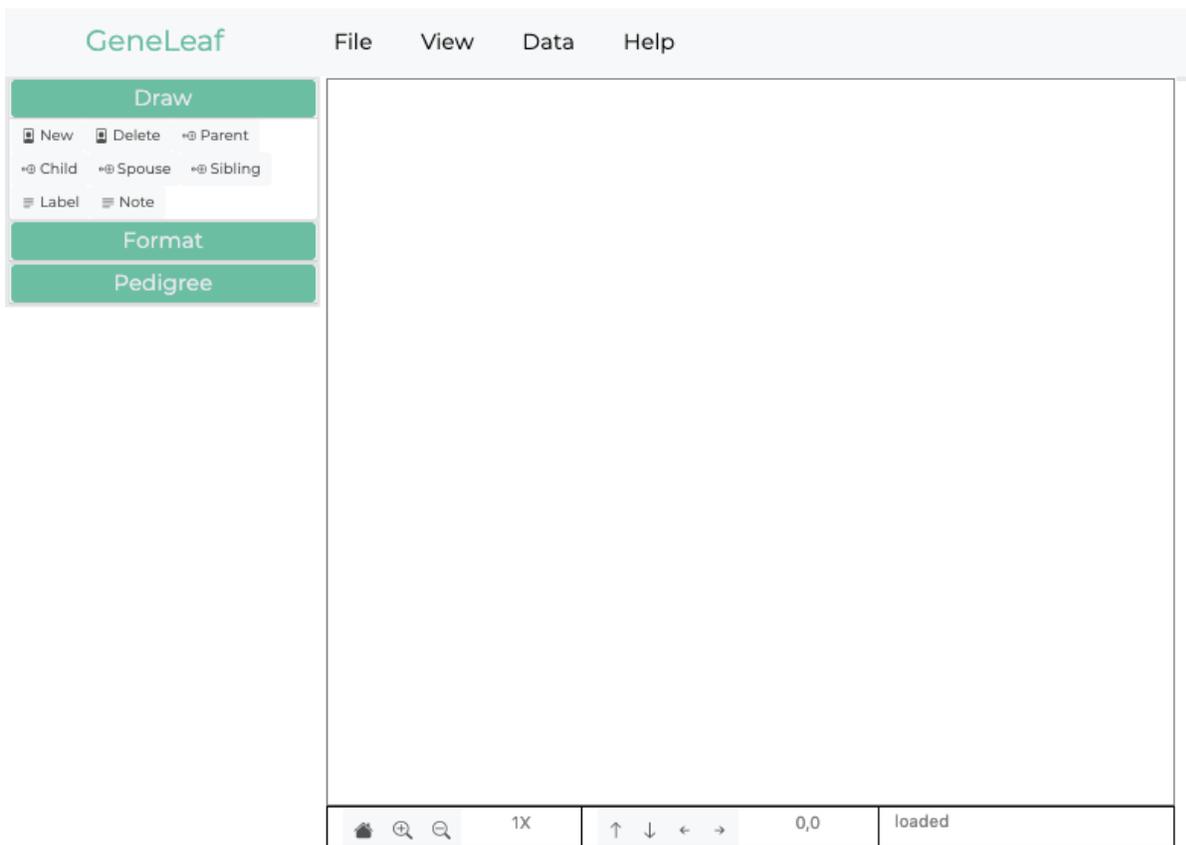
GeneLeaf is designed for ease of use, including intuitive data input modes, including:

- **Drawing Pallet** - click on any of the icons in the drawing pallet and then click on the drawing area to draw that item.

- **Menus** - use menus to navigate to the task you wish to execute.
- **Keyboard shortcuts** - keyboard shortcuts enable you to quickly execute standard tasks.
- **Triggers** - Each pedigree symbol can be selected by clicking on it. This will cause four triggers to be displayed. Clicking ea
  - North - add parent
  - South - add child
  - East - add spouse
  - West - add sibling

## 1.2 Your First Pedigree

After following these simple steps, you will have created a simple pedigree as well as having mastered the basic concepts of pedigree drawing with GeneLeaf. Start by opening the [GeneLeaf App](#) . You will be presented with a screen similar to the following:



1. **Create Individual:** To draw your first individual, double click on the drawing canvas. This will create an individual at the spot where you click and select that individual (showing the 4 blue triggers), opening its properties in the dialog on the right. Here, we should note:
  - Double clicking the canvas is one way to create individuals. However, you could also have clicked on the **new** button under the draw panel on the left.

- Selecting an individual will show the 4 blue triggers and display the selection's properties in the panel on the right.

The screenshot shows the GeneLeaf application window. The top menu bar includes 'File', 'View', 'Data', and 'Help'. On the left, there are three main panels: 'Draw' (containing 'New', 'Delete', 'Parent', 'Child', 'Spouse', 'Sibling', 'Label', and 'Note' buttons), 'Format', and 'Pedigree'. The central pedigree view shows a single individual represented by a blue flower icon. The right-hand panel, titled 'Selection:', displays the following information for the selected individual:

- ID: 89B0017CBD22
- RN: (empty field)
- Category: Adult (dropdown menu)
- First Name: (empty field)
- Middle: (empty field)
- Last Name: (empty field)
- DOB: (empty field)
- Sex:  Male  Female  Other
- Narrative: (empty text area)
- Attributes:
  - proband:
  - preliminary:
  - deceased:
  - adopted:
  - Mode:  Basic
  - affected:
  - carrier:

At the bottom of the pedigree view, there is a status bar with icons for home, search, and zoom, along with '1X' zoom level, navigation arrows, '0,0' coordinates, and a 'selected:' label.

2. **Add Spouse:** Let's add a spouse to this individual. The simplest way to do this is to click on the button labelled 'spouse' in the Draw panel on the left. This will add one spouse to this individual
  - Another way to add a spouse is by clicking the **East Trigger**, on the right of that individual.
  - attempting to add additional spouses is not allowed and will not result in any action.
  - The new spouse's sex is automatically set to the opposite of the current spouse. However, this is done for your convenience and may be manually changed.

This screenshot shows the GeneLeaf application after adding a spouse. The 'Draw' panel on the left now has the 'Spouse' button highlighted. The central pedigree view shows a male (square) and a female (flower) connected by a horizontal line. The right-hand 'Selection:' panel now displays information for the selected female individual:

- ID: 46A4ED4E1346
- RN: (empty field)
- Category: Adult (dropdown menu)
- First Name: (empty field)
- Middle: (empty field)
- Last Name: (empty field)
- DOB: (empty field)
- Sex:  Male  Female  Other
- Narrative: (empty text area)
- Attributes:
  - proband:
  - preliminary:
  - deceased:
  - adopted:
  - Mode:  Basic
  - affected:
  - carrier:

Below the 'Attributes' section, a 'Relations:' table is visible:

Name:	Type:
	Spouse

The status bar at the bottom remains the same as in the previous screenshot.

3. **Add Child:** Let's add a child to this couple. The simplest way to do this is to click on either parent (selecting

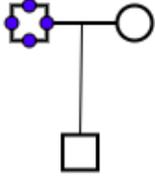
them) and then click on the button labelled ‘child’ in the Draw panel on the left. This will add one child to this couple. Clicking on this again will add additional children.

- Another way to do this is to click on the **South Trigger**, at the bottom of either parent.
- Clicking the trigger additional times will add more children.

The screenshot shows the GeneLeaf software interface. On the left, there are three panels: 'Draw' with buttons for New, Delete, Parent, Child, Spouse, Sibling, Label, and Note; 'Format'; and 'Pedigree'. The central area displays a pedigree diagram with a male and female symbol connected by a horizontal line, and a child symbol below them. On the right, the 'Selection:' panel is open, showing fields for ID (81AA78890D47), RN, Category (Adult), First Name, Middle, Last Name, DOB, Sex (Male selected), and Narrative. Below these are 'Attributes' checkboxes for proband, preliminary, deceased, adopted, affected, and carrier, and a 'Mode' dropdown set to 'Basic'. At the bottom right, a 'Relations:' table shows 'Name:' and 'Type:' with 'Parent' listed twice.

4. **Edit Individual Properties:** Now that we have created our basic family, let’s edit the information of each individual. Start by selecting the first individual you added by clicking on his symbol. This will open the selection panel on the right of the drawing panel. Here you may edit any of the individual’s attributes including name, date of birth, sex, etc.

- **ID:** Each individual is assigned a non-editable ID upon creation. This is displayed in non-editable form.
- **RN(Record Number):** This is a custom record number which may be edited.
- **Category:** This is the general category to which an individual belongs, and includes special symbol categories such as pregnancy, miscarriage, stillbirth, infertility
- **Sex:** Selecting male, female or other results in the corresponding representation of the individual on the pedigree.
- **Attributes:** These include standard attributes commonly represented in pedigree diagrams, including proband and deceased.



Selection:

ID: 89B0017CBD22

RN:

Category: Adult

First Name:

Middle:

Last Name:

DOB:

Sex:  Male  Female  Other

Narrative:

Attributes:

proband:

preliminary:

deceased:

adopted:

Mode:  Basic

affected:

carrier:

Relations:

Name:	Type:
	Spouse
	Child

↓ ← → 0,0 selected:

5. **Add custom data fields:** Let's add a custom data field to this individual. First, select the slider next to **Basic** mode to switch it to **Advanced** mode. This will display the custom data fields below. Since no data fields have been added, click the button labelled **+Add Field** to add a new field. Let's add a field for eyes. Let's set this individual's value to 'blue' (i.e., blue eyes).

Selection:

ID:

RN:

Category:

First Name:

Middle:

Last Name:

DOB:

Sex:  Male  Female  Other

Narrative:

Attributes:

proband:

preliminary:

deceased:

adopted:

Mode:  Advanced

Data Fields:

Field:	Value:
eyes	blue

+Add Field

6. **Add custom formatting rule:** Let's add a custom formatting rule so that when individuals have data field values that match our criteria, they will be displayed to reflect this. \* 6.1 In the left panel, click the tab labelled **Rules**. \* 6.2 Click the button labelled **+Add Formatting Rule**. This will open a dialog box. \* 6.3 Edit the values in the dialog box so that it includes the following:

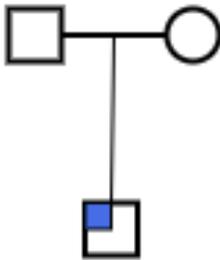
- **Action:** *UL this will color the Upper Left corner of the symbol*
- **Color:** Select the color you want to show.
- **Key:** Select the attribute or data key that you wish to evaluate. Select the field key **eyes** which you just entered.
- **Logic:** Select the operator **Equal**, so that the effect will be applied when the key equals the criteria you specify.
- **Criteria** Enter the value **blue**, so that the effect will be applied when **eyes** equal **blue**
- 6.4 Your rule should look like this:

## Edit Rule

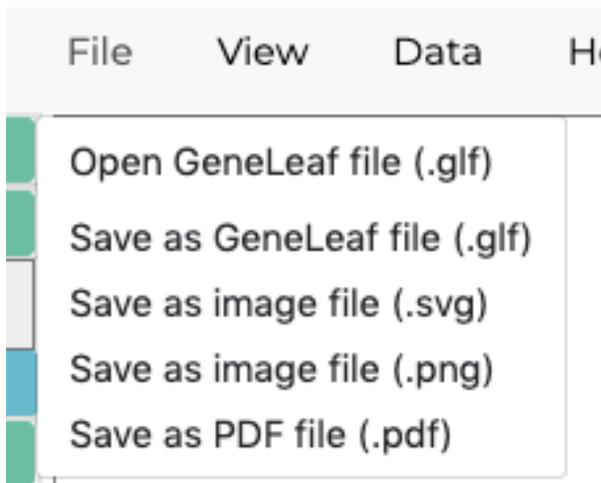
Rule ID:	new
Effect:	Action: UL Color: #2889e9
Key: eyes	eyes
Logic: EQUAL	EQUAL
Criteria: blue	blue

Update Rule Cancel

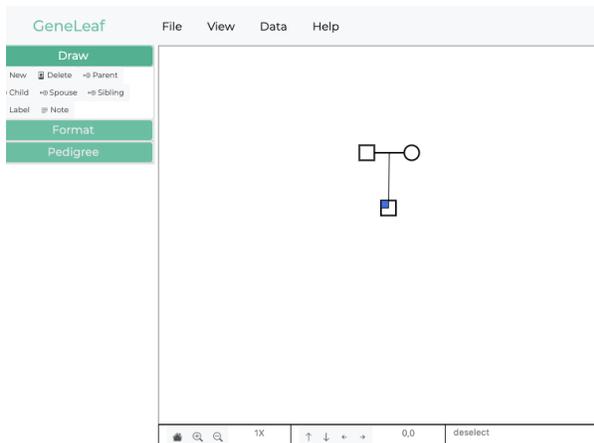
- Click the button **Update Rule** to apply your settings. Your pedigree should look like this:



7. **Save File:** Let's save your pedigree as a GeneLeaf file. Click the file menu and select **Save as GeneLeaf file (.glf)**. Select the location where you wish to save the file on your desktop. \* You may also save the file as an image file (svg, png or pdf).



8. **Open File:** Let's try opening the file you just saved. First, **refresh your browser** to clear the workspace. Next, click the file menu and select **\*\*Open GeneLeaf file (.glf)**. Select the file you saved above. You should see the same pedigree in your browser.

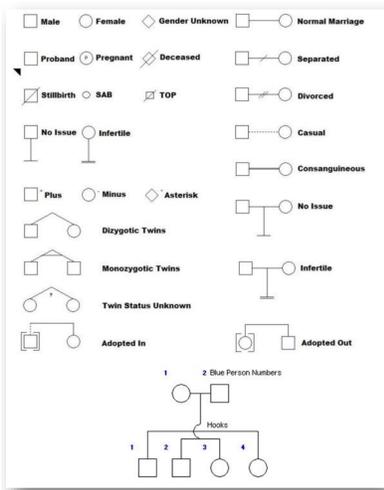


### 1.3 Learning More

- [GeneLeaf Community](#) - join the Geneleaf community.
- [Contact us](#) - email us your questions.

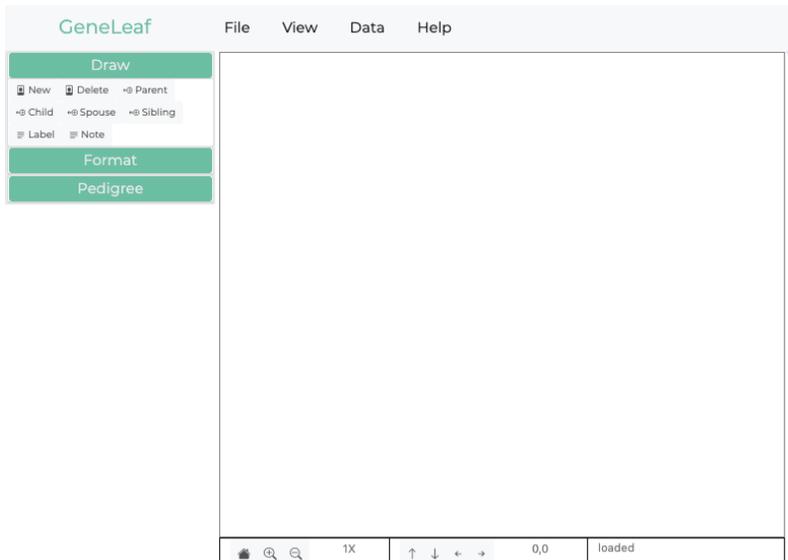
### 1.4 Pedigree Symbol Reference:

GeneLeaf follows widely-supported conventions for Pedigrees, as summarized in the reference sheet below.



### 2.1 a visual editor for genomics

Genetics is the study of family relationships which transfer the information that determines the phenotype of each individual in form of discrete hereditary units called genes. The medium in which most Geneticists typically work is the pedigree diagram or genogram. In these diagrams, the relationships between individuals are represented through a simplified set of symbols, related through lines. Gene Leaf enables users to quickly capture information in this form through an intuitive user interface, which supports rapid entry of information. Gene Leaf allows users to work at their own pace and in their own way. A simplified icon set allows users to add information with simple clicks.



## 2.2 General Concepts

GeneLeaf was developed with the needs of the end user in mind. It is designed to bring powerful analytics to users while reducing the overall effort required to perform key actions.

### 2.2.1 Intuitive

The interface is designed to be as simple as possible, allowing users to find key functionality rapidly through the design approach summarized by:

**TMTOWTDI** - *There's more than one way to do it*

For most common user actions, we support a basic way of doing it, which is easily described as well as a fast shortcut which can be performed rapidly by advanced users.

### 2.2.2 Data-Driven

Pedigrees have always been a unique form of data representation, in that they convey a large amount of detailed information visually. GeneLeaf supports this approach by supporting pedigrees with a rich, extensible data model which enables users to add and maintain a full family history in a compact data format. The data model for each pedigree controls how it is drawn. Users can determine how each data field is represented on the pedigree diagram through the specification of customized formatting rules.

### 2.2.3 Interoperable

One of the frustrating aspects of drawing and maintaining pedigree drawings is the diverse assortment of alternative software applications available for users, which seem to come in and out of fashion on a fairly regular basis. GeneLeaf aims to be a simple, universally compatible tool which allows users to take their pedigrees from any platform, edit and update them and export them in a format which is easily usable by the platform of his or her choice. If you decide to use our platform at an enterprise level, we are here to support you, but we plan to maintain this application as a free tool for all users on a permanent basis.

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## Keyboard Shortcuts

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GeneLeaf allows users to quickly update family information. Use the keyboard shortcuts below to add information with the press of a single key.

Key	Action
p	Create Parent
s	Create Sibling
c	Create Child
m	Create Spouse
-/+	Zoom
PgUp/PgDn	Pan Vertical
Home/End	Pan Horizontal



### 4.1 Beta Testing

Beta testing is under way. We are looking for knowledgeable users to evaluate this application prior to its release and help us to plan for the support of the most important features. If you are interested, contact us at:

[contact@geneleaf.com](mailto:contact@geneleaf.com)



# CHAPTER 5

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Version 1.0 (Spring 2021)

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## 5.1 First Release

The first public release is planned for spring of 2021. To receive updates contact us at:  
[contact@geneleaf.com](mailto:contact@geneleaf.com)



## CHAPTER 6

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### License

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# CHAPTER 7

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## Indices and tables

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