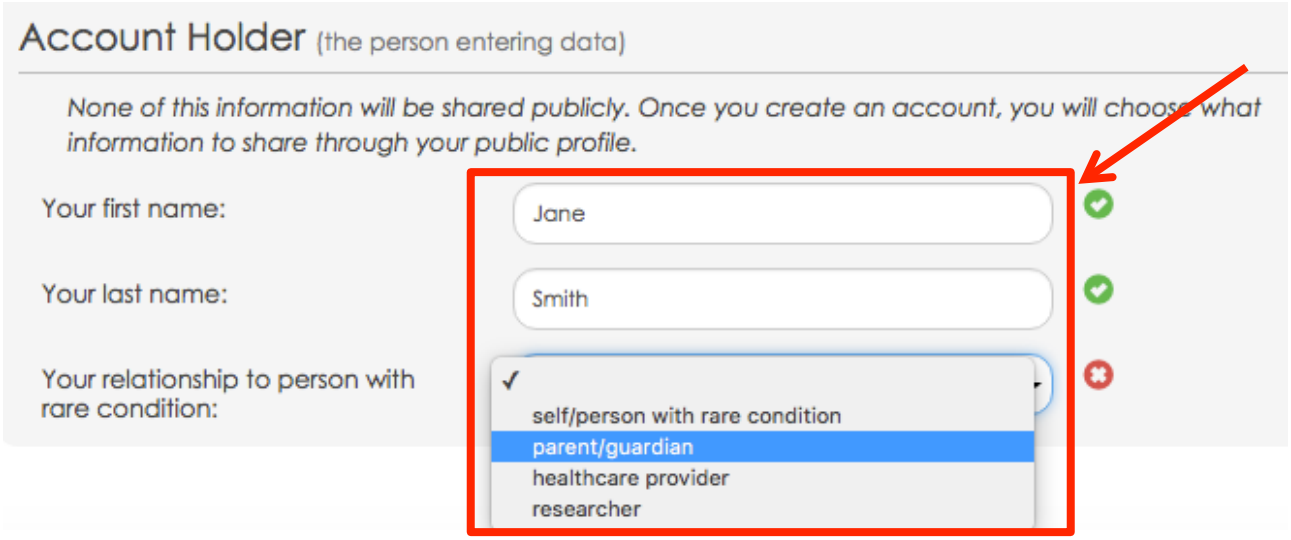
MyGene2 Guide

MyGene2 is a great tool to help you search for and contact other families with individuals who have some of the same characteristics, or mutations in the same gene. By sharing information, you may be able to find others in a similar situation and can gain and offer support. Clinicians and researchers are also able to look at these databases for clues on how to better help their patients.

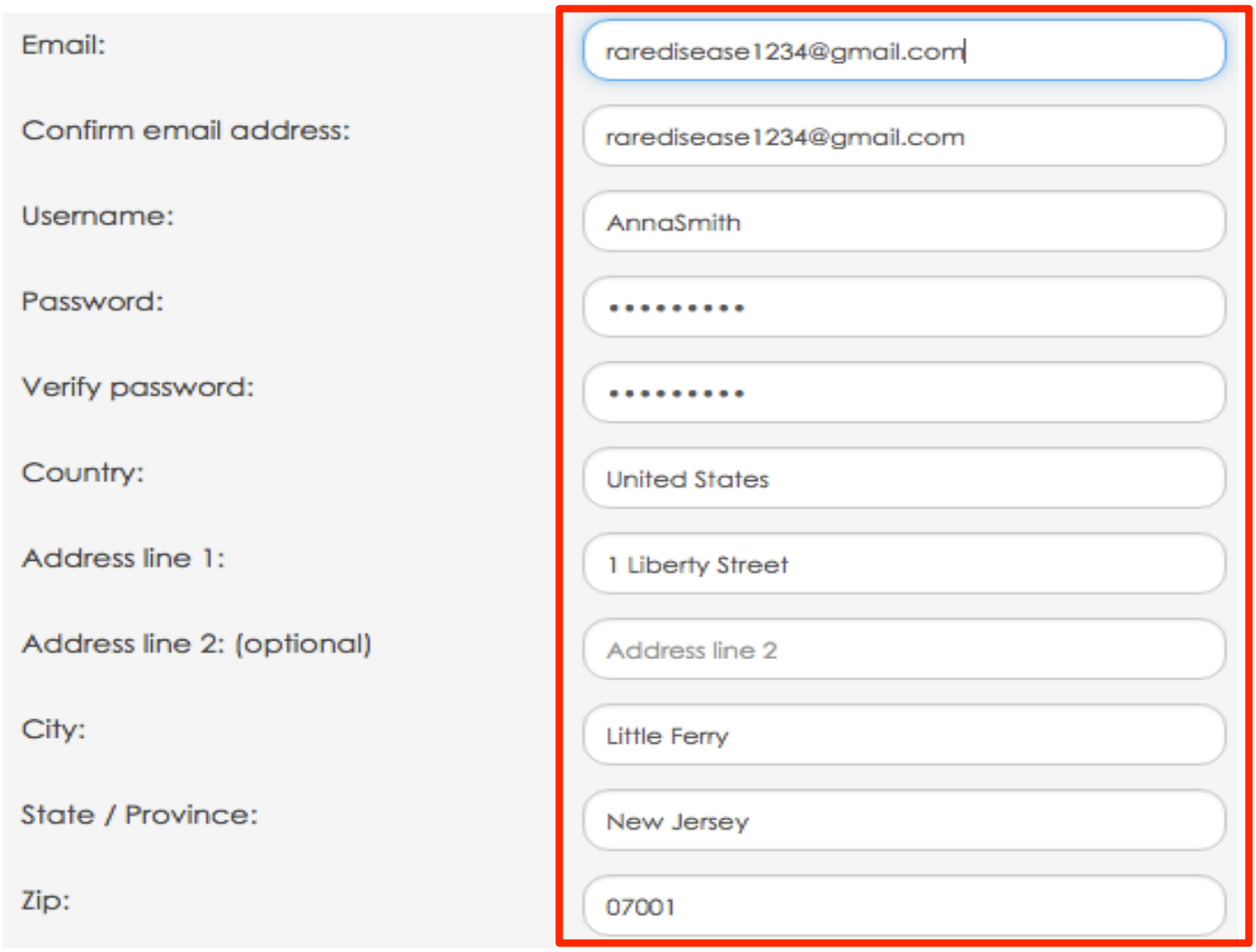
Before beginning, you will need a copy of the lab report that you received either from your doctor or from the lab. MyGene2 will require you to enter the genetic information from this lab report. MyGene2 will also need a short description of the symptoms or signs of the condition you are reporting. This example will use Anna's information from "Example Case Story" provided on our website

When you are ready to begin, go to: https://www.mygene2.org/MyGene2/

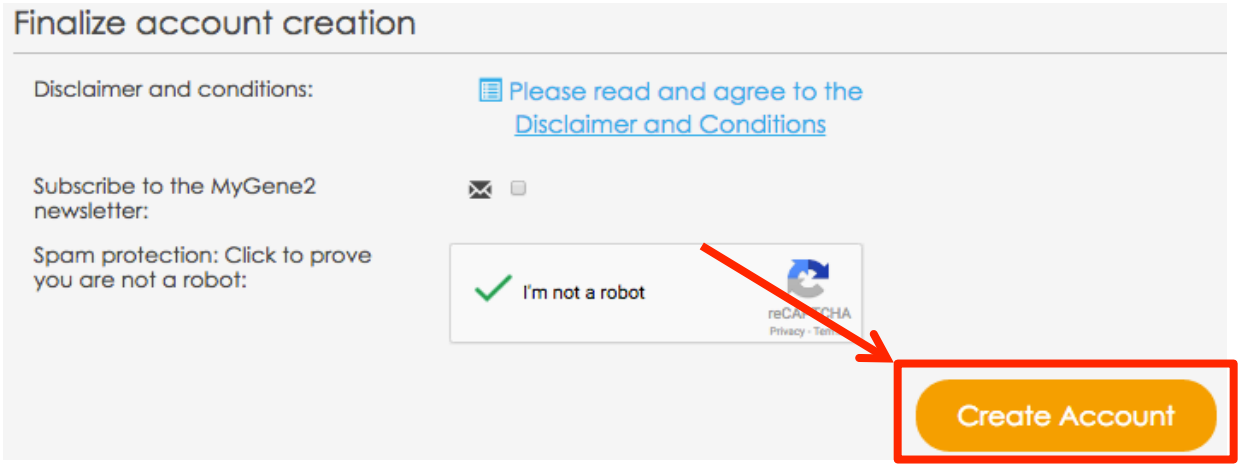
Enter in your first and last name, and from the drop down menu, select the appropriate relationship to the person with the rare condition.



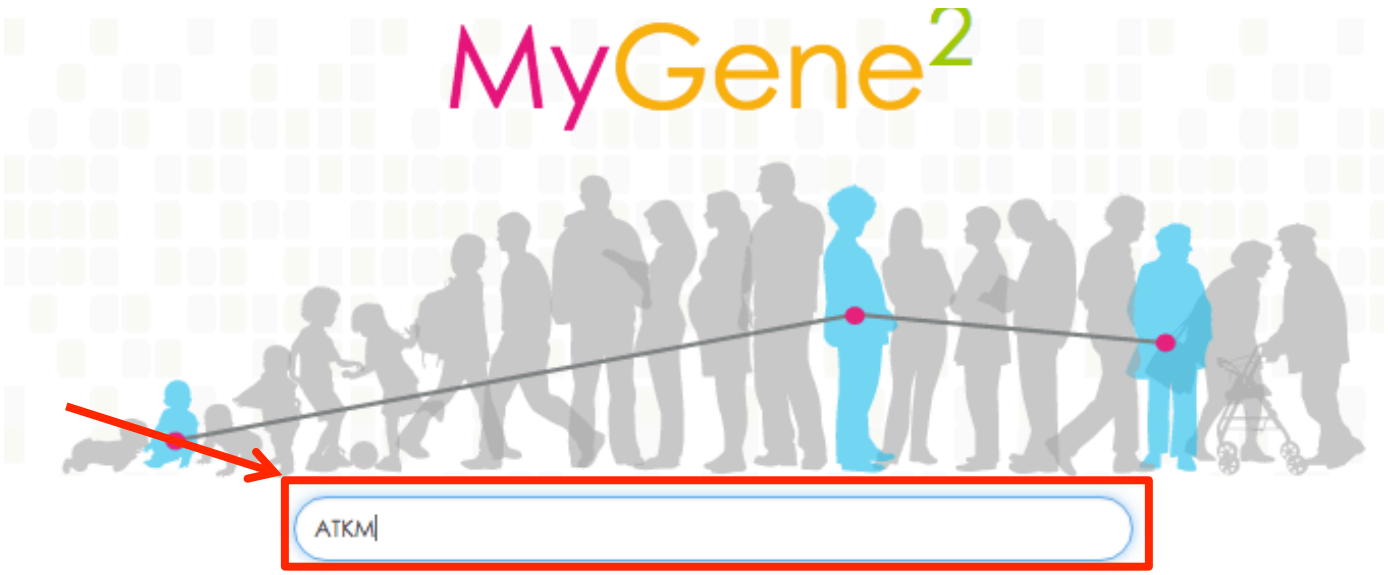
Once you have established your relationship to the person with the rare disease, you need to add this additional information.



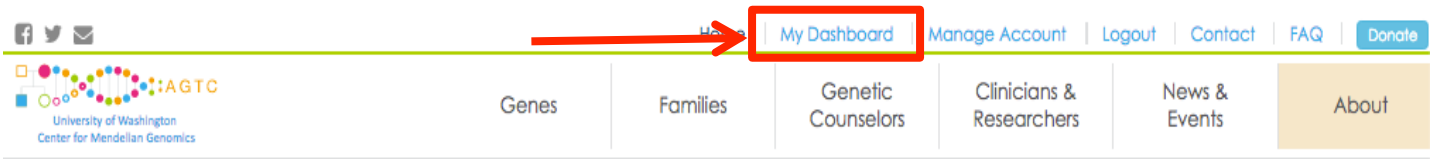
Once you have read and agreed to the "Disclaimer and Conditions" you are ready to create your account!



On the homepage “Search by gene” allows you to see if the gene in your report has already been entered into the database by someone else. This can be helpful in finding other families with a similar condition. There are times when people with different changes in the same gene result in very different conditions. It is important to look at the clinical characteristics of the affected person to see if what they have described is similar to what you or your loved on has experienced. If the gene is new to MyGene2, no dropdown menu will appear and you will be the first to enter in this information.

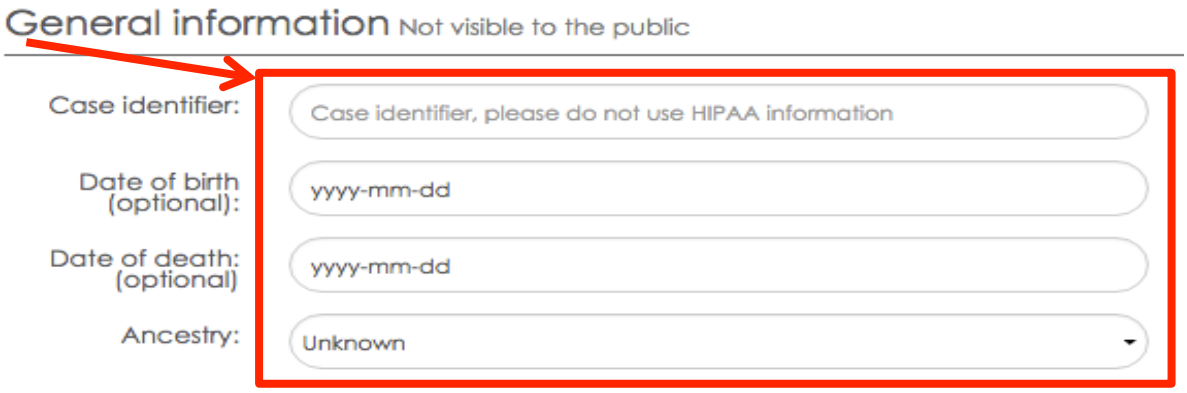


Click on “My Dashboard” at the top of the screen to begin creating your profile which will include genetic information and the description of the condition. MyGene2 refers to individual profiles as “Cases”.

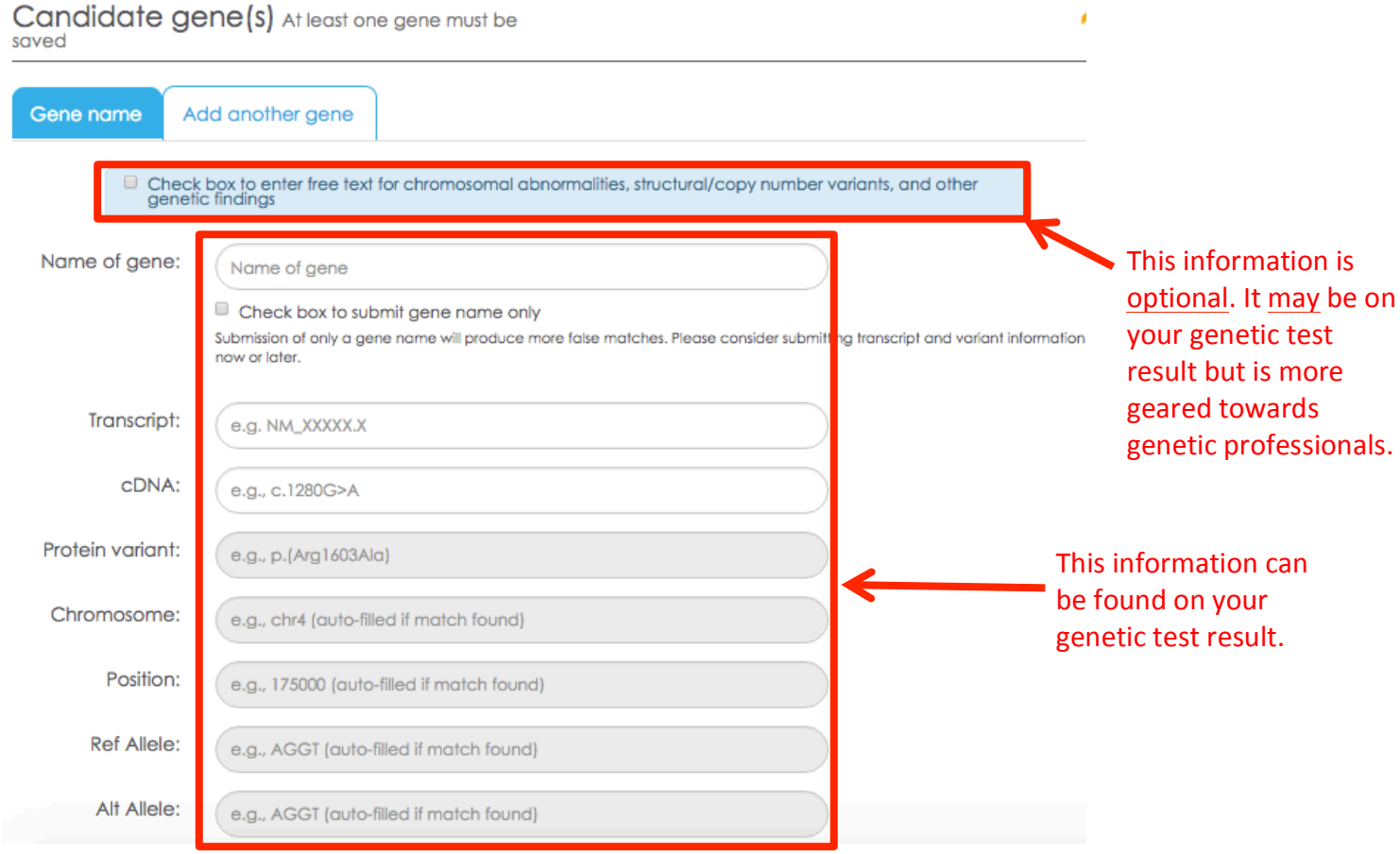


Before you can begin looking at MyGene2’s database, you will need to create your own “Case”. Whether the information is about you, or a family member, once the “Case” has been created, you will have access to all the other Cases that have been placed on the website.

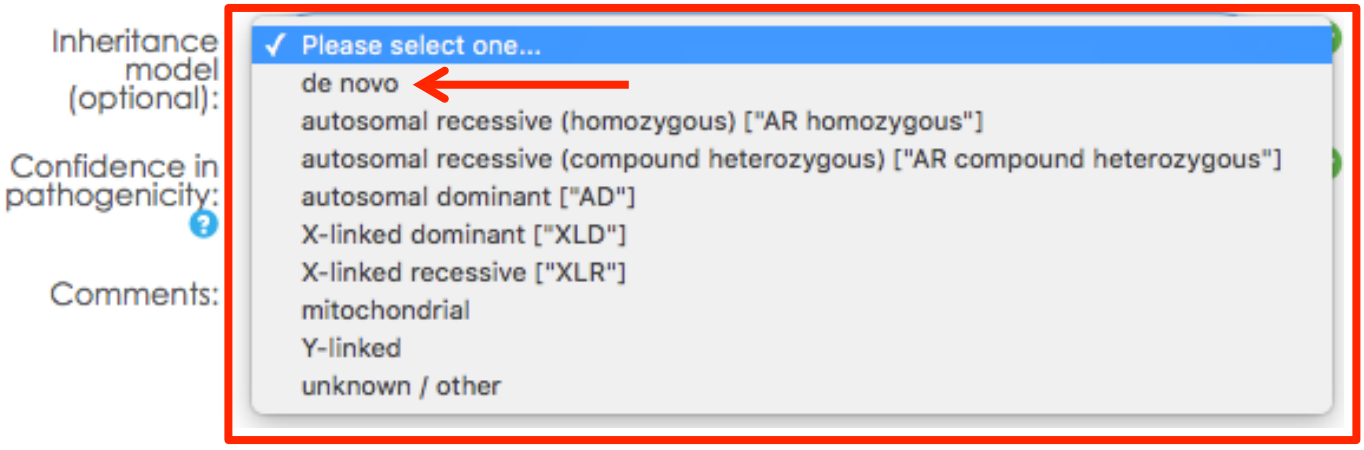
Begin by entering in general information. The “Case identifier” is similar to a username that one might create for any social media page. However this cannot include anyone’s first or last name. An example that could be used in Anna’s case is “ATKMchild.”
“HIPAA" stands for Health Insurance Portability and Accountability Act. "HIPAA information" is personal information that can be used to identify you. MyGene2 requests you do not use HIPAA information for your “Case identifier” for privacy reasons. If you are matched with another individual, your “Case identifier” will be the only information shared with them in the initial contact.



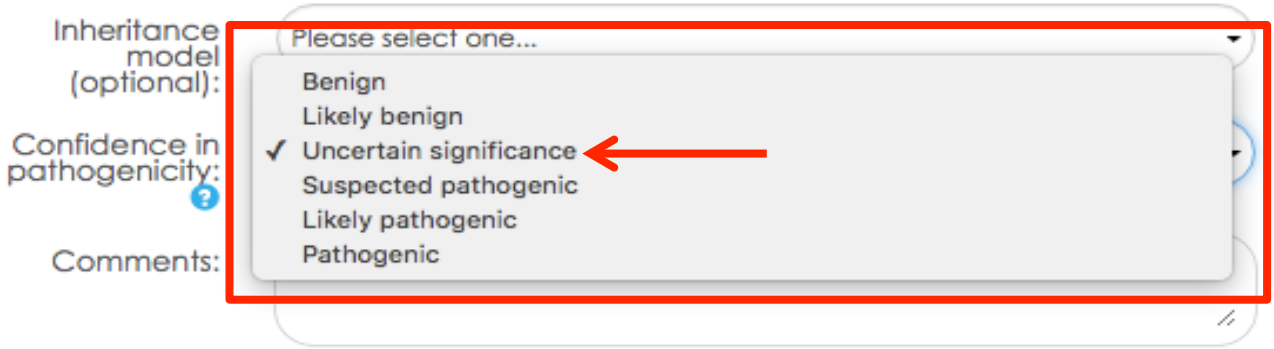
Now you will enter the genetic information about your gene into the “Candidate gene(s)” section. This will require your genetic test results from the lab or your provider. MyGene2 gives examples of the format of the information that is needed. This format should match the one on your lab report. This part can be really tricky but try your best! If your doctor is willing to help, he or she may be a great resource. If you are unsure about some information, leaving the box empty is okay as well. However, the more information you can provide, the more accurately the database will be able to match you with others.



As you scroll down, more information is requested about your genetic test results. “Inheritance model” should also be included. For most N-of-1 cases, this genetic change will often be “de novo” meaning it was new in the affected individual, and not inherited from either parent. If you need more clarification on these terms, you can find descriptions in “Glossary” on the DNA Unicorn website.

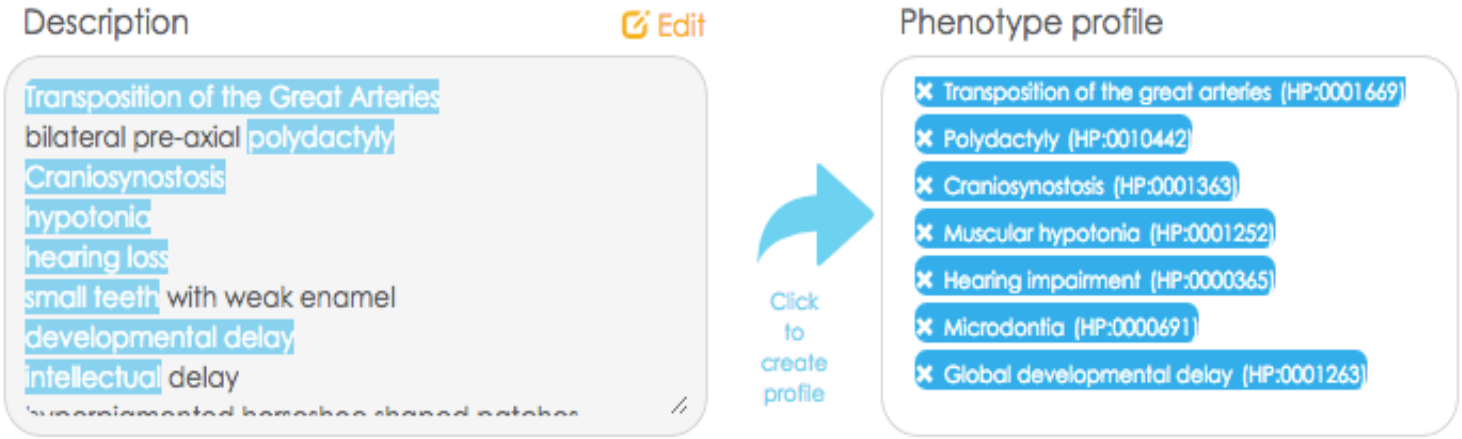


“Confidence in pathogenicity” is something that is determined by the lab. “Pathogenicity” means disease-causing. For many N-of-1 cases, the lab result may say “Variant of Uncertain Significance” meaning this specific genetic change has not been observed in enough individuals for scientists to fully understand whether or not it is causing the condition. As more is understood about a specific genetic change, this “Confidence in pathogenicity” can change to benign or pathogenic. “Benign” means the genetic change does not cause disease and “Pathogenic” means that it does.

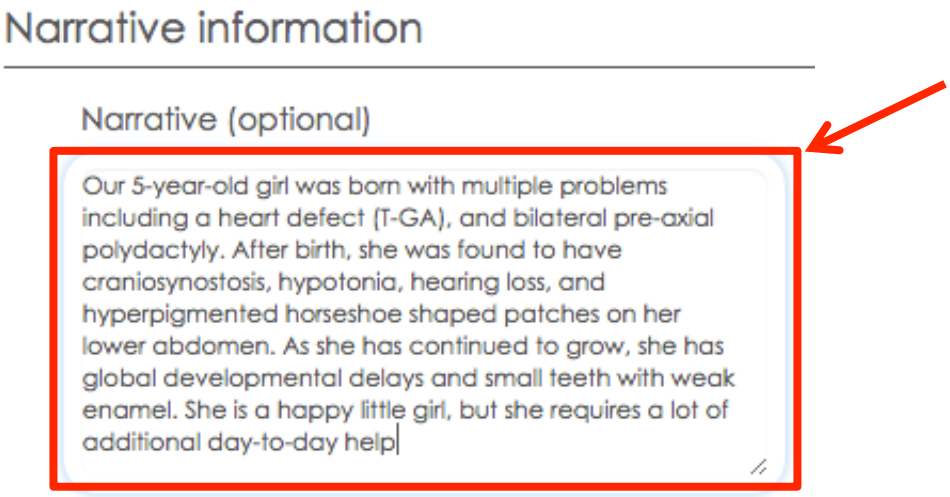


There is a spot where you are asked to input the name of the condition. Belonging to the N-of-1 community also means the condition you are describing most likely does not have a “name”. If at some point there is a name for the condition, you can always come back to change the title.

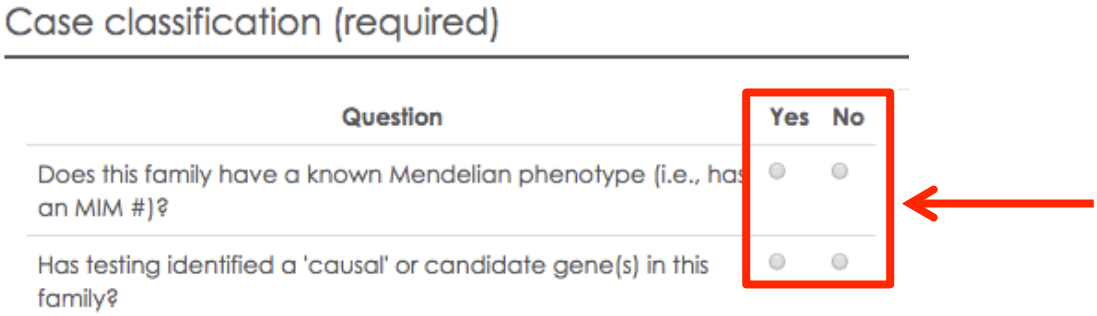
Next you will add descriptions of the condition in the left box. MyGene2 will do it's best to match what you put in with appropriate scientific terms. This process helps them standardize the language to better match individuals within the database. The terms highlighted in blue could be matched. Double-check the column on the right to be certain these terms match your description. Google and our "HPO How-To Guide" may be helpful to complete this step. If you feel the term MyGene2 suggests is not a good match, click on the little "X" to remove it. A description of Anna's symptoms can be seen below.



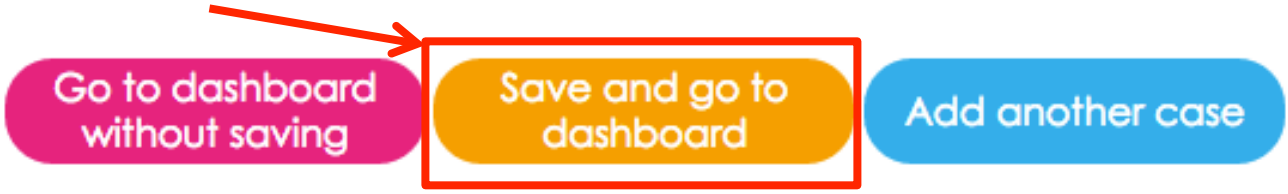
Now you can enter a brief narrative about your, or your family member's condition. What affects day-to-day activities the most? What are some unique symptoms that someone else may be able to relate to?



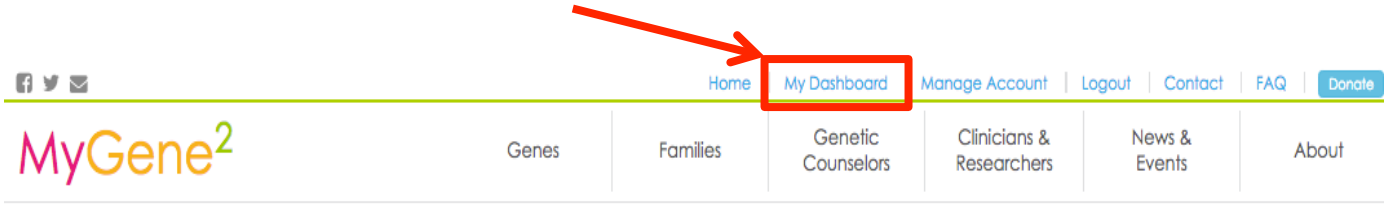
For "Case classification" the first question asks about a gene classification system used by doctors and researchers. If you are aware of a MIM# you should enter it in. The second question asks whether or not the gene identified through testing has been described as a "causal or candidate gene" This means a single gene that may be responsible for the condition.



You have now completed your first MyGene2 case profile! As you continue to scroll down there may be some “Notices and Information” Please read through these. Once you feel ready, at the bottom of the page you can now “Save and go back to dashboard”



Anything you have entered can be changed in the future under “My Dashboard”



MyGene2 also provides additional resources for families, genetic counselors, clinicians, and researchers. The “Families” tab is a great place to learn more about other families using this database.

There is also a “Learning Center for Families.” This will bring you to user-friendly resources that may help you educate yourself about genetics and find out more about the healthcare providers who have accompanied you on your journey.