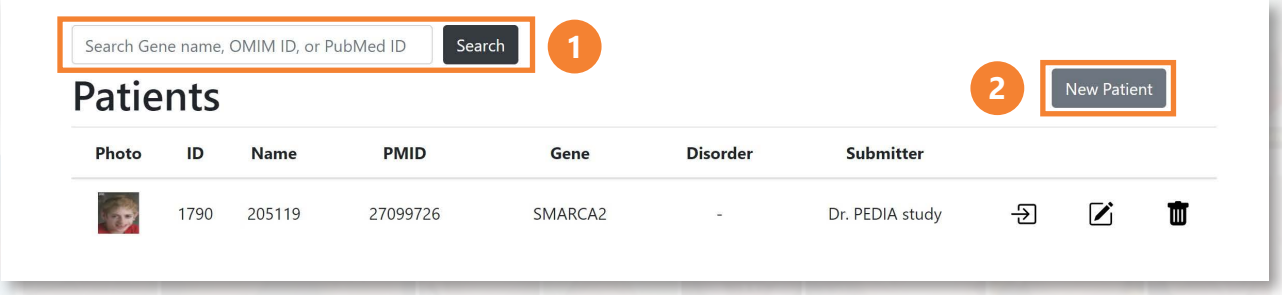
How to:
Contribute a case to GestaltMatcherDB

1 Identify a suitable publication

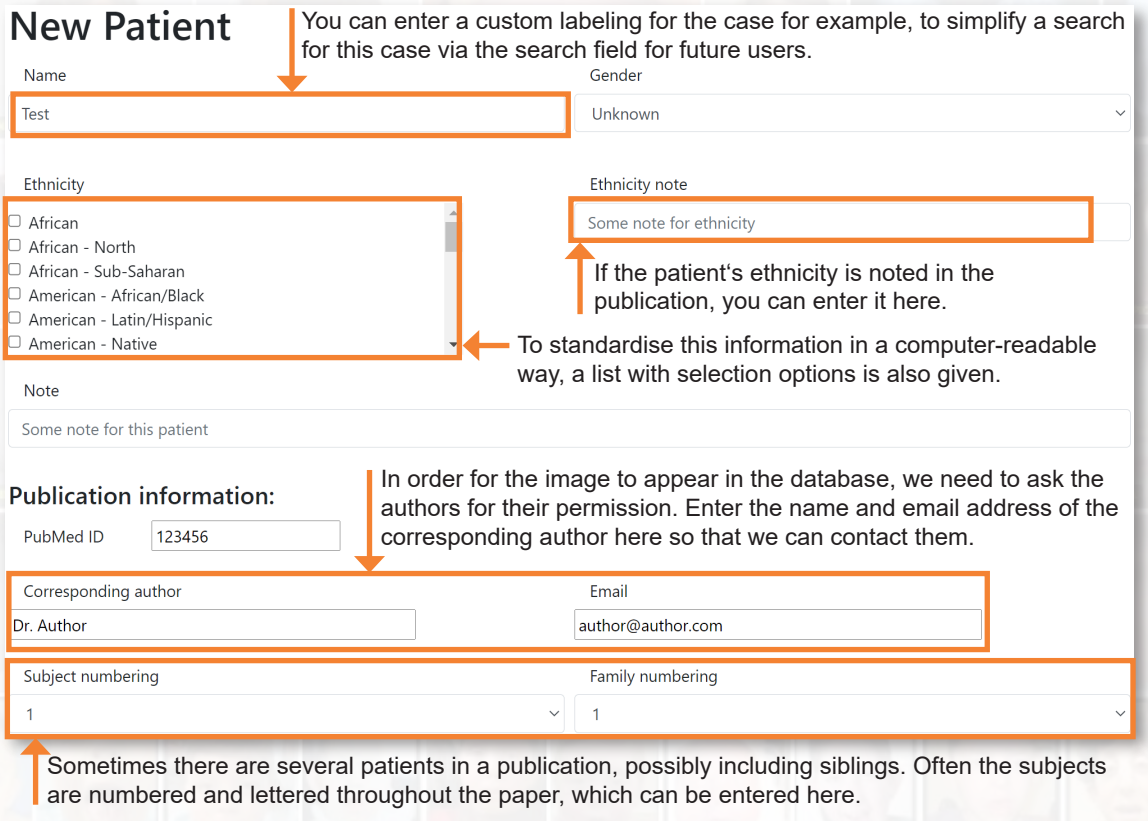
You can find original research papers, case reports and so on, for example, via the gene or syndrome name at Pubmed (https://pubmed.gov/). They should contain at least one portrait of the patient with a clinically or molecularly confirmed diagnosis.
On this page you can see how many patients per gene have already been uploaded: https://gestaltmatcher.gene-talk.de/statistics. Use the search field on https://gestaltmatcher.gene-talk.de/patients to search for the PMID and check whether images from the publication are already available in the database.

2 Open a new case

If you have found a suitable publication that is not yet in the database, click on „New patient“. A new page opens where you can enter the patient‘s details.

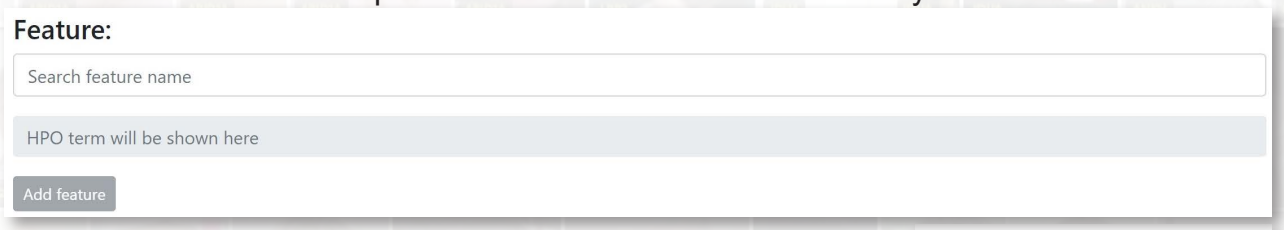


3 Enter patient and publication characteristics

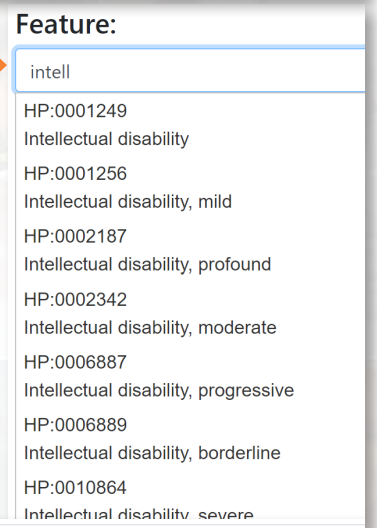


4 Enter the clinical features of the patient

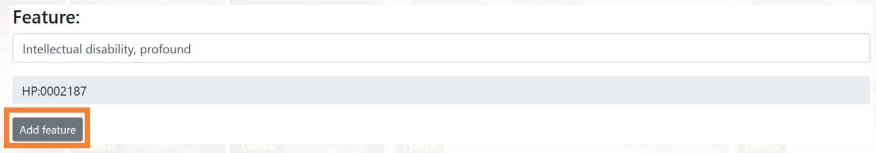
If you scroll down, you will see the part where the clinical features, the disease-causing gene and the corresponding syndrome can be entered. The symptoms and phenotypic features mentioned in the publication can be entered here one by one:



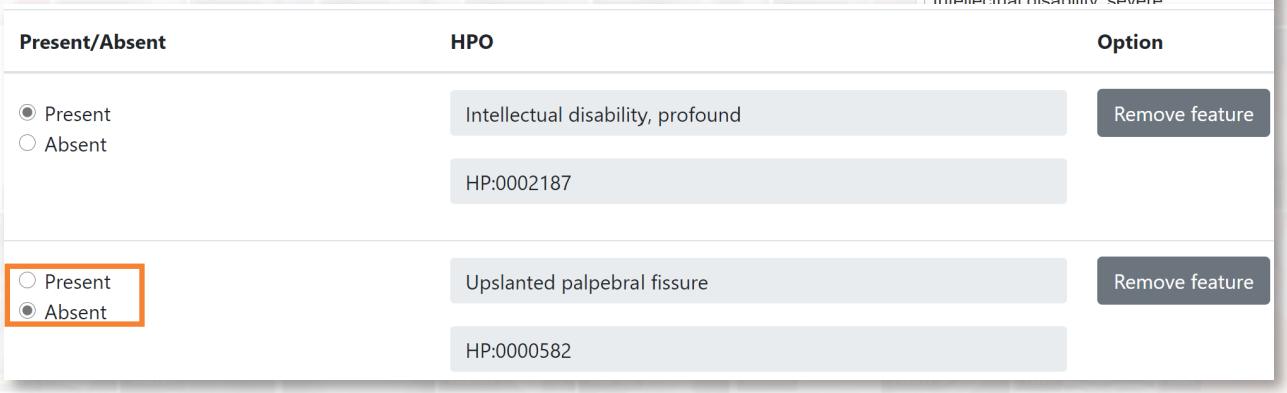
GestaltMatcher offers an AutoFill function in order to simplify the selection of the correct HPO term.



When you have found the most suitable HPO term, click on „Add Feature“ to add it to the list:

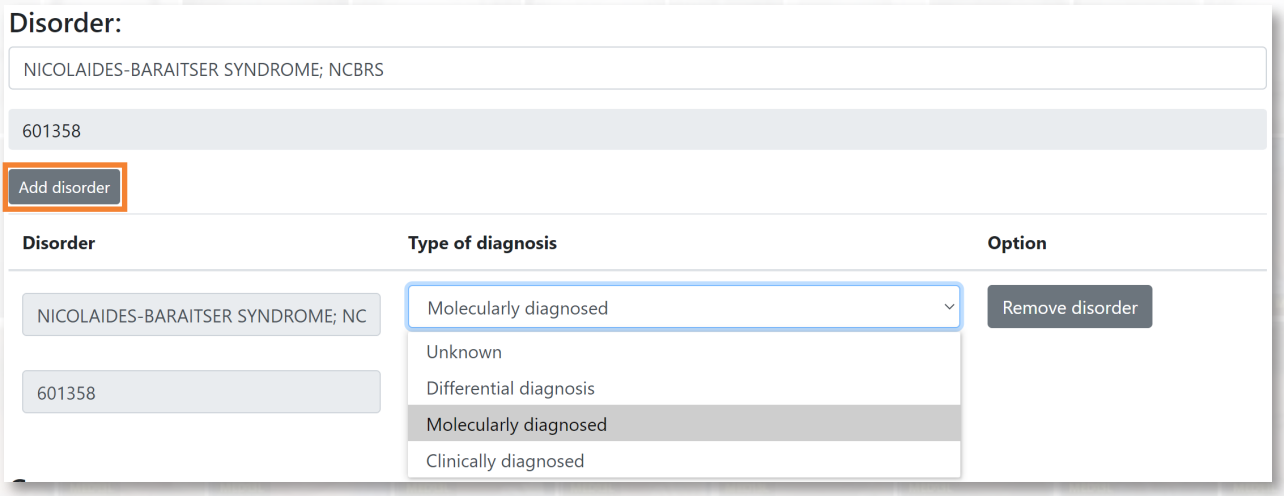


Sometimes a publication explicitly mentions that a feature could not be detected in the patient. In this case, please enter the feature as described above and then mark it as „Absent“ in the list:



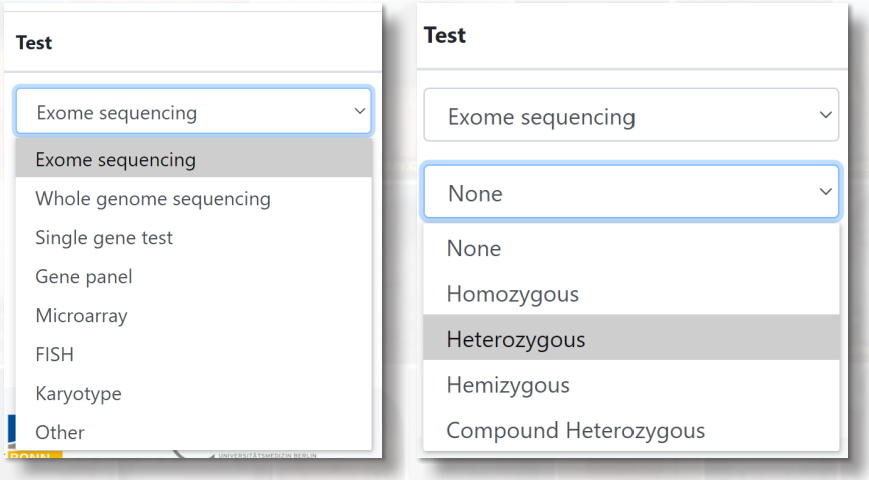
5 Enter the diagnosis of the patient

As with the feature selection, there is also an AutoFill function for the diagnosis. After you have selected the appropriate diagnosis, do not forget to click on „Add Disorder“ (if the patient has multiple syndromes, you can add more). You can then select whether the diagnosis was made clinically or whether it could be confirmed molecularly:

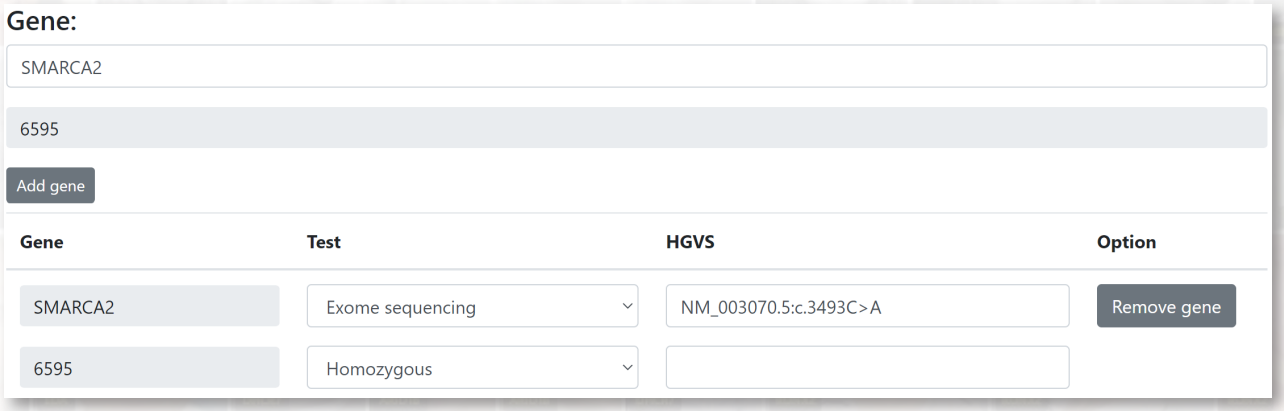


6 Enter the disease-causing genetic change

First you have to enter the affected gene. You do this in a similar way to how you entered the features and the syndrome before.
You can then select the test method and the mode of inheritance:



If available, enter the HGVS code in the free text field. Please use the following nomenclature: https://varnomen.hgvs.org/. If you are unsure, the syntax checker of the Mutalyzer can assist you in detecting possible errors: https://mutalyzer.nl/syntax-checker. You can use the second free text field for the second mutation, if it is a compound heterozygous inheritance.

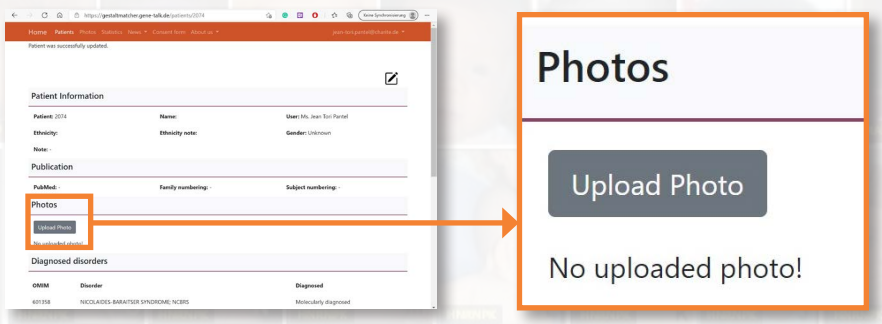


7 Save the patient information

Once you have gone through the previous steps, click on „Create patient“. You can edit and delete the information at any time later.

8 Upload photos and medical images

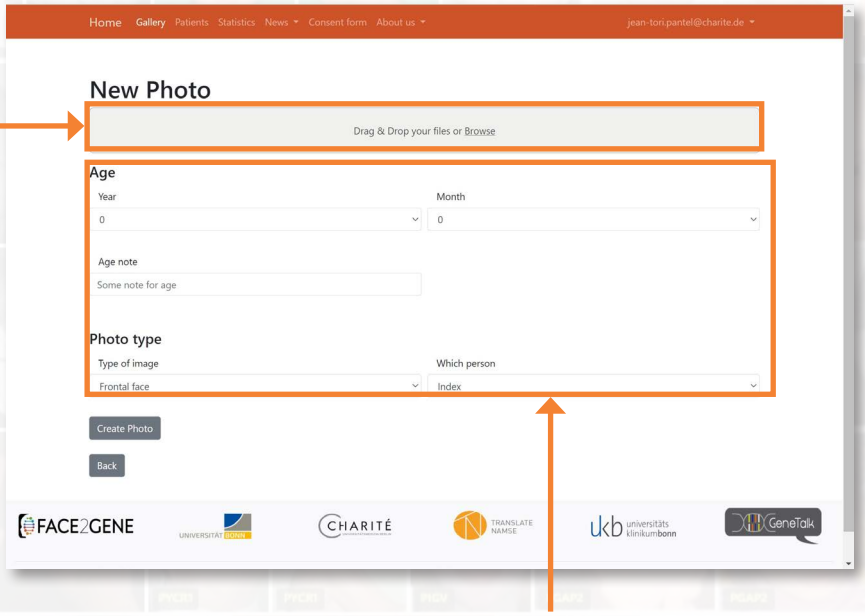
On the next page you will see an overview of the data entered. You can also upload the pictures belonging to the patient here: Portrait pictures and profile pictures, possibly with different ages, pictures of the family and medical imaging data such as X-rays or funduscopies.



8 Upload photos and medical images *(continued from last page)*

There are multiple options to upload a photo to the database. The easiest one is to drag the picture directly from the publication to the „Drag & Drop“ field. This only works for publications implemented in a website - it does not work for publications in PDF format. In this case you need to take screenshots of the photos in the publication.

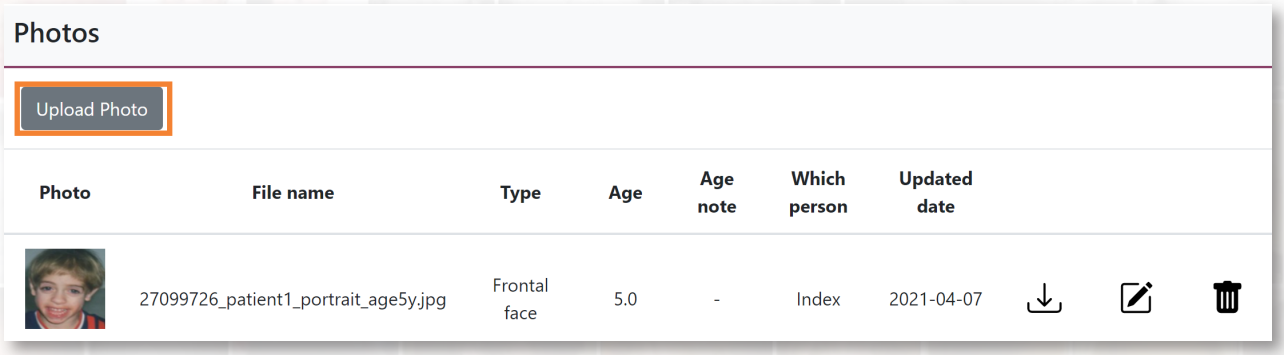
To do this, you can use the screenshot tool that is usually pre-installed on your computer. The screenshot works best if you load the image in the best quality and filling your screen. You can copy the image from the screenshot programme and paste it on the displayed page using the familiar keyboard commands, or you can drag it from the programme into the corresponding field.



Alternatively you can save the images on your hard drive. It is best to give them unique names, such as „#PMID_patient1_hand-Xray.png“. Upload one of the photos by clicking on „Choose file“ and selecting the desired image from the explorer.

You can then select the age of the person pictured and the type of photo - i.e. whether it is a hand X-ray or a portrait photo and if the person on the picture is the patient or, for example, his or her brother.

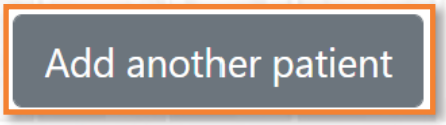
Click on the „Create Photo“ button to upload the image and to save the corresponding data on the server. You will be redirected back to the case overview, where you should now also see the uploaded photo. You can upload more photos by clicking on „Upload photo“.



9 Done!

Congratulations, you have uploaded your first case to the GestaltMatcher DB. Thank you for your important contribution!

If you want to upload another patient from the same paper, click on „Add another patient“ and the core data will be copied to the new case.



If you have any questions, please do not hesitate to contact us. We are always happy to receive feedback! Email: pkrawitz@uni-bonn.de, Twitter: @GestaltMatcher.