

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.

Photo	ID	Name	PMID	Gene	Disorder	Submitter
	1790	205119	27099726	SMARCA2	-	Dr. PEDIA study

3 Enter patient and publication characteristics

New Patient

You can enter a custom labeling for the case for example, to simplify a search for this case via the search field for future users.

Name: Test
Gender: Unknown

Ethnicity:
 African
 African - North
 African - Sub-Saharan
 American - African/Black
 American - Latin/Hispanic
 American - Native

Ethnicity note: Some note for ethnicity
If the patient's ethnicity is noted in the publication, you can enter it here.

Note: Some note for this patient
To standardise this information in a computer-readable way, a list with selection options is also given.

Publication information:
PubMed ID: 123456
Corresponding author: Dr. Author
Email: author@author.com
In order for the image to appear in the database, we need to ask the authors for their permission. Enter the name and email address of the corresponding author here so that we can contact them.

Subject numbering: 1
Family numbering: 1
Sometimes there are several patients in a publication, possibly including siblings. Often the subjects are numbered and lettered throughout the paper, which can be entered here.

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:

Feature:

HPO term will be shown here

Add feature

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

Feature:

intell

HP:0001249

Intellectual disability

HP:0001256

Intellectual disability, mild

HP:0002187

Intellectual disability, profound

HP:0002342

Intellectual disability, moderate

HP:0006887

Intellectual disability, progressive

HP:0006889

Intellectual disability, borderline

HP:0010864

Intellectual disability, severe

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

Feature:

HP:0002187

Add feature

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:

Present/Absent

HPO

Option

Present

Absent

Intellectual disability, profound

Remove feature

HP:0002187

Present

Absent

Upslanted palpebral fissure

Remove feature

HP:0000582

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:

Disorder:

601358

Add disorder

Disorder

Type of diagnosis

Option

NICOLAIDES-BARAITSER SYNDROME; NC

Molecularly diagnosed

Remove disorder

601358

Unknown

Differential diagnosis

Molecularly diagnosed

Clinically diagnosed

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:

Test

Exome sequencing

Exome sequencing

Whole genome sequencing

Single gene test

Gene panel

Microarray

FISH

Karyotype

Other

Test

Exome sequencing

None

None

Homozygous

Heterozygous

Hemizygous

Compound Heterozygous

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.

Gene:

SMARCA2

6595

Add gene

Gene	Test	HGVS	Option
SMARCA2	Exome sequencing	NM_003070.5:c.3493C>A	Remove gene
6595	Homozygous		

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.

Create Patient

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.

Patient Information

Name: User Mr. Jean-Eric Perard

Ethnicity note: Gender: Unknown

Note:

Publication

PubMed: Family numbering: Subject numbering:

Photos

Diagnosed disorders

OMIM: Disorder: Diagnosed

607108: NICOLAIDES-SARATIS SYNDROME; NCERS: Molecularly diagnosed

Photos

Upload Photo

No uploaded photo!

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.

The screenshot shows the 'New Photo' form with the following fields and options:

- Drag & Drop your files or Browse** (highlighted with an orange box)
- Age**: Year (0) and Month (0) dropdowns
- Age note**: Text input field with placeholder 'Some note for age'
- Photo type**:
 - Type of image: Frontal face (dropdown)
 - Which person: Index (dropdown)
- Create Photo** (button)
- Back** (button)

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“.

Photos

[Upload Photo](#)

Photo	File name	Type	Age	Age note	Which person	Updated date			
	27099726_patient1_portrait_age5y.jpg	Frontal face	5.0	-	Index	2021-04-07			

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.

[Add another patient](#)

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](https://twitter.com/GestaltMatcher).