

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.

The screenshot shows a table titled "Patients" with one row of data. The columns are Photo, ID, Name, PMID, Gene, Disorder, and Submitter. The data row is: Photo (small portrait), ID (1790), Name (205119), PMID (27099726), Gene (SMARCA2), Disorder (-), Submitter (Dr. PEDIA study). To the right of the table is a "New Patient" button. At the top left is a search bar with placeholder "Search Gene name, OMIM ID, or PubMed ID" and a "Search" button. A circled "1" is next to the search bar, and a circled "2" is next to the "New Patient" button.

3 Enter patient and publication characteristics

The screenshot shows the "New Patient" form with several fields highlighted and annotated:

- Name:** Test (highlighted with an orange border)
- Gender:** Unknown
- Ethnicity:** A list of options including African, African - North, African - Sub-Saharan, American - African/Black, American - Latin/Hispanic, and American - Native (highlighted with an orange border).
- Ethnicity note:** Some note for ethnicity
- Note:** Some note for this patient
- Publication information:** PubMed ID: 123456 (highlighted with an orange border)
- Corresponding author:** Dr. Author
- Email:** author@author.com
- Subject numbering:** 1
- Family numbering:** 1

Annotations provide additional context:

- An arrow points to the "Name" field with the text: "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."
- An arrow points to the "Ethnicity" list with the text: "To standardise this information in a computer-readable way, a list with selection options is also given."
- An arrow points to the "Ethnicity note" field with the text: "If the patient's ethnicity is noted in the publication, you can enter it here."
- An arrow points to the "Publication information" section with the text: "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."
- An arrow points to the "Subject numbering" field with the text: "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:

Search feature name

HPO term will be shown here

Add feature

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:

Feature:

Intellectual disability, profound

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

- Present
- Absent

HPO

Intellectual disability, profound

Option

Remove feature

HP:0002187

- Present
- Absent

Upslanted palpebral fissure

Remove feature

HP:0000582

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

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:

NICOLAIDES-BARAITSER SYNDROME; NCBRS

601358

Add disorder

Disorder

Type of diagnosis

Option

NICOLAIDES-BARAITSER SYNDROME; NC

Molecularly diagnosed

Remove disorder

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:

The image consists of two side-by-side screenshots of a dropdown menu. The left screenshot shows a list of test methods: Exome sequencing (selected), Whole genome sequencing, Single gene test, Gene panel, Microarray, FISH, Karyotype, and Other. The right screenshot shows a list of inheritance modes: None (selected), Homozygous, Heterozygous, Hemizygous, and 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	<button>Remove gene</button>
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.

The image is a screenshot of a web-based patient information form. In the 'Photos' section, there is a button labeled 'Upload Photo' with a red box drawn around it. Below the button, the text 'No uploaded photo!' is displayed.

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 a web-based form titled "New Photo". At the top, there is a large orange-bordered input field labeled "Drag & Drop your files or Browse". Below this, there are fields for "Age" (Year and Month dropdowns), "Age note" (text input), and "Photo type" (dropdowns for "Type of image" (Frontal face) and "Which person" (Index)). At the bottom left are "Create Photo" and "Back" buttons, and at the bottom right are logos for FACE/GENE, CHARITÉ, TRANSLATE NAMEE, ukb universitäts klinikum bonn, and GeneTalk.

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

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](#).