Information about the GestaltMatcher database for patients and research participants

Until the correct diagnosis is made, patients with rare diseases and their families often experience a nerve-wracking odyssey. In many cases, it is crucial to meet a specialist who has the right idea. In recent years, improvements in digital image analysis technology have enabled the development of computer programs that can speed up the diagnostic process while being cost-effective and widely available.

For instance, typical facial features can be found in many genetic syndromes, providing valuable information about the underlying disease. Specialists in human genetics rely on their clinical experience to identify and correctly classify these characteristics. In order for a computer program to reliably recognize such features, a large amount of data is needed to train the algorithms - but after the training, the program can be used by pediatricians, for example.

The GestaltMatcher database will provide such a basis for experts in medicine and computer science to develop and improve diagnostic programs for rare diseases. You can support us by providing us with pictures of you or your child. On the following pages you will find information on how to upload your or your child's data.

1 How to participate in the GMDB project

We are pleased that you have become aware of our research project and that we have your interest. You may have been **A** approached by your physician, **B** contacted by us through your patient association, or you may have **C** otherwise encountered this page.

To upload your data, you will need a personal access link. You can get this link as follows:

A After your doctor has created a case for you via his/her profile in the GMDB, the link visible only to him/her will appear on the case page, which he/she will pass on to you. With this link you can then also access your case page.

Shown is a screenshot of how the case description is displayed for your physician. The personal access link with which you get access to your page is placed in the upper left corner.

B Please contact the head of your patient association. We will create case pages for you and send you the personal link by email.

C Please contact us directly so that we can discuss the study with you, set up a case page and send you the link.

❷ Requirements for participation

We are pleased to include photos and data of you or your child if the diagnosis has been confirmed by laboratory diagnosis. In the case that the diagnosis is not yet known, we would like to refer you to a research project based on the GMDB with the aim to identify still unknown genetic syndromes with the help of image analysis: http://gestaltmatcher.org/.

When you access your personal link, you will first be shown the study information, which you will need to sign for the following steps. Please read them carefully and do not hesitate to contact us if you have any questions. This is a digital consent, which you can withdraw at any time and without giving any reason.

Here you can see a screenshot of the consent form that is displayed when you open your personal access link. To participate in the GestaltMatcher database we need your consent to store and process your data. You can sign digitally directly in your browser.

If you have read the study information carefully and completely understood all aspects, please fill in the relevant fields, add your signature and click on :

Submit consent

In the next step, the consent form will be displayed again with your signature - feel free to print this page for your records. Clicking on the button shown on the right will take you to the input mask for your data.

Edit patient record

❸ How to enter data into the GMDB

In the input mask, various details about you or your child are requested, which are required for the training of intelligent computer programs. For example, the certain features of the face may differ depending on gender or ethnicity. Your doctor may have already entered some or all of the data. You can correct and add to these. If you do not know what to enter in a field, feel free to ask or simply leave the field blank.

If you are part of a patient association, you can select your group here. If your patient association is not yet listed here, then feel free to let us know so that we can add it.

You can use this field for information that might be of interest to us but is not covered by the other fields. Please do not enter any personal data here that makes you identifiable.

The entry of your or your child's symptoms, diagnosis and molecular findings will be handled by your physician.

Alternatively, you can share your medical findings with one of our physicians, who will then be happy to transfer the data.

Please do not forget to save your data by clicking on this button after you have entered all data

To save the entered data, you need to click on "Update". The window will then be refreshed. If you want to edit the data, you can click on the edit icon in the upper right corner of the screen.

4 How to upload images to GMDB

After you have clicked on "Update patient" as described in the previous step, an overview of the entered data appears. Here you can finally upload portrait and profile pictures, possibly from different ages, of yourself or your child under the "Photos" section.

Photos

Upload Photo

A There are several ways to upload a photo to the database. The easiest is to drag the picture directly from the folder on your PC into the "Drag & Drop" field. Alternatively, you can click on "Browse" and select the desired image from the file explorer.

New Photo

A Drag & Drop your files or Browse

Age **B**

Year
0

Month
0

Age note
Some note for age

Photo type

Type of image
Frontal face

Which person
Index

Distinctiveness

How would you rate the relevance of the facial gestalt in establishing the clinical diagnosis of this patient?

Supportive

Create Photo **C**

B You can then select the age of the person pictured and the type of photo - for example, whether it is a portrait or profile photo.

C Click the "Create Photo" button to upload the image and save the corresponding data on the server.

You will be taken back to the case overview, where you should now also see the uploaded photo. You can upload more photos by clicking on "Upload photo" again.

For open questions, suggestions and requests

The study director, Prof. Dr. med. Peter Krawitz, can be contacted at the following e-mail address: pkrawitz@uni-bonn.de.

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GeneTalk