GestaltMatcher research study and GestaltMatcher database.

Study information

Dear patient,

a rare mutation has been found in your genes, which has led to some unique features in you compared to other children of your age.

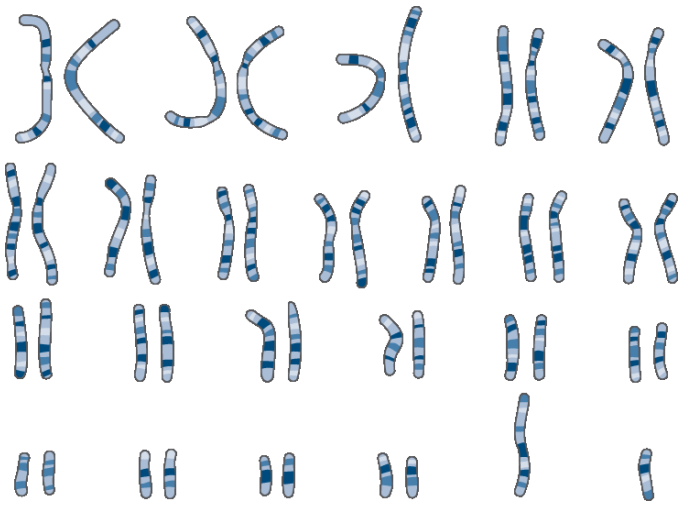

Figure 1: Chromosomes

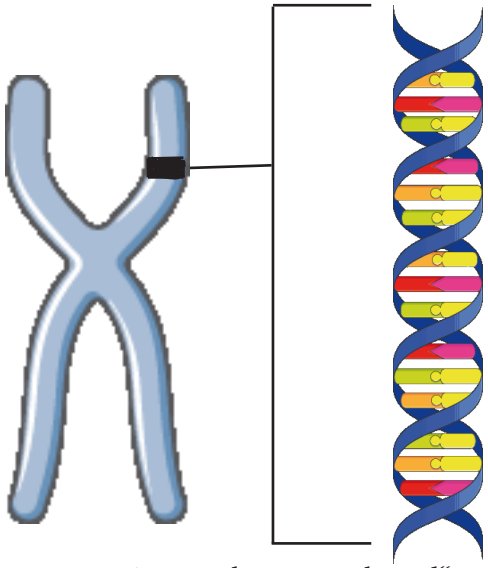

Figure 2: A gene that is „packaged“ in the chromosome

A gene is part of the blueprint of every living being. Genes are tiny and we all have many different ones, all with special functions. They are found in all the cells of our body. However, they are not just floating around freely, but are „packaged“ in the chromosomes. Among other things, our genes determine our characteristics, such as eye or hair color. We inherit them from our parents, which is why family members usually look alike. However, the genes are not exactly the same in all people, there are always changes that make us unique.

In your case, a change has occurred that is very rare and has led to very special characteristics and symptoms in you.

A doctor was able to find this change in you with a special genetic analysis and since then has been able to better understand your symptoms and therefore treat them better. Sometimes, however, it takes a long time to find such rare genetic changes and during this time it is not possible to help the patients as well as we want to. With our research, we would like to make sure that children who have similar changes to yours can get the help and support they need sooner.

There are other children who have the same or a similar change in the same gene as you do. You may have noticed that you share some of your characteristics. You can often see such similar features in the face as well. Maybe you have heard about Down syndrome. Here the similarities in the face are very impressive. Do you also recognize these similarities in the pictures below? So if a doctor suspects a genetic change in a child, he or she will look particularly closely at the face and may be able to make a guess as to which genetic change is responsible. To be able to do this, however, he or she needs a lot of experience.

Image 3: Down syndrome - Do you recognize any similarities?

There are now also computer programs that can recognize these features. But even these computer programs have to learn the features first, so that they can recognize them and match them to a known disease. In order to teach the system, we would like to show your picture to the computer system.

If a child with similar facial features to yours will upload his or her picture to the program, the system may recognize your similarities and will suggest to the doctor to screen the other child for your genetic change. The more patients provide their pictures, the better the program can learn the features and thereby detect the disorder. So with your picture you could help to shorten the time until another child gets his diagnosis.

For our study we do not need any information about your name or where you live and we do not need to perform any further tests on you. We only need a picture of you and the results of your genetic laboratory test.
Any data we store will not be linked to your name, so no one will be able to link this data to you.

We also want to share the results of the study with other scientists so they can be better informed about rare genetic disorders.

You are free to decide whether you want to participate in the research study. You can choose to participate or not.

If you have any further questions about our research study, please feel free to contact us by phone, email or mail:

info@gestaltmatcher.org


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