GestaltMatcher research study and GestaltMatcher database.

Study information

Dear patient,

You are seeing us because there is a suspicion that there is a rare change in your genes, which has led to some unique features in you compared to other children of your age.

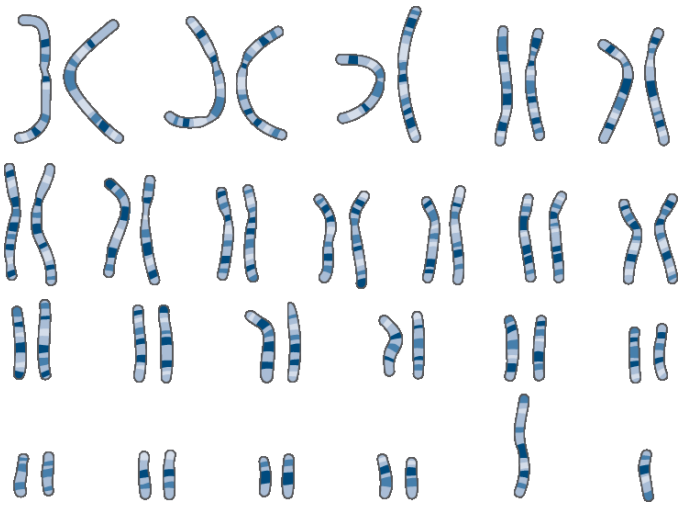

Figure 1: Chromosomes

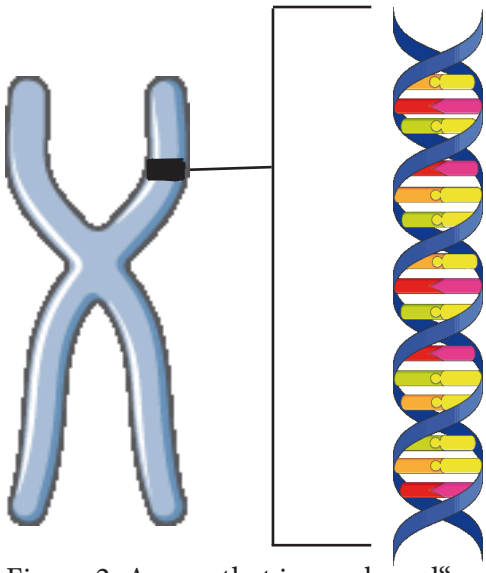

Figure 2: A gene that is „packaged“ in the chromosome

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, it is possible that a change has occurred that is very rare and has led to very special characteristics and symptoms in you.

Sometimes 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 would like to. Maybe you have already been waiting for a long time to find the reason for your unique characteristics. With our research we would like to achieve finding such genetic causes faster. Then children like you can get the help and support they need sooner.

Children who have the same change in their genes sometimes look more alike to each other than to their siblings. You may have heard of Down syndrome. In this syndrome, but also in many other genetic disorders, you can often see similar features in the face. Maybe you also recognize the 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 about which genetic change might be the cause.

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

There are also computer programs that can recognize these characteristics. We would like to show your picture to such a computer program. If there is a picture of a child with similar facial features to yours in our image collection, the computer system will recognize your similarities. It will then suggest to the doctor that you be tested for the genetic change that is present in the child. In this way, we may be able to find the cause of your special features more quickly.

If we find a genetic change in you, then the computer system also can learn from your photo and recognize the change in another child even more confidently. Perhaps we will not find a similarity to another child in you at first. Perhaps we will not find a similarity to another child in you at first, but If we store your photo, then we can compare it with all children who upload their picture later. In the future, we may find a child with the same characteristics as yours and can then investigate whether you have the same genetic mutation.

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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