Patient Information for Children (7-11 years)

on the scientific study

Identifying the Causes of Rare Diseases using Genome-Wide Sequencing

Dear Girl, Dear Boy!

Thank you for your interest in our study and for reading this information sheet.

We would like to invite you to take part in a study that will help us to find out more about where **rare diseases** come from, how to recognise them better and how they can be treated best. This investigation is completely voluntary, but you can help us so that we can better help other people like you.

You keep coming to our clinic because you have a condition that only a few persons have. That's why we call it a **rare disease.** They often appear from birth because there is a defect in the genetic material, the so-called genes. In each person, the "genes" provide a kind of plan for building the body, like the instructions for a house made of Lego bricks.

In order to be able to help people with rare diseases in a better way, it is very important to find out about mistakes in the genes, e.g. also in your own building plan, and also to investigate the disturbances in your body's metabolism caused by them.

In our study, we are looking at the whole building plan of many children, adolescents, and adults who have a rare disease, to see if there are any mistakes. If we find a mistake, we might be able to help these people better in the future, for example with new or different medicines.

We therefore want to know as much as possible about your disorder. For this purpose, we would like to take **a little more blood for the examination of your building plan during a blood sample taking that is planned anyway.** We also want to do more tests, so we might ask you for a few more biosamples (next page):

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Public Law Institution

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Your supervising physician will decide together with you and your parents which of your biosamples may still be needed.

- Fingernail sample
- Urine sample (spontaneous urine)
- 24-hour urine sample
- Saliva
- Skin biopsy (tissue sample from the skin only for children for whom a skin sample is taken anyway as part of the medical diagnosis or treatment, no additional sample is taken)
- Muscle tissue (only if a muscle sample is taken anyway as part of the medical diagnosis or treatment, no additional sample is taken)
- Cerebrospinal fluid (only as an additional sample up to a maximum of 5 ml during a routine puncture that is being performed anyway, no additional puncture is performed)

We want to keep the information about your disease and your donated body samples for a very long time. **When you turn 18, you will be asked whether we can still keep the information about you and your body samples and further examine them.**

If you no longer want the information about your illness and your donated body samples to be with us and to be further examined by us, you can stop this at any time. Neither you nor your parents will suffer any disadvantages. If you do, we will delete all the information we have about you and throw away your donated body samples.

We hope you see how important it is for us to protect the information about your disease and your donated biosamples. Your help to better detect and treat people with rare diseases is very important to us.

If you help us with our work, we would be very happy about it! Of course, you can also say that you don't want to take part.

If you have any questions, please contact us or your parents!

If you would like to join this study, please write your name on this line:

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