



GENETIC AND FUNCTIONAL STUDIES OF PROSPECTIVE MONOGENIC DISEASES

WHY HAVE YOU BEEN ASKED TO TAKE PART?

We ask you to be a part of this study because we don't know why you are ill. We would like to help you and make you feel better, but we need to find out what is making you ill. We ask for your blood / skin piece / bone marrow, which is a fluid inside your bones/ poop sample and brushing from your skin and inside your mouth, these are called biological samples. Each biological sample contains cells, which are like small bricks that build your body. Almost all cells have inside them DNA, which is like a long string composed of 4 letters: A, G, T, C. These letters are set in a special way to form a code, different for every person and scientist can read this code. Sometimes there are errors in the DNA, for example some letters are missing, and this makes cells behave differently and make people ill. What we want to do is to see the order of these 4 letters in your code and to find out if and what kind of errors there are. In the future we might be able to correct those mistakes, which will make cells to behave normally and eliminate the illness.

WHAT WILL HAPPEN IF YOU TAKE PART?

Your parents will bring you to the hospital where you will be examined by a doctor and you will have to answer his questions. If you and your parents agree to participate in this study, you will be asked to give your biological samples: blood/ bone marrow/ cerebrospinal fluid/ tumor and skin pieces/ poop sample and brushing from your skin and inside your mouth. The blood will be collected by a nurse and it will only hurt a little, but it is not dangerous. Bone marrow, cerebrospinal fluid, tumor and skin pieces will only be taken if your doctor decides that you need to have a surgery. Then you will be sleeping during the whole time and you won't feel any pain. Your parents will be with you all the time. If something is hurting you, a nurse and the doctor will help you. After a surgery you will stay at the hospital for few nights and you will probably meet other children who also participate in this study. Poop sample and brushing will be collected by a nurse during your time at the hospital, it won't hurt at all.

WHAT WILL HAPPEN IF YOU DON'T TAKE PART?

If you don't want to you don't have to agree to be a part of this study. You don't have to give your samples to scientist. There won't be any negative consequences for you and your parents if you won't participate in the study. The treatment at the hospital won't change if you don't participate.

You can also agree to participate in the study and only give one or two selected by you and your parents biological samples.