



## 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 you are healthy and we want you to donate your biological samples to help children who are ill. We would like to help them and make them feel better, but we need to find out what is making them ill. We ask for your blood / skin piece, which 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 up 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. To find out what kind of errors there are, we want to see the order of these 4 letters in sick children's code and compare it with yours. In the future we might be able to correct those errors, and this 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, piece of skin, poop sample and brushing from your skin and inside your mouth. A nurse will collect the blood and it will only hurt a little, but it is not dangerous. 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. A nurse will collect poop sample and brushing during your time at the hospital, it will not hurt at all.

### WHAT WILL HAPPEN IF YOU DON'T TAKE PART?

If you do not want to, you do not 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 biological samples selected by you and your parents.