Appendix II

Informed consent form 'Research genetic predisposition in Blount disease': DNA sequencing.

1. Purpose of the Project

We would like to invite you to participate in a research project called 'genetic predisposition in Blount disease'. The purpose of the research is to discover genetic changes associated with the disease. This should lead to a better understanding of the aetiology of Blount disease and later on, lead to better ways to prevent, detect, and treat the disease.

Body tissues are made up of cells. Cells contain DNA, which is your unique genetic material that carries the instructions for your body's development and function. Your DNA is a combination of the DNA from you parents. Many diseases can result from changes in a person's genetic material that cause cells to not work properly. Diseases who have a genetic predisposition can often be found in the DNA of the parents as well. They don't have to be affected by the disease but can be, so called 'carriers' who have the genetic predisposition but not the disease. Currently, researchers and doctors know some of the genetic changes that can cause some disease, but they do not know all of the genetic changes that can cause diseases. So far, they never looked into Blount disease. We think there might be a genetic predisposition in Blount disease. This is why we would like to invite you and your parents to participate in this research. We will perform this same process with some other patients and their parents who have agreed to participate in this research project. Combining these results, we will be able to say more about the possibly genetic predisposition in Blount disease.

2. Description of the Research

Collection of Samples and Medical Information:

- We will collect two samples from you by drawing about 4 tablespoons of blood from a vein in your arm for each sample. If you object to having blood drawn or are not sure if we drew enough blood, we will collect tissue from you by swabbing cells from the inside of your cheeks.
- We also will collect information from your medical records, including your age, ethnic background, diagnosis, disease history, medical treatments, and response to treatments.

Coding of Tissue Samples and Medical Information

- Your blood (or other tissue) sample and medical information will be labelled with a code.
- Only members of the research team 'genetic predisposition in Blount disease' at (Maastricht University medical centre, The Netherlands and St. John of God hospital, Ghana) will have the information that matches the code to traditionally-used identifying information, such as your name, address, phone number. The research team will keep the information that matches the code to this traditionally-used identifying information in a safeguarded database. Only very few, authorized people, who have specifically agreed to protect your identity, will have access to this database. All other researchers and personnel, including those who will be working with your samples and medical information, will not have access to any of the traditionally-used identifying information about you.

Storage and Release of Samples and Medical Information

- Your coded blood (or other tissue) samples will be sent to The Netherlands for detailed analysis. Remaining portions of your samples will be stored for an unlimited period of time for future use in research related to diseases.
- Information from analyses of your coded samples and your coded medical information will be put into a database along with information from the other research participants.

3. Financial Compensation/Costs

You will not be paid to participate in this project. Your blood (or other tissue) samples and your medical information will be used for research purposes only and will not be sold. It is possible that some of the research conducted using your samples or information eventually will lead to the development of new diagnostic tests, new drugs or other commercial products. Should this occur, there is no plan to provide you with any part of the profits generated from such products.

4. Potential Benefits of Participating in the Project

You should not expect to personally benefit from this research. The main reason you may want to participate is to help researchers and health professionals around the world to better understand the cause of your disease so that they can find better ways to prevent, detect and treat Blount disease. You may feel good knowing that you may be helping future patients. If it turns out there is a genetic predisposition in Blount disease, this could be useful information for you, knowing your children may be affected as well.

5. Potential Risks of Participating in the Project

Physical Risks

- Possible side effects from drawing the blood sample include mild pain, bleeding, bruising, and infection at the site of the needle insertion. Fainting or light-headedness can sometimes occur, but usually last only a few minutes.

Psychological or Social Risks Associated with Loss of Privacy

- Your privacy is very important to us and we will use many safety measures to protect your privacy. However, in spite of all of the safety measure that we will use, we cannot guarantee that your identity will never become known.

6. Confidentiality

We will make every attempt to protect your confidentiality and to make sure that your personal identity does not become known. This signed consent form will be stored in a locked file that will be accessible only to a very small number of authorized people involved in this project. We will carefully follow the coding, storage, and release plan explained in the 'Description of the Research' section of this document.

7. Project Results

In general, results from this research project will not be given back to you or put into your medical records. In some situations, the results might be important to your health or medical care. If this occurs, we will contact you to see if you want to learn more. If research from this project is published in professional journals, there will be no traditionally-used identifying

information, such as your name, address, telephone number, or insurance number, included in the publications.

8. Alternatives to Participating in the Project

The alternative option is not to participate.

9. Voluntary Participation

The choice to participate in this research by donating your tissues and medical information is completely up to you. No matter what you decide to do, your decision will not affect your medical care. Refusal to participate will involve no other penalties or loss of benefits to which you are entitled.

10. Withdrawal from the Project

Once data are generated from the samples you provided, and those data are placed in the database as described elsewhere in this consent, you will not be able to withdraw the data, only the samples. If you would like to withdraw from this project you can contact dr. H. Staal, department of orthopaedics, MUMC, The Netherlands and she will destroy any remaining tissue samples of yours that have been obtained for the study. In addition, it may be possible for her to destroy the link between you and your genetic and medical information.

However, the samples and data that have already been distributed to other research centres or placed in the research databases will not be able to be withdrawn.

11. Contact Information

If you have any questions about the project, about your rights as a research participant, or about any research-

related injury, please contact N. Jansen (n.j.jansen@amc.nl).

Source:

The national human genome research institute medical sequencing program - Consent Form: Example 2 (DNA Sequencing)

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Agreeing to Participate in the Project

To participate in this research, you must agree to ALL of the following statements:

- I voluntarily agree to donate two blood samples and/or a cheek tissue sample to be used for this research project.
- I agree to release information from my medical records for this research project.
- I agree to have my coded genetic information and coded medical information placed in a secure database.
- I understand that there is a risk that someone in the future might be able to use information in this database to identify me.

Please sign your name here if you agree with the above four statements.

Your signature: _____

Date: _____

Signature of Doctor/Researcher_____