



THE UNIVERSITY
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AUSTRALIA

Information Sheet for Data Collection

The Queensland Bone Dysplasia Registry (QBDR) A registry for patients with genetic disorders of the skeleton

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A. What is the purpose of this study?

Bone dysplasias are genetic disorders affecting the normal growth and development of the skeleton. They can cause a variety of medical problems including short stature, limb deformities, joint pain and bone fragility. There are more than 300 different kinds of bone dysplasias and most of them are very rare (less than 1 in 10'000 individuals). Since bone dysplasias are so rare, very little is known about them. For many disorders we still don't know what causes them, which medical problems we need to expect and how to treat them effectively. Studies on bone dysplasias are sparse and hampered by difficulties in recruiting sufficiently large numbers of patients and following them long-term.

To facilitate research on bone dysplasias, the Queensland Bone Dysplasia Registry collects information on patients with bone dysplasias and their families and makes it available to medical researchers. Information is collected through interviews with patients and family members, sending out questionnaires or reviewing existing medical records. The information collected by the registry is stored in an electronic database. The information collected by the registry can be accessed by doctors and researchers to study various aspects of bone dysplasias. Such studies will help improve our understanding of bone dysplasias and will eventually result in better treatment for patients.

B. What does participation in the study entail?

We ask for your permission to access your (or your child's) medical records. Any information considered important for research on bone dysplasias will be stored in the registry database. We might access your medical records in the future to update the information in the database. We may contact you in the future to update the information in our database, or to ask you to participate in future studies or to supply you with information relevant to your condition.



C. How long will I participate in this study?

Since one of the goals of the registry is to collect long-term information on patients with bone dysplasias and their families, we would like you to participate as long as possible. The information we have collected on you will be stored and used indefinitely unless you tell us to remove or destroy them. We might review your medical records in the future or contact you to update the information in our database.

D. Are there any risks in participating?

There is the theoretical risk that someone could gain unauthorized access to the registry database and could get access to your data. However, we will take all the necessary precautions to avoid unauthorized access (password protection, data encryption etc.).

E. Are there any benefits in participating?

Your participation in this study might help researchers to develop new tests or treatments for bone dysplasias or related disorders. It will also help to provide doctors and patients with more precise information about bone dysplasias and the best way to manage them.

The results of some of the research projects might have a direct benefit to you. The identification of a genetic defect in you or your family would allow us to give you a precise risk for recurrence of the bone dysplasia in other family members including your own children. It would also open the possibility of genetic testing during a pregnancy to find out if a child will be affected.

We might be able to enroll you into drug or other therapeutic trials which could give you access to new treatments for bone dysplasias.

If future studies identify preventive measures or treatments that can improve the quality of life of patients with certain bone dysplasias, we might be able to pass this information on to you and your doctors. We also plan to provide a newsletter with information on bone dysplasias to all interested participants.

F. What happens if I do not want to participate in this study or if I want to withdraw from the study later?

You are under no obligation to participate in this study. You can withdraw from the study at any time by contacting the Principal Investigator Dr Andreas Zankl at the address given on the first page of this info sheet. If you ask us to do so, we will destroy all data that we have stored for you. We will also contact all researchers that have accessed your data in the past and ask them to destroy them.

If you are a patient of a doctor involved in the registry, you can continue to see this doctor and will receive the same treatment as if you participated in the registry. If you feel there is a conflict of interests and you would prefer to see a different doctor, this can be arranged, too.

G. Will my data be treated confidentially?

During the course of the study, we will collect "identifiable" and "non-identifiable" data on you (or your child). Identifiable information refers to things such as name, address, phone number, medical

record number etc. It also includes full face photographs and comparable images. Non-identifiable information refers to things such as lab results or xrays after names or other identifiers have been removed.

All information is stored in a password-protected database that is only accessible to the registry team. We will share your non-identifiable information with other researchers who require this information for their studies. We will not share your identifiable information with other researchers without your permission.

Some studies involve publication of photographs of patients at medical conferences or in medical journals. Medical conferences are usually only attended by medical professionals and medical journals are usually only read by medical professionals and cannot be bought at new kiosks or bookshops. It is therefore unlikely that someone you know will recognize you on one of these photos. If you want to give us permission to publish photos of you at a medical conference or in a medical journal, please tick the appropriate box on the consent form. We will not publish (or allow other researchers to publish) any photographs that could identify you without your consent.

H. Will I get access to any results?

For practical reasons, we cannot guarantee that you will get access to the results of all studies that have utilised your data or materials. However, we will try to transmit results that are of practical relevance (identification of genetic defects, identification of risks for specific medical problems, improvements in treatment etc.) back to the study participants or their doctors. Some of these results could have far reaching implications for you and your family. Results could indicate that you are at risk for certain medical complications or that you are at risk for passing a genetic disorder on to your children. We assume that most participants would prefer to receive such results so that appropriate life style changes or medical interventions can be initiated if necessary. Please make sure you inform us of any changes in your address, phone number or email address so we will be able to contact you. If you do not wish to be informed of any test results, please let us know.

We will also inform study participants about study results that are of general interest through occasional newsletters. Please make sure to inform us of any changes in your address, phone number, or email address.

I. Will I get paid to participate in this study?

You will not be paid for participating in this study.

J. Who can I contact if I have any questions or problems in relation to this study?

If you have any questions or problems in relation to this study, please contact the Principal Investigator Dr Andreas Zankl. His contact details are given on the front page of this info sheet.

This study has been cleared by one of the human ethics committees of the University of Queensland in accordance with the National Health and Medical Research Council's guidelines. If you would like to speak to an officer of the University not involved in the study, you may contact the Ethics Officer on 3365 3924.



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Statement of Informed Consent for Data Collection

I have read and understood the information presented in the participant information sheet and consent to participating in this study. I authorise the registry to access my (or my child's) medical records and to store any relevant information in the registry database.

I understand that my (or my child's) data will be treated confidentially. Non-identifiable data will be shared with other researchers. Identifiable information (name, phone number, photographs etc.) should be treated as follows:

- I give permission to pass identifiable information on to other researchers who require this information for their studies.
- I give permission to publish photographs of me (or my child) at a medical conference or in a medical journal.
- I want to be contacted and asked for permission before my identifiable information is passed on to other researchers or published at a medical conference or in a medical journal.

I understand that I will not be paid to participate in this study and that I can withdraw from this study at any time without penalty.

Name of Participant

Date

Signature of Participant

Name of Guardian

Relationship to Participant

Date

Signature of Guardian