



Children's Hospital Boston

RESEARCH CONSENT FORM

Use Plate or Print:

Protocol Title: Molecular analysis of skeletal disease

MRN#:

DOB:

Principal Investigator: Matthew L. Warman, M.D.

Subject's Name:

Gender:

Why is this research study being conducted. What is its purpose?

We are performing a research study to better understand how genes affect the growth and maintenance of bones and joints. We also want to correlate changes in genes with specific signs and symptoms of skeletal disease. You (or your child) have been invited to participate in this research study because a member of your family has a skeletal disease.

Who is conducting this research study, and where is it being conducted?

Dr. Matthew L. Warman at Boston Children's Hospital is conducting this research study. Dr. Warman collaborates with other physicians and scientists who are at Boston Children's Hospital or at other medical centers and academic institutions.

How are individuals selected for this research study? How many will participate?

Individuals are invited to participate in this research study if they or their relatives have a skeletal disease for which there may be a genetic predisposition or cause. Dr. Warman and his collaborators expect to enroll more than 2000 participants with skeletal disease during the course of this study. Individuals who are contacted and asked to participate in this research are under no obligation to do so.

What may I be asked to do if I agree to participate in this research study?

If you (or your child) are personally affected with a skeletal disease we may ask you to provide:

- 1) Your (or your child's) medical information and/or records: You will be asked questions about yourself, your children, and other family members. You will be asked to give us permission to obtain from your health care provider(s) copies of your (or your child's) relevant medical records, including radiographs.
- 2) A blood, saliva, and/or urine sample: You (or your child) will be asked to provide a small amount of blood from a vein in your (or your child's) arm from which we can isolate your (or your child's) genetic material and also living cells. From children we will collect less than 3 teaspoonfuls of blood. From adults we will collect less than 6 teaspoonfuls of blood. Alternatively, you may be asked to provide a saliva sample or to rub a swab on the inside of your cheek, from which we can isolate smaller amounts of genetic material, but not living cells. You may also be asked to provide a urine sample, which we will use to look at substances made by bones and joints that are excreted in urine.
- 3) Tissue sample(s): If there is a tissue sample that is available from a previous procedure you have had, we may ask to have the sample shipped to us from its current location. During any future medically

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indicated procedure you may have, such as elective surgery, we may ask to receive tissue that would normally be removed as part of the procedure and then discarded.

If your relative has a skeletal disease, but you (or your child) personally have no signs or symptoms of skeletal disease, we may ask you to provide:

- 1) Your (or your child's) medical information and/or records: You will be asked questions about yourself, your children, and other family members. You will be asked to give us permission to obtain from your health care provider(s) copies of your (or your child's) relevant medical records, including radiographs.
- 2) A blood, saliva, and/or urine sample: You (or your child) will be asked to provide a small amount of blood from a vein in your (or your child's) arm from which we can isolate your (or your child's) genetic material and also living cells. From children we will collect less than 3 teaspoonfuls of blood. From adults we will collect less than 6 teaspoonfuls of blood. Alternatively, you may be asked to provide a saliva sample or to rub a swab on the inside of your cheek, from which we can isolate smaller amounts of genetic material, but not living cells. You may also be asked to provide a urine sample, which we will use to look at substances made by bones and joints that are excreted in urine.

What are the risks of this research study? What could go wrong?

Risks associated with collecting blood, saliva, or urine: You (or your child) may experience minor discomfort, bruising, or rarely dizziness or fainting as a result of having blood drawn. When possible, we will collect blood at the time of a medically indicated procedure so that you will not need to have blood drawn only for research purposes. There are no known risks associated with providing a saliva or cheek swab sample. There are no known risks associated with collecting urine.

There are no known risks of allowing us to study a previously obtained tissue sample or to retain a tissue sample that was obtained as part of a medically indicated procedure and would otherwise have been discarded.

Some people involved in genetic studies feel anxious about the possibility of carrying an altered gene that places them at risk or that may be passed on to children. If these feelings arise at any time during the study, you may contact us and we will arrange for you to speak with a genetic counselor.

In genetic research that involves testing multiple family members, it is possible that family information such as mistaken paternity and adoption may be discovered. If you wish, you may let us know in confidence if this is a possibility, since it may otherwise interfere with our analysis. In all cases, this information will be kept in the strictest confidence and will not be divulged to anyone.

You should also be aware that there might be social and economic disadvantages, which can be associated with the gathering of genetic information. You should understand that our testing might find an inherited defective gene, which puts you or a relative at risk for a genetic disorder in the future. Genetic information divulged to the wrong source, could affect you and your family if an insurance company or employer acquired this genetic information. We will do our best to keep all information confidential and only with your permission would we make this information available to others.

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What are the benefits of this research study?

There is no direct benefit to you or your family for participating in this study. However, we hope to gain information about the role(s) of specific genes in skeletal disease that will be of future benefit to skeletal health in humans.

Because we are a research laboratory and not a clinical laboratory with certified procedures for reporting patient results, we cannot directly release results from this study to you. If we obtain information that we think might be significant to your family (e.g., identification of a mutation that has caused the disease, disorder, or condition we are studying), we may be able to have these results confirmed by a CLIA-certified clinical laboratory. A CLIA lab is a lab that is authorized to release results from patient tests for clinical and diagnostic purposes. There will most likely be a charge associated with this testing, which will vary depending on the laboratory. Most CLIA laboratories will ask for fresh blood samples in order to ensure the accuracy of the results. If your results are confirmed, they will be reported to your physician and made available to you with proper genetic counseling. Please indicate below whether or not you wish to be informed if results become available. If you choose to be contacted, we can only do so through your own health care provider. Therefore, please provide the name of the healthcare provider we should contact to discuss making arrangements with a certified lab. We will make every reasonable effort to get in touch with the person you specify.

____ Please contact my health care provider if results become available in the future:

Physician's name: _____

Phone: _____

Address: _____

Are there costs associated with this research study? Will I receive any payments?

The study will be conducted at no cost to you or your insurance.

You will be compensated for parking, blood drawing, and sample shipping costs that you incur while participating in this study, if they are not covered by other sources. We can reimburse you up to \$100, but we will need the receipts for these expenses in order to be able to reimburse you.

During the research we may collect different samples from you (or your child) such as blood, urine or tissue, as described in the informed consent document. It is possible that what we learn or create from these samples, or the samples themselves, may be made available to other hospitals, universities and businesses, for further research. The research may also lead to new products, research tools, or inventions that are patented. If these lead to payments to Children's Hospital, the money the Hospital receives will be used to support biomedical research or provide healthcare to our patients, except that people who make the discoveries may receive some portion of the amount. We believe that devoting such payments to

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research and health care is the best way to benefit patients as whole, so we do not transfer those payments to research participants.

What will happen with the information obtained as part of this research study? What about confidentiality?

Any information about you or your family obtained from this research will be kept confidential. When results of this study are incorporated into a scientific report for publication the identification of those taking part is withheld.

The results of the tests performed for research purposes will not be placed in your (or your child's) medical record. In this manner it will be unlikely that others within the hospital, an insurance company or employer would ever learn of such results

If I do not want to take part in this research study, what are the other choices?

Participation in this research study is voluntary. Your (or your child's) only alternative to participating in this study is to not participate in this study.

What are my rights as a research participant?

Your participation in this research study is voluntary. Refusing to participate will not alter your usual health care or involve any penalty or loss of benefits to which you are otherwise entitled. If you decide to participate in this study, you may withdraw at any time and for any reason without penalty or loss of benefits. If information generated from this study is published or presented, your identity will not be revealed. In the event new information becomes available that may affect the risks or benefits associated with this study or your willingness to participate in it, you will be notified so that you can decide whether or not to continue participating. If you experience physical injury or illness as a result of participating in this research study, medical care is available at Boston Children's Hospital, or elsewhere; however, Boston Children's Hospital will not provide free care or compensation for lost wages.

Are there other things I should know about?

Request to store remaining samples

At the completion of this study, we would like to store any remaining sample for possible future use. The remaining samples may be stored indefinitely and may be used for future studies of genetic causes of your (or your relatives') disease. The samples will be stored in Dr. Warman's laboratory. Your sample will be given a unique identification number and stored without your name or other identifiers. Only the investigator will have a list to know which sample is linked to which patient/family member and this list will be kept confidential in a secure location. If the investigator distributes these samples, to other individuals who have an interest in the genetic causes of disease, it will be released with the unique identifier without any names or medical record numbers. If at any time you would like to have the

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sample removed from storage, please let us know and it will be transferred or destroyed according to your wishes.

What information do I need to know about the Health Insurance Portability and Accountability Act (HIPAA)?

During this research, information about your (or your child's) health will be collected. In general, under federal law, information about patients is private, but there are exceptions and you should know who will have access to this information and might see it.

Researchers may be collecting information about you or your child from medical records. They may also learn things from procedures that are part of the research itself such as tests, office visits, questionnaires and interviews.

The following people will be able to see this information:

- Medical and research staff at Children's Hospital, including people listed on your informed consent.
- Medical staff who are directly involved in your care that is related to the research or arise from it.
- People who oversee, advise or conduct research at Children's Hospital, and people who oversee or evaluate research and care, including the Committee on Clinical Investigation, staff working on quality improvement, and other clinicians and administrative staff of Children's Hospital.
- People from agencies and organizations that provide independent accreditation and oversight of research
- Sponsors or others involved in funding the research
- Federal agencies that oversee or review research information.
- Government agencies and sponsors.
- If some law or court requires us to share the information, we would have to follow that law or final ruling

You (or your child) should be aware that the federal privacy rule does not cover all of these possible uses. This means that once some of the above mentioned users receive your/your child's health information they do not have to follow the same rules. Other laws may or may not protect sharing of private health information. If you have a question about this you may contact the Children's Hospital Privacy Officer at 617-355-5502

There is no set time for destroying this information and no time limit for its use. Researchers continue to analyze data for many years and it is not possible to know when they will be done.

You (or your child) do not have to sign this form. If the form is not signed, however, you won't be able to participate in the study. Not signing will not affect your care or your child's care at Children's Hospital in any way now or in the future. Also, there will be no penalty or loss of benefits if you choose not to sign and participate.

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You (or your child) also have the right to withdraw from this study at any time. You have the right to end your permission for Children's Hospital to use or share the protected information about you or your child that was collected as part of the research.

Researchers may also continue to use information already collected to protect the integrity of the study. This means that your withdrawal won't make the whole study useless. Once you remove your permission and you (or your child) is no longer in the study, no more private health information will be collected. If you wish to withdraw you will need to do so in writing. Your investigator will have a form for you to use. If you (or your child) decide to share private information with anyone not involved in the study, the federal law designed to protect privacy may no longer apply to this information.

Although there are some legal limitations, you (or your child) have the right to get protected information resulting from this research that relates to your treatment or to payments. This information is available after the study analysis is done. To request the information, please contact the Hospital's Privacy Officer at 617-355-5502. If you have questions, please be sure to ask for answers.

Research at Children's Hospital: Children's Hospital has recently developed a web-based, interactive educational program for parents called "A Parent's Guide to Medical Research." To find out more about research at Children's Hospital, please visit the program at www.researchchildren.org

Children's Hospital is interested in hearing your comments, answering your questions and responding to any concerns regarding clinical research at Children's Hospital. If you would like further information about the type of clinical research performed at the hospital or have suggestions, questions or concerns regarding clinical research you may send an email to cci@childrens.harvard.edu or call 617 355-7052 between the hours of 8:30 and 5:00.

INVESTIGATOR'S AND/OR ASSOCIATE'S STATEMENT:

I have fully explained to _____ [participant/parent/guardian] the nature and purpose of the above-described procedures and the risks involved in its performance. I have provided the subject/family with the Privacy Rule if requested. I have answered and will answer all questions to the best of my ability. I will inform the participant of any changes in the procedures or the risks and benefits if any should occur during or after the course of the study. I have given a copy of the consent/authorization form to the subject/family.

Date (MM/DD/YEAR) Signature of **Investigator or Associate**

CONSENT/AUTHORIZATION:

If the child to be involved in this research study is a foster child or a ward of the state please notify the researcher or their staff who is obtaining your consent.

