UNIVERSITY OF CALIFORNIA, SAN FRANCISCO
CONSENT TO PARTICIPATE IN A RESEARCH STUDY

Study Title: MOLECULAR GENETIC ANALYSIS OF LIMB MALFORMATIONS

This is a medical research study. Your study doctor(s), Dr Nadav Ahituv from the Institute for Human Genetics and Dr Anne Slavotinek of Medical Genetics and the Division of Neurology in the Department of Pediatrics at the University of California, San Francisco (UCSF), will explain this study to you.

Medical research studies include only people who choose to take part. Take your time to make your decision about participating. You may discuss your decision with your family and friends and with your health care team. If you have any questions, you may ask your study doctor.

You are being asked to take part in this study because you or one of your close relatives have limb malformations. We would like to ask you to participate in this study so that we can try to find out the cause for limb malformations.

Why is this study being done?

The purpose of this study is to learn more about the genetic causes of limb malformations in humans. The genetic cause(s) for several of these conditions is not known at present or incompletely known.

This study is sponsored by UCSF from funds of the Department of Biopharmaceutical Sciences and the Institute for Human Genetics.

How many people will take part in this study?

About 300 people per year will take part in this study.

What will happen if I take part in this research study?

- If you agree to be in this study, a blood sample or a saliva sample, if you do not want a blood sample to be drawn, will be used to obtain the genetic material for laboratory testing. The blood sample will be drawn by inserting a needle into a vein in your arm. About three ounces of blood will be drawn from adults (8-10 teaspoons) and two ounces (5-7 teaspoons) from adolescents depending on weight. This blood will be drawn at UCSF or by your physician. The blood draw will take about five minutes. A permanent cell line may also be made from the blood sample so that more research tests can be done in the future. Repeat studies may be needed in some cases and you may be asked to give another blood sample in the future. However, you may choose not to donate the second sample. If a saliva sample will be taken, you will spit saliva into the Oragene kit container provided. DNA samples and cell lines will be housed at UCSF in the lab of Dr. Nadav Ahituv and will only be used for research into limb malformations.

- If you take part in this study, medical information regarding limb malformations regarding either you or your family may be collected by the study investigators. We may publish a chart that shows your family tree and who is affected with the condition, but we will not use your family's name. If you have a unique family, others may still be able to recognize the family tree, but we will use the smallest amount of information possible in order to make the family

tree less recognizable. We may also publish photographs of the limb malformation without showing your face. However, we must first discuss this with you and obtain your written permission. If we determine that it would be helpful to enroll other family members in this study, we will contact them only with your assistance and their permission.

- Material (DNA) taken from your blood sample may be given to other researchers for other studies on the same or similar conditions approved by UCSF. However, your name will not be given to the other researchers. On completion of the study, the DNA samples and cell lines will be stored indefinitely at UCSF in the lab of Dr. Nadav Ahituv or at the institution where the work was carried out. The samples will not be given to other researchers for studies on different medical conditions. If you decide later that you do not want your sample and information to be used for future research, you can tell us by contacting Dr Nadav Ahituv, principal investigator of the study, either by phone, 415 476 1838, or by e-mail (nadav.ahituv@ucsf.edu), and we will destroy any remaining identifiable sample.

Can I stop being in the study?

Yes. You can decide to stop at any time. Just tell the study doctor you want to stop by contacting Dr. Nadav Ahituv at 415 476 1838 or nadav.ahituv@ucsf.edu , or Dr. Anne Slavotinek at (415) 514-1783 or page her on (415) 719-8473.

What side effects or risks can I expect from being in the study?

Drawing blood may cause temporary discomfort from the needle stick, bruising, and very rarely infection.

Confidentiality: Donating specimens may involve a loss of privacy, but information about you will be handled as confidentially as possible. Your name will not be used in any published reports from research performed using your specimen. Dr. Ahituv and his staff members will have access to information about you but they will not release any identifying information about you to researchers using your specimen. The UCSF Committee on Human Research and other University of California personnel also may see information about you to check on the tissue bank. Once your health information is disclosed to the research team it is not protected under the Health Information Portability and Accountability Act (HIPAA). The tissue bank staff will continue to protect your personally identifiable health information as described in this consent form. The University of California complies with the requirements of HIPAA and its privacy regulations, and with all other applicable laws that protect the confidentiality of your health information.

Are there benefits to taking part in the study?

There will be no direct benefit to you from participating in this study. However, this study will help doctors learn more about the genetic cause of limb malformations, and it is hoped that this information will help in the treatment of future patients with limb malformations. In addition, the results may be used for the development of tests, products, or discoveries that may have potential commercial value but you will not be paid for taking part in this study.

What other choices do I have if I do not take part in this study?

You are free to choose not to participate in the study. If you decide not to take part in this study, there will be no penalty to you. You will not lose any of your regular benefits, and you can still get your care from our institution the way you usually do.

Will my medical information be kept private?

We will do our best to make sure that the personal information in your medical record is kept private. However, we cannot guarantee total privacy. Your personal information may be given out if required by law. If information from this study is published or presented at scientific meetings, your name and other personal information will not be used.

The study team that may look at and/or copy your medical records for research, quality assurance, and data analysis include Dr. Nadav Ahituv and his lab members, Dr. Anne Slavotinek, and UCSF's Committee on Human Research.

Participation in research may involve a loss of privacy, but information about you will be handled as confidentially as possible. A medical record will be created because of your participation in this study. Your consent form and some of your research test results will be included in this record. Therefore, your other doctors may become aware of your participation. Hospital regulations require that all health care providers treat information in medical records confidentially. The paper research records and photocopies of medical records and patient correspondence will be stored in a locked drawer or in a password-controlled computer file in a locked office. Additional information containing patient identifiers such as copies of medical letters, correspondence related to patient inclusion in the protocol and email transcripts will also be stored under the same secure conditions. We will code laboratory specimens upon their arrival at the UCSF laboratory. The PI will keep the key to the coding that will be kept in a locked drawer in a locked laboratory or office. The key may also be stored in a password-protected computer file. The paper research records and clinical information files will be kept for the duration of the study. At the time that the study is closed, a decision will be made whether to keep the data or to destroy it.

What are the costs of taking part in this study?

You will not be charged for any of the study treatments or procedures. All costs for this study including the blood draw will be covered by Dr. Nadav Ahituv.

Will I be paid for taking part in this study?

You will not be paid for taking part in this study.

What happens if I am injured because I took part in this study?

It is important that you tell your study doctor, Dr. Nadav Ahituv, if you feel that you have been injured because of taking part in this study. You can tell the doctor in person or call him at 415 476 1838.

> **Treatment and Compensation for Injury:** If you are injured as a result of being in this study, treatment will be available. The costs of the treatment may be covered by the University of California, depending on a number of factors. The University does not normally provide any other form of compensation for injury. For further information

about this, you may call the office of the Committee on Human Research at 415- 476-1814.

What are my rights if I take part in this study?

Taking part in this study is your choice. You may choose either to take part or not to take part in the study. If you decide to take part in this study, you may leave the study at any time. No matter what decision you make, there will be no penalty to you and you will not lose any of your regular benefits. Leaving the study will not affect your medical care. You can still get your medical care from our institution.

We will tell you about new information or changes in the study that may affect your health or your willingness to continue in the study.

In the case of injury resulting from this study, you do not lose any of your legal rights to seek payment by signing this form.

Who can answer my questions about the study?

You can talk to your study doctor about any questions or concerns you have about this study. Contact your study doctor Nadav Ahituv at 415 476 1838 or nadav.ahituv@ucsf.edu , or Dr. Anne Slavotinek at (415) 514-1783 or page her on (415) 719-8473.

For questions about your rights while taking part in this study, call the office of the **Committee on Human Research**, UCSF's Institutional Review Board (a group of people who review the research to protect your rights) at **415-476-1814**.

****

CONSENT

Please read each sentence below and think about your choice. **After reading each sentence, put your initials in the "Yes" or "No" box**. If you have any questions, please talk to your doctor or nurse, or call our research review board at IRB's phone number.

No matter what you decide to do, it will not affect your care.

1. A blood or saliva sample will be taken for this study.

YES	NO

2. My tissue may be kept for use in research to learn about, prevent, or treat limb malformations.

YES	NO

3. Photographs and X-rays may be taken for this research, and may be used for publication in a medical journal.

YES	NO

4. I may be contacted regarding participation in future studies.

YES	NO

You have been given copies of this consent form and the Experimental Subject's Bill of Rights to keep.

You will be asked to sign a separate form authorizing access, use, creation, or disclosure of health information about you.

PARTICIPATION IN RESEARCH IS VOLUNTARY. You have the right to decline to participate or to withdraw at any point in this study without penalty or loss of benefits to which you are otherwise entitled.

If you wish to participate in this study, you should sign below.

_____ _____
Date Participant's Signature for Consent

_____ _____
Date Witness's Signature (only required if the participant is a non-English speaker)

_____ _____
Date Person Obtaining Consent