

18 years of age or over: Consent only

## The Ohio State University Consent to Participate in Research

**Study Title:** Genetics, Molecular Diagnostics, and Mechanisms of Juvenile Cobalamin Deficiency

**Principal Investigator:** Stephan Tanner, PhD

**Sponsor:**

- **This is a consent form for research participation.** It contains important information about this study and what to expect if you decide to participate. Please consider the information carefully. Feel free to discuss the study with your friends and family and to ask questions before making your decision whether or not to participate.
- **Your participation is voluntary.** You may refuse to participate in this study. If you decide to take part in the 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 usual benefits. Your decision will not affect your future relationship with The Ohio State University. If you are a student or employee at Ohio State, your decision will not affect your grades or employment status.
- **You may or may not benefit as a result of participating in this study.** Also, as explained below, your participation may result in unintended or harmful effects for you that may be minor or may be serious depending on the nature of the research.
- **You will be provided with any new information that develops during the study that may affect your decision whether or not to continue to participate.** If you decide to participate, you will be asked to sign this form and will receive a copy of the signed form. You are being asked to consider participating in this study for the reasons explained below.

### 1. Why is this study being done?

This study is being done to better understand the genetic causes of juvenile cobalamin (vitamin B12) deficiency (JCD).

### 2. How many people will take part in this study?

Approximately 500 people will take part in this study.

### 3. What will happen if I take part in this study?

Upon enrollment in this study, you will be asked to provide demographic, family history information and release your personal medical history information relating to your diagnosis of juvenile cobalamin deficiency (JCD). You may also choose to provide

contact information for your health care provider, so that we may follow-up with results of this study.

To participate in this study, you will need to provide a blood or cheek cell sample. If you choose to donate blood, a small needle will be inserted into a vein in your arm and about 4 tablespoons of blood will be taken. If you are not able to meet with us at Ohio State University Medical Center to have your blood drawn, a blood kit will be sent to you via FedEx. You will need to have blood drawn by a certified phlebotomist at your physician's office. The specimen will need to be packaged and sent to us using a provided postage-paid FedEx envelope. You will not be required to pay for shipping of the sample, however, you may need to pay a small fee for blood drawing (usually \$10-\$20).

If you are uncomfortable providing a blood sample, you can provide a cheek cell sample by donating a saliva sample, buccal swab sample, or mouthwash sample. Donating saliva involves spitting into a small cup until the desired amount is obtained (typically about 2 tablespoons, which takes approximately 1 minute). Donating a buccal swab sample involves rubbing a soft brush on the inside of your cheek, and donating a mouthwash sample involves rinsing your mouth with a mouthwash (like Scope) and spitting it back into a collection cup. These samples can be collected at OSUMC or at home with a provided kit, and sent back to us at our expense. You will need to let us know which option you prefer.

Your sample will be sent to a laboratory at The Ohio State University, where DNA/RNA (genetic material) will be removed from the cells. Your genetic code will be analyzed for changes in certain genes (called GIF, AMN, CUBN). Changes in these genes have been associated with juvenile cobalamin (vitamin B12) deficiency (JCD). The sample will also be used for research to identify other genes that may be associated with JCD. Samples will be destroyed 3 years after the completion of this study.

You have the option to receive genetic test results from this study. If you would like to be contacted by a genetic counselor with your test results, you must specify this on the consent form (#13). You will have time to discuss questions or concerns with the counselor over the phone, or in person, if you prefer. You, and/or other family members will also be given the option to participate in this research study and receive results.

#### **4. How long will I be in the study?**

Your participation in this study involves a one-time donation of blood or saliva, which should only take a few minutes. However, the DNA/RNA extracted from your sample will be used in research over the next several years, and may not be discarded for up to 3 years after research is complete. We estimate that research will be complete in the year 2012, so samples will be kept until 2015. Future use of the data or specimens will be restricted to this study and not used for other, unspecified research projects.

You have the choice whether or not to receive your test results from this study. Results should take approximately 2-6 months.

#### **5. Can I stop being in the study?**

You may leave the study at any time. You will only need to submit a request, in writing, to study investigators. If this happens, your sample and all related information will be destroyed. If you decide to stop participating in the study, there will be no penalty to you, and you will not lose any benefits to which you are otherwise entitled. Your decision will not affect your future relationship with The Ohio State University.

#### **6. What risks, side effects or discomforts can I expect from being in the study?**

You may experience some physical discomfort when your blood is drawn. You may have some slight bruising at the site of needle insertion and there is a small chance of infection. There are no known risks for cheek cell collection.

Some individuals have concerns regarding confidentiality of genetic testing and/or the potential for discrimination based on genetic test results. All efforts will be made to keep your test results confidential; results will be released only to you and your health care provider (if requested). There are federal and state laws to protect you from health insurance discrimination based on genetic information. This protection extends to all group health care plans (HMP, PPO, etc), but does not include private health insurance plans. There are no protections for life insurance or disability discrimination. While discrimination related to genetic testing is possible, very few documented cases of any type of discrimination based on genetic information currently exist.

Research genetic testing should not be used for treatment decisions until results are confirmed in a certified (CLIA approved) laboratory. When and if you are informed of the results from research testing, we will let you know if CLIA approved laboratory testing is available.

#### **7. What benefits can I expect from being in the study?**

Participation in this study may be beneficial to you, and/or family members, as it may help to identify the genetic mutations in your family that predispose you, or family members to JCD. A better understanding of these genetic changes will assist researchers in developing a clinical genetic test. A clinical genetic test could be used to identify individuals in a family at increased risk to inherit JCD, and would enable early treatment to prevent complications from this disease.

#### **8. What other choices do I have if I do not take part in the study?**

You may choose not to participate without penalty or loss of benefits to which you are otherwise entitled. If your doctor suspects you have JCD, you will receive the usual

treatment for this condition regardless of your participation in this study. Other options for genetic testing for JCD do not currently exist. You may also elect to participate in this study without receiving genetic test results (specify preference under #13).

### **9. Will my study-related information be kept private?**

Efforts will be made to keep your study-related information confidential. Only investigators and co-investigators will have access to research data. All records related to the study will be stored in locked file cabinets. Laboratory specimens will be coded and stored in the laboratory of a co-investigator at The Ohio State University. Computer files with information will be kept locked with restricted access passwords.

However, there may be circumstances where this information must be released. For example, personal information regarding your participation in this study may be disclosed if required by state law. Also, your records may be reviewed by the following groups (as applicable to the research):

- Office for Human Research Protections or other federal, state, or international regulatory agencies;
- U.S. Food and Drug Administration;
- The Ohio State University Institutional Review Board or Office of Responsible Research Practices;
- The sponsor supporting the study, their agents or study monitors; and
- Your insurance company (if charges are billed to insurance).

If the study involves the use of your protected health information, you may also be asked to sign a separate Health Insurance Portability and Accountability Act (HIPAA) research authorization form.

### **10. What are the costs of taking part in this study?**

You may be responsible for the cost of getting your blood drawn. This fee is typically \$10-\$20 depending on the physician office. You may request a cheek cell collection kit if you prefer, in order to avoid the phlebotomy fee. You will not be responsible for the cost of the cheek cell kit or blood kit, shipping of the sample, or cost of research genetic testing or counseling.

### **11. Will I be paid for taking part in this study?**

You will not be paid to participate in this study.

### **12. What happens if I am injured because I took part in this study?**

There is minimal risk involved with participation in this study, since it only requires a blood draw or donation of saliva. If you suffer complications as a result of your blood

draw, you should notify your doctor immediately, who will determine if you should obtain medical treatment.

The cost for this treatment will be billed to you or your medical or hospital insurance. The Ohio State University has no funds set aside for the payment of health care expenses for this study.

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

If you choose to participate in the study, you may discontinue participation at any time without penalty or loss of benefits. Research subjects who are minors when they enter the study will have the option of being re-consented once they reach adulthood or request that their specimen and associated data be discarded.

You will be provided with any new information that develops during the course of the research that may affect your decision whether or not to continue participation in the study. You may refuse to participate in this study without penalty or loss of benefits to which you are otherwise entitled.

An Institutional Review Board responsible for human subjects research at The Ohio State University reviewed this research project and found it to be acceptable, according to applicable state and federal regulations and University policies designed to protect the rights and welfare of participants in research.

You may choose whether or not you want to receive the results of your genetic testing (see check boxes below). If you choose to receive genetic test results, they will be communicated to you by a genetic counselor in person, or by telephone, when they become available. Research genetic test results should not be used for clinical management until they are confirmed in a CLIA-approved laboratory. Currently, clinical testing through a CLIA-approved laboratory is available through GeneDx. The cost of clinical confirmation testing will not be covered by the research study.

If you choose not to have your test results released, they will be kept confidential in your research chart at Ohio State University and stored in a password protected database.

**Would you like to receive results of your genetic testing once it is available?**

Yes     No \_\_\_\_\_ (initial and date)

If you would also like your health care provider to be contacted with results, please provide his/her name, phone number and address of your health care provider:

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\_\_\_\_\_  
\_\_\_\_\_

**14. Who can answer my questions about the study?**

For questions, concerns, or complaints about the study you may contact Amy Sturm at (614) 293-6694.

For questions about your rights as a participant in this study or to discuss other study-related concerns or complaints with someone who is not part of the research team, you may contact Ms. Sandra Meadows in the Office of Responsible Research Practices at 1-800-678-6251.

If you are injured as a result of participating in this study or for questions about a study-related injury, you may contact Amy Sturm at (614) 293-6694 or 1-888-329-1654 (toll free).

**15. Signing the consent form**

I have read (or someone has read to me) this form and I am aware that I am being asked to participate in a research study. I have had the opportunity to ask questions and have had them answered to my satisfaction. I voluntarily agree to participate in this study.

I am not giving up any legal rights by signing this form. I will be given a copy of this signed form.

<b>Printed name of subject</b>	<b>Signature of subject</b>
	_____ <b>AM/PM</b>
	<b>Date and time</b>
<b>Printed name of person authorized to consent for subject (when applicable)</b>	<b>Signature of person authorized to consent for subject (when applicable)</b>
	_____ <b>AM/PM</b>
<b>Relationship to the subject</b>	<b>Date and time</b>

**Investigator/Research Staff**

I have explained the research to the participant or his/her representative before requesting the signature(s) above. There are no blanks in this document. A signed copy of this form has been given to the participant or his/her representative.

<b>Printed name of person obtaining consent</b>	<b>Signature of person obtaining consent</b>
	_____ <b>AM/PM</b>
	<b>Date and time</b>

**Witness(es) - May be left blank if not required by the IRB**

<b>Printed name of witness</b>	<b>Signature of witness</b>
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**CONSENT**

**IRB Protocol Number: 2005H0201**

**IRB Approval date: 10/31/07**

**Version: 6**

**AM/PM**

\_\_\_\_\_  
**Date and time**

\_\_\_\_\_  
**Printed name of witness**

\_\_\_\_\_  
**Signature of witness**

**AM/PM**

\_\_\_\_\_  
**Date and time**