

## **ROHHAD Biomarkers Study**

The Center for Autonomic Medicine in Pediatrics (CAMP) at Lurie Children's is working with a group of international collaborators with expertise in genetics to identify gene(s) or biomarkers which may contribute to or cause ROHHAD (rapid-onset obesity with hypothalamic dysfunction, hypoventilation and autonomic dysregulation).

The purpose of this IRB-approved study is to gain a better understanding behind ROHHAD. The study aims to recruit patients from around the world. This project will go a long way toward improving diagnosis and care of ROHHAD patients. We appreciate your help.

### **Participate in the Study**

Participation in this study will require samples of blood, saliva, buccal, dental, and/or tumor tissue and signed consent form(s). Please see below for further details about specimen preparation and consent forms.

### **Blood or DNA Sample**

We are collecting small blood sample or DNA sample from both patients with ROHHAD and their family members, especially the parents of ROHHAD patients. Parental samples will allow us to determine if something we identify in a ROHHAD patient is inherited, and thus NOT likely a factor in ROHHAD, or if it is de novo (which means a new variation in the DNA, not inherited, and thus much more likely to play a role in ROHHAD). In addition to patient DNA and parental DNA, we will also need DNA of any available siblings.

We will need consent and a blood or DNA sample from each participant. Please see consents at the end of this document.

### **Specimen Preparation:**

For the ROHHAD patient:

9 ml of blood (3 mL in a lavender/purple-top EDTA-coated vacutainer and 6 mL in a red silicone-coated vacutainer) is preferred.

For the parents/siblings:

3-6 ml of blood (in a lavender/purple-top EDTA-coated vacutainer) is preferred.

Please prepare and ship the samples immediately or place the tubes into a refrigerator if immediate shipping is not possible. DO NOT FREEZE. Because of transportation issues, it is best not to ship blood on Friday, Saturday, or Sunday.

- Please be sure to label tubes carefully and clearly
- Please place tubes containing blood into plastic 50 ml tubes (or similar) and screw cap on tightly. This will protect the tube while in transit.
- Place this tube into a plastic bag with absorbent paper towels (or similar) and seal the plastic bag.
- Place plastic bag back into sturdy shipping box and seal with tape.
- Signed consent forms and other information should be included.
- Shipping can take place at ambient temperature.

You may schedule a pickup using Fedex.com, or simply drop the container off at your local FedEx office or similar courier.

Ship overnight or using 2-3 day priority delivery to the address below:

Rush University Medical Center  
Section of Genetics  
Attention: Lili Zhou-Casey Rand  
1750 W. Harrison St. Rm. 1501 Jelke  
Chicago, IL 60612  
Phone: 312.942.6298

If shipping DNA samples, we ask for as much DNA that is available or a minimum of 300  $\mu\text{L}$  of DNA at a concentration of 300  $\text{ng}/\mu\text{L}$  from 2  $\mu\text{g}/\mu\text{L}$  of blood. DNA samples can be shipped at ambient temperature to the address listed above.

### **Saliva Samples**

We are collecting saliva samples only from the parents and siblings of patients with ROHHAD. DNA samples from family members will allow us to determine if a genetic marker we identify in a ROHHAD patient is inherited, and thus NOT likely a factor in ROHHAD, or if it is de novo (which means it is a new variation in the DNA, not inherited) and thus much more likely to play a role in ROHHAD development. Email [CAMPResearch@luriechildrens.com](mailto:CAMPResearch@luriechildrens.com) to express your interest in contributing one or more saliva samples and we will send collection kit to your mailing address. Please ship the sample/s to the address below:

Rush University Medical Center  
Section of Genetics  
Attention Lili Zhou-Casey Rand  
1750 W. Harrison St. Rm. 1501 Jelke  
Chicago, IL 60612  
Phone 312-942-6298

### **Cord Blood**

We are collecting cord blood from ROHHAD patients (blood from the umbilical cord). This may have been stored at the hospital where the ROHHAD patient was born. Please call the hospital if you are unsure. We are looking at differences in the blood prior and after disease onset to see any differences can be found. This study will involve sending us any of the banked or stored cord blood of ROHHAD patients.

Please contact CAMP at [CAMPResearch@LurieChildrens.org](mailto:CAMPResearch@LurieChildrens.org) or 312.227.3300 regarding specimen preparation of cord blood.

## **Tumor Tissue**

We are also collecting tumor tissue. When tumor tissue is removed, it is often banked/stored for future analysis. If tumor tissue is stored, participation in this study will require sending us the tumor tissue from ROHHAD patients so that our team of experts may analyze the tumor.

### **Specimen Preparation:**

Frozen tumor tissue is preferred. Please prepare and ship the frozen tissue immediately. Please be sure to label all tubes and provide information on tissue type, source, and date of extraction in paperwork.

### **Frozen tissue:**

If surgery to remove tissue is being performed at this time:

- 1) Place frozen tissue into cryotube and seal lid.
- 2) Place frozen tube in biohazard bag
- 3) Place 2-3 inches of dry ice in the bottom of the Styrofoam cooler, place biohazard bag in the center of the cooler on top of the dry ice, then fill the cooler with dry ice (preferably pelleted).
- 4) Place a single paper-towel or piece of paper across the top of the ice, place lid on the cooler and tape the lid tightly to the cooler, sealing all the way around the lid.
- 5) Place the cooler in the cardboard box with all paperwork associated with the case on top of the cooler and seal the box.
- 6) Ship overnight to the address below.

If frozen tissue has previously been removed:

Follow steps 2-6 above.

### **Fixed tissue:**

- 1) Fixed tissue should be placed into a sealed tube for shipment
- 2) Place sealed tube into sealed bag
- 3) Place bag into secure shipping container
- 4) Ship to the address below (does not need to ship overnight)

Rush University Medical Center  
Section of Genetics  
Attention Lili Zhou-Casey Rand  
1750 W. Harrison St. Rm. 1501 Jelke  
Chicago, IL 60612  
Phone 312.942.6298

We will accept fixed tumor tissue if that is the only one available. Fixed tumor tissue can be shipped at ambient temperature to the address above.

## **Buccal Sample**

We are collecting buccal samples. This involves swabbing the inside of the cheek of ROHHAD patients. Buccal collection kits will be sent to your mailing address. Please complete and ship to the address below:

Rush University Medical Center  
Section of Genetics  
Attention Lili Zhou-Casey Rand  
1750 W. Harrison St. Rm. 1501 Jelke  
Chicago, IL 60612  
Phone 312.942.6298

### **Dental Sample**

We are collecting dental samples from ROHHAD patients that are losing baby teeth soon or are getting any teeth removed. Each sample must be processed for shipping by being placed into a tube of tube media within 30 minutes of falling out/being removed. Please contact us CAMP beforehand at [CAMPResearch@LurieChildrens.org](mailto:CAMPResearch@LurieChildrens.org) or 312.227.3300 to allow for a dental sample collection kit, with more detailed instructions, to be sent to your mailing address ahead of time.

### **Consents**

For any samples (blood, DNA, cord blood, buccal, dental, tumor tissue) from the ROHHAD patient, we will need a signed consent(s).

#### **For the ROHHAD patient:**

- 1) [Subject Adult Proband Consent](#) – should be signed individually by the patient if the patient is 18 years or older
- 2) [Subject Parent of Proband Consent](#) – should be signed by parents for the patient if the patient is under the age of 18 years old
- 3) [Subject Adolescent Assent](#) – should be signed by the patient if the child is between the ages of 12-17 years, this is done in addition to the parent consent.

For any family members sending samples (blood or DNA), we will need a signed consent for each member of the family.

#### **For the parents and/or siblings (if sending parent/sibling blood or DNA samples):**

- 1) [Control Adult Consent](#) – should be signed individually for each participating parent or sibling (only if the sibling is 18 years or older). Each parent and/or sibling will need a separate consent.
- 2) [Control Parent Consent](#) – should be signed by parents for each participating sibling under the age of 18 years old. One consent will need to be signed for each child.
- 3) [Control Adolescent Assent](#) – should be signed by each sibling that is between the ages of 12-17 years, this is done in addition to the parent consent.

If you have any questions, please contact CAMP at Lurie Children's at [CAMPResearch@LurieChildrens.org](mailto:CAMPResearch@LurieChildrens.org) or 312.227.3300 with any questions and to review the consents. Once signed, please scan and email, fax, or mail the consents to CAMP. Thank you very much for considering participation in this project.