Simons VIP: Skin Biopsy Information for Families

What is a skin biopsy?
A skin biopsy is the removal of a small piece of skin.

Who is asked to provide a skin biopsy sample?
Each individual participating in the Simons VIP study with either a 16p11.2 deletion or 16p11.2 duplication will be asked to consider providing a skin sample.

Why is this study collecting skin samples?
Investigators would like to be able to study brain cells in individuals with 16p11.2 deletions and 16p11.2 duplications to understand why the brain functions differently. However, it is not possible to directly study brain cells in a living person. Advances in science now allow us to take skin cells and make them into brain-like cells. This process requires first making the skin cells into immature cells called induced pluripotent stem cells (iPS cells). The induced pluripotential stem cells can then be made into brain cells.

In this way, we can take skin cells from individuals with 16p11.2 deletions and 16p11.2 duplications and make brain cells that can be studied in the laboratory. Scientists can then try to understand if and how those brain cells work differently. Ultimately, this could give us important clues about how to treat individuals with 16p11.2 deletions and 16p11.2 duplications.

How is the skin biopsy performed?
In order to get the cells we need, a punch skin biopsy is done. A punch biopsy takes a small round piece of skin 3 mm in diameter. See the picture below for an example.

What will happen during the skin biopsy?
The doctor performing the biopsy may ask you in advance about any allergic reactions you have had to anesthetic medications, about any blood thinner or anticoagulant medications you may be taking, or any problems with bleeding in the past. If you are pregnant, we will not perform the skin biopsy. Typically, there are no conditions that prevent physician from performing a skin biopsy, but these questions help the doctor anticipate and better manage any potential complications (eg, excess bleeding). Otherwise, there is no special preparation you need to take prior to the procedure.
The area of the skin biopsy is cleansed with alcohol or antiseptic solution, and local anesthetic (numbing medicine) is injected at the site of the biopsy. The doctor will wait about 2 minutes for the numbing medicine to take effect. The area will be cleaned thoroughly. Using sterile instruments, the doctor takes a small round sample of skin, and the tissue is removed. The site may bleed, so the doctor will apply pressure with gauze. The doctor may use a medicine to help stop the bleeding. Antibacterial ointment and a small dressing are placed over the site.

You will then keep the area clean and dry for the next 5 days. After that, it may be gently cleaned with soap and water. Applying antibacterial ointment may be done on a daily basis until the skin heals.

**Where will the biopsy be taken from?**
The site of the skin biopsy is largely up to you. Many patients prefer having the biopsy done on the inner arm or upper thigh.

**Will the biopsy leave a scar?**
The biopsy site could have a colorless scar that looks like this:

![Image of a colorless scar on skin]

**Are there any risks from doing the skin biopsy?**
With the skin biopsy there may be bleeding, pain with injection of the numbing medicine, infection, or a small, colorless scar. The skin biopsy will be performed by an experienced doctor to minimize these risks.

**My child will be having surgery. Can we get a skin sample then?**
For most surgical procedures in which an incision is made in the skin, a small piece of skin can be collected at the incision site and does not affect the wound healing or add risk of infection. The skin removed from a circumcision can also be used by our study. We can coordinate having these skin samples submitted to the study if surgical procedures are planned.

**I have more questions, who can I talk to?**
Our staff is happy to talk to you about the skin biopsy procedure and your options for this study. Please contact us!
Website: [www.simonsvipconnect.com](http://www.simonsvipconnect.com)
E-mail: Coordinator@SimonsVIPConnect.com
Phone: 888-493-6682

Principle Investigator: Dr. Wendy Chung, (212) 851-5313, wkc15@columbia.edu

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<tr>
<th>If you are going to</th>
<th>Contact</th>
<th>E-mail</th>
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</thead>
<tbody>
<tr>
<td>Boston (Children’s Hospital Boston)</td>
<td>Dr. Ellen Hanson</td>
<td><a href="mailto:Ellen.Hanson@childrens.harvard.edu">Ellen.Hanson@childrens.harvard.edu</a></td>
</tr>
<tr>
<td>Houston (Baylor College of Medicine)</td>
<td>Dr. Robin Kochel</td>
<td><a href="mailto:kochel@bcm.edu">kochel@bcm.edu</a></td>
</tr>
<tr>
<td>Seattle (University of Washington)</td>
<td>Dr. Raphe Bernier</td>
<td><a href="mailto:rab2@u.washington.edu">rab2@u.washington.edu</a></td>
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