DEPARTMENT OF HEALTH AND HUMAN SERVICES Public Health Service

National Human Genome Research Institute
National Institutes of Health
10 Center Drive
Building 10, Room 3C710
Bethesda, MD 20892
Tel. (301) 435-2832, Fax (301) 435-3495

Questions and Answers about NIH Study

Who are the researchers conducting the study?

This study is being conducted by a group of researchers and clinicians at the National Human Genome Research Institute at the National Institutes of Health (NIH) who have been studying overgrowth conditions for over 10 years. This study is headed by Dr. Leslie Biesecker, a medical geneticist. You can learn more about Dr. Biesecker's research by visiting http://www.genome.gov/10000356.

What is the goal of the study?

Our protocol began with the study of Proteus syndrome, a very rare overgrowth condition. Over the years, we have learned a lot about Proteus syndrome, including how it affects different people over time, some specific health concerns patients and doctors should be aware of, and what the most effective treatments for it are. We now hope to learn more about other overgrowth conditions so that we can improve doctors' ability to diagnose and treat those conditions as well. We also hope to learn more about the genetic causes of overgrowth conditions.

Who is eligible to participate in the study?

If you or your child has an overgrowth condition, such as CLOVES syndrome, hemihypertrophy or hemihypertrophy multiple lipomatosis (HHML), fibroadipose overgrowth, or macrodactyly, you (or your child) may be eligible to participate. We usually ask for the a letter from a doctor describing a person's condition along with photographs of the affected parts of the body to evaluate eligibility.

What does participation involve?

Participation in this study is entirely voluntary. The study has a clinical arm, where patients and families are asked to come the NIH for an in-person evaluation, and a laboratory arm, which involves sending specimens like blood and tissue samples to us for genetic testing. Which arm of the study any individual family joins depends on the patient's presentation, the family's goals, and other factors. Participation in the study is free and we can cover most of the costs associated with participation, including shipment of specimens, and travel, lodging, and food expenses associated with an NIH visit.

For laboratory arm participants, we will ask for a blood sample and a tissue sample (most often a skin biopsy) to be sent to us for genetic testing. We will test this sample for gene changes known or suspected to cause overgrowth and provide participants with our results. We will invite some participants to come to

the NIH for an in-person evaluation (this is the clinical arm of the study). Each NIH evaluation is tailored to the individual patient but typically includes evaluations with geneticists, dermatologists, and orthopedic surgeons and imaging studies such as ultrasounds and x-rays. A member of our research team will always review our procedures and the consent form for the study with you before you decide to participate.

What kind of results might I learn as part of this study?

We hope to learn more about overgrowth conditions by studying a few specific genes known or believed to play a role in causing overgrowth. Because of this, genetic testing of DNA from blood and/or tissue samples is a major part of our study. If we are able to find a gene change that we believe caused your or your child's overgrowth, we will verify this result in a clinical laboratory and share our findings with you. You can choose to visit the NIH to get your results in person, or, we can work with you to find a genetics specialist in your area who can review the results with you. We think it is important to talk with someone in person about any results, and so we will not give you your results over the phone. This process can take months to years to complete, but we will always notify you if any results do become available.

What are the risks and benefits to participating in this study?

Some physical risks associated with being in this study include complications that can result from a blood draw or skin biopsy, such as pain and infection. Some of the procedures and evaluations people in the clinical arm of the study may be asked to consider may have risks, such as the risk of radiation exposure from x-rays. Some people find that learning about their genetic information is upsetting or worry about other people learning this information. We take precautions to minimize these kinds of risks. For example, we take your privacy and confidentiality very seriously and we will not share any information about you with anyone else without your express permission. While we hope that this study will increase our understanding of overgrowth conditions, you may or may not directly benefit from taking part. We will review the risks and benefits of participating in the study thoroughly with you before you decide whether or not you want to participate.

Who do I contact for more information?

You can contact Lauren Ivey, research assistant, by calling (301) 435-6689 or by email at [iveyle@mail.nih.gov].