Research Update

Over the past year the cutis laxa research study has expanded! Dr. Urban, his collaborators, and the research team have had two patient research clinics in 2011. This has allowed us to collect significantly more information on cutis laxa. This fall, Dr. Urban traveled to Lyon, France to meet with researchers from Europe and attend the Cutis Laxa Internationale meeting. Dr. Urban participated in the 4th Days on Cutis Laxa Conference organized by the patient support group Cutis Laxa Internationale. As a part of the information day he gave a talk on the current use of animal models to better understand the mechanisms that cause cutis laxa and to develop new treatments. In September 2011 we launched our own University of Pittsburgh cutis laxa research website (www.hgen.pitt.edu/projects/cutislaxa/). Our website contains information on what cutis laxa is, how it is inherited, FAQs, research updates, and important links for our participants and anyone who is interested in learning more about cutis laxa or has any questions about our research. If you would like to enroll in our study or give us updated information on you and/or your child please feel free to contact either of our two research coordinators, Meghan Mac Neal and Kara Levine, at cutislax@pitt.edu or 412-383-7369.

Recruitment

Recruitment for the cutis laxa study has been very active this year and we are pleased with the progress of data collection. The table below shows total recruitment numbers for individuals and families participating through the research clinics in Pittsburgh and through their local physician. A special thanks goes out to all of our families who participate in this study. Every person we recruit helps us learn more, and we could not do this research without you! In 2012 we will be looking for both new participants and updates on people already enrolled.

<table>
<thead>
<tr>
<th>Type of participation</th>
<th>Families</th>
<th>Individuals</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pittsburgh Research Clinic Participants</td>
<td>23</td>
<td>26</td>
</tr>
<tr>
<td>Total Participants</td>
<td>125</td>
<td>228</td>
</tr>
</tbody>
</table>
Upcoming Clinic

We will be having a research clinic on June 21-23, 2012 for 12 participants. Please check your inboxes for official information and let either Meghan or Kara know if you would like more information about the upcoming clinic. We are hoping for a great turnout and look forward both to meeting new participants and seeing previous ones again. If you have already participated in a research clinic and would like to know if you would be eligible to come back this summer, please email or call one of the research coordinators:

Kara Levine or Meghan Mac Neal
Phone: 412-383-7369
Fax: 412-624-3020
Email: cutislax@pitt.edu

Spreading the Word

Over the past year, Dr. Urban and his research team have given multiple talks and research presentations to other members of the scientific and medical community. Below you will find a list of our research publications from 2011. Look on page 4 for our 2011 presentations. Slowly but surely, more and more people are hearing about cutis laxa!

We encourage the families in our research study to become active in the cutis laxa community. Visit our website at www.hgen.pitt.edu/projects/cutislaxa/related_links.php for links to some of the support groups available to families. We would also encourage the formation of an official non-profit organization for cutis laxa in the US. If you are interested in helping with this endeavor, please contact our research team for more information. Many participants have commented that it is really beneficial meeting/talking with other families who have been through similar situations.

Cutis Laxa 2011 publications


Our Team

The Cutis Laxa Research Team consists of Dr. Zsolt Urban, his collaborators, and his research staff. As the principal investigator, Dr. Urban manages the research both from a laboratory and clinical perspective. Meghan MacNeal and Kara Levine are the study coordinators, whose main responsibilities include recruitment of new participants, coordination of the Pittsburgh clinics, and the management of data as well as information flow between our research team and the study participants. Dr. Madan-Khetarpal and Juliann McConnell are a part of the clinical component of our team. They perform the genetic evaluation at our cutis laxa clinics. We have several individuals working in the lab including a new lab manager Elizabeth Lawrence, doctoral student Sandeep Khatri, visiting student interns Bianca Schulz and Gulab Sher, masters student Chi-Ting Su, and an undergraduate student, Maureen McGowan.
Cutis Laxa 2011 Presentations

- Invited Seminar, Department of Anatomy, Biochemistry and Physiology, John A. Burns School of Medicine, University of Hawaii, Honolulu, HI, January 10, 2011. “The molecular basis of elastic fiber assembly and TGFβ signaling: insights from human genetics.”

- Invited Seminar, Department of Environmental and Occupational Health, Graduate School of Public Health, University of Pittsburgh, Pittsburgh, PA, April 28, 2011. “An elastic view of genes, environment and disease.”

  - Khatri S, Maxfield AB, Mecham RP, Joseph EM, Urban Z. Fibulin-4b is required for cardiovascular system development and as an antagonist of transforming growth factors tgfβ2 and tgfβ3.


- Invited Seminar, Department of Anatomy and Cell Biology, McGill University, Montreal, Canada, October 12, 2011. “Human genetic insights into the elastic fiber/TGFβ network.”
