

The last newsbit, was basically about the final planning stages of our annual convention, a while back, which was held in Shell Knob, Missouri. Hosted by Kim O. While there, it was decided that we would meet every-other-year because of the expense in hosting this event, and to give families the opportunity to have other family vacations on the off year.

The content of this newsletter is mainly to inform you of the conversations and planning strategies of further and ongoing research. Please read, then if you have any questions feel free to call Kim or Karla (e-mail is preferred).

***Following the June 2000 Convention in Missouri, we received an e-mail from Dr. Han Brunner about our interest in having more research done about Robinow Syndrome. Here is a summary of what he had to say:***

October 16, 2000

*. . . When I visited your group in June, we discussed the possibility for a US national study to develop clinical criteria for the diagnosis of Robinow syndrome, for the assessment of the nature and frequency of significant complications, and for things like natural history and molecular genetic diagnosis.*

*At the last Human Genetics Conference in Philadelphia I briefly discussed this possibility with another who said that there is group at the National Institute of Health that might be interested to do this by evaluating a group of Robinow patients in Bethesda, MD.*

*Please let me know what you think. I believe that the syndrome can be, and should be, more precisely defined as nobody seems to have done any major clinical work on this since Dr. Butler in 1987, and with the possible exception of Dr. Balci from Turkey.*

Dr. Han Brunner

***We responded to Dr. Brunner's E-mail. From there we also sent an E-mail to Dr. Clair F. Following are some of the responses we received***

November 2000

Dear Kim and Karla,

*Thank you very much for your note. I am indeed extremely interested in working with you to answer the research questions you have addressed. I apologize for the long delay in responding to your message; please do not interpret this as a lack of interest or enthusiasm. I agree that a team approach would be very valuable, both from the point of view of the*

*patients and also the research questions.*

*Ms. Emily G. is a research assistant with me at the National Human Genome Research Institute. She will be contacting you to help in defining the next steps. If you would be interested in traveling to the Washington area to meet about this, I would be delighted to arrange for your travel and expenses to be covered while you are here.*

*I look forward to our future correspondence, to meeting with you, and to furthering the cause of research and patient care for families with Robinow syndrome.*

Many thanks,  
Dr. Clair F.

November 8, 2000

*My name is Emily G. and I would like to introduce myself and offer my assistance to you in coordinating future research endeavors on Robinow Syndrome. I am a Post-Baccalaureate Fellow in the National Human Genome Research Institute working for Dr. Clair F., whom you contacted in reference to research on Robinow Syndrome. Dr. Clair F. and myself are both very interested in furthering the research and patient care opportunities for patients and their families with Robinow Syndrome.*

*I look forward to speaking with you about future projects.*

Emily

***Soon we will be in contact with the NIH in making arrangements for travel to Bethesda, MD. All families that wish to be a part of this study will be contacted to coordinate the best time for their family to make the trip. Accommodations and travel arrangements will be at the courtesy of the NIH [Affected child(ren) and one parent]. This study will probably be over the course of one-two years. The survey's that we have been receiving from each family will help begin the clinical research that Dr. Clair F. and her team will be starting soon. Please keep sending us your surveys so that we can be prepared for the initial start of this project. This will be one great step for all of us and the many unanswered questions we all have. We hope that most families will be willing to take advantage of this study.***

Special thanks to Kim O. for keeping in contact with them and for putting such effort into helping coordinate this.

ROBINOW SYNDROME CONVENTION "2002"  
will be held June 20 - June 23

Hosts: Kevin & Karla Kruger  
15955 Uplander Street NW, Andover, MN 55304  
Phone: (763) 434-1152

Come and join us for a truly unique experience.  
Meet others who will share their stories and knowledge of this syndrome.

**Please:** RSVP to Karla (call or e-mail) so appropriate plans can be made for food and entertainment, etc.

**LODGING INFORMATION**

Campfires, fun, food, lots of learning,  
reminiscing & conversation  
Mall of America (approx. 45 min. drive)

***A+ FOR EFFORT***

Sara Hutton is still going strong selling her newsletters within her neighborhood. Her handwritten newsletters, which include all information pertinent to her neighbors, are sold for a minimal price and she has also, collected other monetary donations. These funds (almost \$300) are then sent to the foundation. Is she an entrepreneur in the making or what? What an example she is setting for us.

***Thanks Sara!!***

***"For Your Information"***

Lately the foundation has been receiving a remarkable amount of e-mails requesting information about this syndrome from teacher's aides, paras, case managers for different programs, and students in both High School and in College. Most are for biology reports they are doing on a genetic disorder. How and where did they even hear about Robinow Syndrome? We must be doing something right, because the word is obviously getting out there. I wish it was more "out there" many years ago, so we could all be more informed and educated!! Modern technology (computers & internet) is definitely paying off, because people worldwide are viewing what is on our website. If you want to add something or make changes please contact the Foundation.

For those of you who were at the meeting in Shell Knob, and contributed cheek swabs for DNA testing, the latest word from Dr. Brunner is that he still has not completed all the testing, and he does need more DNA from a few of us. Here is a portion of the most recent e-mail from him:

*Robinow Syndrome Foundation:  
Kim Oritz and Karla Kruger,*

*. . . We are at long last finalizing the results for ROR2 mutation testing of your son. I am ashamed to realize how long it has taken us. I shall let you know as soon as the data are complete.*

*I got some lovely pictures from the meeting. I am using one of these in my talks regularly to show that this type of research can be fun, as well as intellectually stimulating.*

*We haven't really been getting much new in the way of the recessive type of Robinow syndrome lately. We are pretty much waiting for the biochemists to tell us more about the normal functions of ROR2. We hope to then use that information as springboard for other studies to do with the syndrome. I have no idea when that will be. In the meantime we have sent some DNA samples to a researcher in Brazil who works on the related ROR1 gene. Maybe she will turn up something that is of interest.*

*There is one exciting new development though. I have established contact with a genetic counselor . . . in California. They have seen a very large kindred which looks like dominant Robinow syndrome, although maybe not quite. I haven't seen the pictures yet. In any case they are interested in letting us study that family, and maybe this will give us some new clues. We'll have to see how this develops.*

*I hope that you and Karla can keep the group together. It has a lot to offer to parents and children alike. Yet I know that the work that goes into these groups tends to increase all the time. At some point another parent may need to take over the work.*

*Here all is well. Best regards, and best personal wishes,  
Han G. Brunner, MD*

I'm thinking the location of the next meeting (2004) should be out East (where the study will be taking place) so the findings can be presented to us - providing they have been completed at that time. You may be contacted and it is your choice to participate if you wish to do so. Here is a partial excerpt of a recent letter received from her:

*Robinow Syndrome Foundation:*

*. . . a note in response to an e-mail from Kim*

*Thank you for your notes. We have been experiencing a long string of bumps and snags in the process of getting our clinical research approved in Baltimore. However, we are getting closer and closer, and I sincerely hope that by mid - March we will be able to start bringing patients in. I am still very interested in working with your group to answer the many clinical questions that are still out there concerning Robinow Syndrome. I appreciate your patience in this long transition, and hope we will be able to get going very soon . . .*

*Clair*