

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.

