

# CLINICAL RESEARCH

## RESEARCH BEING DONE ON ROBINOW SYNDROME AROUND THE WORLD:

There are a handful of researchers doing research on Robinow syndrome around the world. Affected families, as well as the scientists, want to see this research translated into usable information for patients.

The National Institute of Health Office of Rare Diseases is sponsoring a new program called the Collaboration, Education and Test Translation Program (CETT) to get research findings into the hands of the people who can use it. The government website summarizes the program as follows: "Collaboration is the key word in the CETT Program's goal to help facilitate the translation of new tests for rare genetic diseases. The program's goals are to translate as many tests as possible and ensure that the best possible test is offered, whether the specific genetic condition affects 5 people or 50,000." (<http://www.cettprogram.org/>)

Researchers are working to discover new genes that are associated with this syndrome. Because research labs are not designed to test and treat individual patients, people who want to be tested for a gene need to be referred to a clinical laboratory. It takes time and money for clinical laboratories to create new tests. Some of these tests may be performed infrequently because of the low incidence of the syndrome. The CETT program provides funding for clinical laboratories to create new tests for genes that have been discovered in the research lab in order to help affected families. This program also encourages continuing collaboration between the clinical laboratory, researchers and patient advocates. This insures that research will continue even after the test is moved from the research lab into the clinical setting.

Karla Kruger, Kim Ortiz-Kremeier and **The Robinow Syndrome Foundation** in collaboration with the University of Chicago Genetic Services laboratories and Dr. Han Brunner and Dr. Hans Scheffer at the Radboud University Nijmegen Medical Centre in The Netherlands have received a CETT grant to develop a clinical test for ROR2, the gene related to autosomal recessive Robinow syndrome. This test should be available at The University of Chicago Genetic Services Laboratories sometime this summer.

Please contact Melissa Dempsey ([mdempsey@bsd.uchicago.edu](mailto:mdempsey@bsd.uchicago.edu); 773-834-1606) for more information.

### DEFINITION OF CLINICAL TESTING:

**Testing that is ordered by a physician, billed for and usually covered by an insurance company, and a report is written and sent to the ordering physician.**

**As a physician needs to order the testing, we will not be drawing blood at the meeting in Minneapolis, but we will distribute paperwork about the testing that families can take to their physician.**