

"Multi-Lingual" Webpage

We are currently seeking assistance in aiding us to have the webpage in multi-lingual. Those buttons, will be on the webpage when they become available. German, Spanish, Italian & French have been our biggest requests. So, if you know of someone who is willing and can translate - please forward their names and phone numbers to the foundation so I can discuss with them the details, including a timeline, content, payment, etc.

THE FOUNDATIONS FINANCIAL INFORMATION

For those of you who are wondering where the fundraising money is, and what it is spent on, I will gladly send the financial status of the Foundation, to you. Please send a note requesting a copy. Include a "Self-Addressed, Stamped Envelope".

The website is going to undergo some minor changes. The "General Info" button will have corrections made. The "Newsletter" button will be updated as well. New buttons to be created are: "Donations". This is where instructions for a tax-deductible contribution will be listed. Remember, many, many businesses offer "Matching Funds" donations. I encourage you all to pursue that option where available. Another will be where all the information from the first package will be. Package #2 (when completed) will too be added.

"Info. Package #1 & #2"

I have currently run out of pkg. #1, and will need more immediately. pkg. #1 is still provided free of charge to new families. To date, I personally have sent out over 85 of those envelopes. Kim Ortiz has also provided pkg. #1 to many families. Info. package #2 is presently being compiled - the cost of photocopying both packages will be incredibly huge - even with a discount. It should be available within the next year. If you want to receive pkg. #2 via "snail-mail" the cost will be somewhere between \$12-\$15 depending on the weight and mailing address when finished. It will also be placed on the website, for those who have internet access, to obtain.

Some of you will find enclosed in this mailing a form letter with your personal information that the foundation has. Updates are necessary and any incorrect or outdated info. will need to be changed. Please complete or correct your info. and send in to the foundation. Please note that I will need information about relatives that may be affected or possibly affected, siblings, etc. Email responses will be fine for those who have it. As noted on the letter - if you do have e-mail, all future newsletters will be sent to you that way. Thus, saving the foundation many dollars in postage. The current newsletter will be posted on the website.

The latest in Genetic Research

Mutation of ROR2 tyrosine kinase gene causes autosomal recessive Robinow syndrome.

Hans van Bokhoven¹, Jacopo Celli¹, Hülya Kayserilif, Ellen van Beusekom¹, Sevim Balci², Wim Brussel³, Flemming Skovby⁴, Bronwyn Kerr⁵, E. Ferda Percin⁷, Nurten Akarsu³, Han G. Brunner¹

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- 5 Department of Clinical Genetics, Rigshospitalet, Copenhagen - Denmark
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Robinow syndrome is a short-limbed dwarfism characterized by abnormal morphogenesis of the face, vertebral segmentation, and the external genitalia. The recessive form of Robinow syndrome (RRS; OMIM 268310) has a high incidence of abnormalities of the vertebral column such as hemivertebrae and rib fusions, which are not seen in the dominant form. Some patients have cardiac malformations or facial clefting. We have mapped a gene for RRS to 9q21-q23 in eleven families.

Prominent features of Recessive Robinow Syndrome (RRS) are hypertelorism, short stature, mesomelic shortening of the limbs, hypoplastic genitalia, and rib and vertebral anomalies.

Finally, it is now possible to test whether the recessive and autosomal dominant forms of Robinow syndrome are allelic due to ROR2 mutations.

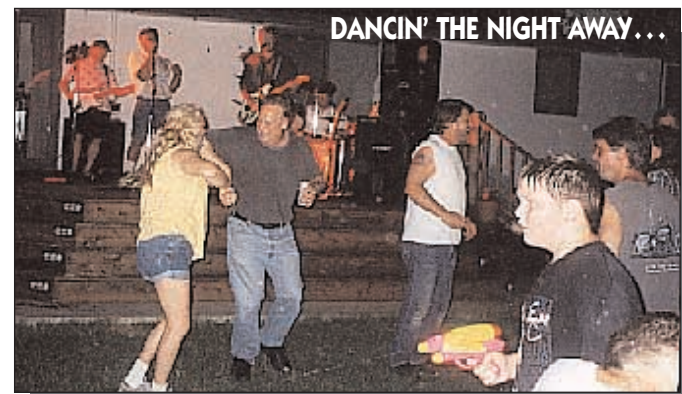
Key words: Robinow syndrome, linkage, Ror, Tyrosine kinase receptor, segmentation, skeletal dysplasia

New Research on the dominant form of Robinow syndrome is now making progress. The location of the gene mutation that causes the dominant form of Robinow may be available for testing in about one year.

MORE CONVENTION HIGHLIGHTS:



THE MEETING . . . KIM, KARLA, ED & JULIANA

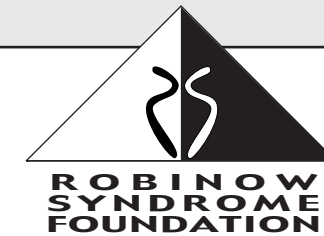


SMALL ABOUT ROBINOW SYNDROME TALK

We are a "Little" group of families reaching out for one another's support on dealing with this medical rarity and how it affects all of our lives.

ROBINOW SYNDROME FOUNDATION

FALL 2002



The 6th Annual Robinow Syndrome Convention was held in Andover, Minnesota from June 18-25, hosted by Kevin & Karla Kruger. We had a wonderful turnout. Many of us had a blast with our existing friendships and all of us are thankful to have met the new families for the first time. We are hopeful that all families will continue to keep in contact with each other and the foundation for many years to come.

This convention brought together families from all over the US & Canada who need to find new and available resources that provide information and support for this rare condition. "We are one big happy family - with a bond that is so unbelievable it is hard to explain!"

This is such a rare opportunity for both the children and adults to be with others with the same or similar medical issues. The bonds we develop with each other is priceless to both our self esteem and emotional support.

Guest speaker this year was Dr. Juliana Mazzeu, coming all the way from Brazil, who is actively researching the gene(s) that causes this rare genetic disorder. She says "...there is not yet available, any type of 'gene therapy' to assist in the treatment of this disorder..." but, "we are continually getting closer and closer to understanding how this syndrome is developed." Juliana has located the genetic mutation that is believed to be the cause of RS. These new findings will allow for proper and early diagnosis.

Each family had their own personal and private time with Juliana to discuss any of their own concerns with her. That time, was very beneficial to the families because there are so many unanswered questions that we all have.

The Kruger's provided food, entertainment and even lodging (for some). We had a variety of entertainment for both the adults and children arranged, some made it and others

- McDonald's, Coon Rapids
- Pizza Hut, Anoka
- Target, Coon Rapids

And those families, who so generously donated:

- Ms. B. Davis
- Ms. S. Otten
- Ms. M. Moorhead
- Ms. M. Stocksleth
- Ms. M. Bernhagen
- Ms. K. Ortiz
- Mr. & Mrs. S. Burnham
- Mr. & Mrs. D. Petermeier
- Mr. & Mrs. F. Unger
- Mr. & Mrs. D. LaFlamme
- Mr. & Mrs. E. Motzkin
- Mr. & Mrs. G. Cook
- Mr. & Mrs. F. Hellman
- Mr. & Mrs. K. Notch
- Mr. & Mrs. H. Kruger
- Mr. & Mrs. T. Kruger

didn't even show up. Those are some of the obstacles one can not control when planning for these events.

Everyone really enjoyed the live 'pop-rock' music, which was played on our deck Saturday night by 'The Boneshakers', and the dinner music which was provided by Soloist Kama Otten, and of course, from the additional 14 people staying in our home. It was very stressful, financially draining, and took at least 6 months of planning, but so incredibly necessary, very satisfying & worthwhile.

This year's convention could not have happened as smoothly as it did, without the help of these contributors. A sincere 'Thank you' to all of them for their help. (not in any particular order):

- Salvation Army, Alexandria, VA
- City of Andover
- Billy's Bar & Grill, Anoka
- Festival Foods, Coon Rapids
- Tracy Printing, St. Anthony
- Bill's Superette, Coon Rapids
- Lashinski Septic, Andover
- Wal-Mart, Coon Rapids
- Cub Foods, Coon Rapids
- Rainbow Foods, Coon Rapids
- VFW Post #6702, Andover
- Old Dutch, Roseville
- American Legion Post #334, Coon Rapids
- Anoka County Parks
- Kinko's, Coon Rapids
- Mello Smello, New Hope
- Brush Masters, Plymouth

And thanks to all our neighbors for their understanding and patience throughout the event.

Contributions made during and after the convention are in the account and will go towards the next convention and to aid in the areas listed below.

Our next convention is set for June, 2004 to be held in, New York. Anita Hutton has volunteered to coordinate the event. Fundraising efforts are continuous to help maintain the Foundation's website, research, newsletters, postage, P.O. box, medical and travel assistance and to help in aiding families ability to attend our bi-annual gatherings, but are unable to because of the financial strain due to medical expenses.

Thanks to all who attended - we had a blast and I look forward to seeing you again in 2004!

"2002 CONVENTION HIGHLIGHTS"

JUNE 18 - 24 • ANDOVER, MINNESOTA



A: Sondra (holding) Hope, Mark & Layne Stevanus
B: Beth Thurman
C: Elizabeth, Valerie & Jeremy Montgomery-Bull
D: Keeyla, Adrian (holding) Adrian Jr. Martinez
E: Brett, Veronica (standing) Nelson & Aunt Kris
F: Kim Ortiz & Ryland Kuehn
G: Ed, Joyce & Cori Motzkin
H: Guest Speaker - Juliana Mazzeu
I: Ralph, Amy, Laura & Jean Patterson.



J: Christina Nazaruk
K: Bonnie & Angela Tirrell
L: Bob, Barbara & Robert Winsor
M: Catherine Johnson (inset) Kyla
N: Anita, Sarra & Lauren Hutton
O: Calvin, Nancy & Adrian Cash
P: Kelsey, Kevin, Kadean & Karla Kruger

