

ROBINOW SYNDROME FOUNDATION

9th Biennial CONVENTION

Minneapolis, Minnesota
June 25th - 29th
2008

AGENDA:

*The scheduled events listed are **PRELIMINARY** only. As the final weeks pass by please keep checking this site as more definitive details still may be arranged & confirmed.*

WEDNESDAY, JUNE 25th:

- Arrival of participating patients and families at various times throughout the day
- Check-in at Marriott Airport Hotel or Holiday Inn Airport Select
- Gather poolside at Marriott Hotel for meet-and-greet / socialization
- Welcome meeting for new (first time) families: location & time TBA

THURSDAY, JUNE 26th:

- **Daytime:** Social event for patients families to be announced and organized by **The Robinow Syndrome Foundation**
- **Evening:** Dinner at Famous Dave's Restaurant at the Mall of America from 4:30 p.m. to 6:00 p.m. Provided by **The Robinow Syndrome Foundation**. *Meet in lobby of Marriott at 4:15 p.m.*
- Arrival of scientific speakers and attendees

FRIDAY, JUNE 27th:

- **Morning:** Scientific sessions
(See tentatively scheduled scientific session - Friday - outline on pg.3)
- **Afternoon:** Breakout sessions with clinicians and researchers along with meetings and research participation
(See tentatively scheduled clinical sessions - Friday - outline on pages 5 & 6)
- **Evening:** Faculty dinner and meeting for discussion of ongoing research projects and collaborations *(Dr. Lohr to host)*
Families enjoy free social time - other evening events; planning is underway.

SATURDAY, JUNE 28th:

- **Morning:** Scientific sessions
(See tentatively scheduled scientific session - Saturday - outline on pg. 4)
- **Afternoon:** Breakout sessions with clinicians and researchers along with meetings and research participation
(See tentatively scheduled clinical sessions - Saturday outline on pages 5 & 6)
- **Evening:** Unscheduled / To be announced

SUNDAY, JUNE 29th:

- **Morning:** Farewell breakfast *(not yet scheduled or confirmed)*
- Departures

8:00 - 9:00 a.m.	Continental breakfast provided Welcome (Karla Kruger / Jamie Lohr)
9:00 - 9:45 a.m.	Keynote Speaker Lisa Schimmenti, M.D. <i>Associate Professor of Pediatrics and Ophthalmology, University of Minnesota, Minneapolis, MN</i> <i>“Pathways and Models: finding genes for Human Genetic Conditions”</i>
9:45 - 10:15 a.m.	Jamie Lohr, M.D. <i>Assistant Professor of Pediatrics, Division of Pediatric Cardiology, University of Minnesota, Minneapolis, MN</i> <i>“Molecular Causes of Autosomal Dominant Robinow Syndrome”</i>
10:15 - 10:45 a.m.	Hans van Bokhoven, Ph.D. <i>Radboud University, Nijmegen Medical Center, The Netherlands</i> <i>“Chromosomal Abnormalities Associated with Dominant Robinow Syndrome”</i>
10:45 - 11:00 a.m.	Break
11:00 - 11:30 a.m.	Soraya Beiraghi, D.D.S. <i>Professor of Dentistry University of Minnesota, Minneapolis, MN</i> <i>“Craniofacial and Dental Complications of Robinow Syndrome: Diagnosis and Treatment”</i>
11:30 - 12:00 p.m.	Cheryl Anderson-Cermin, D.D.S. <i>“Common Orthodontic Problems in Robinow Syndrome and other Conditions”</i>
12:00 - 12:30 p.m.	Anna Thurmes, M.A., <i>Doctoral Student (Craniofacial Clinic), University of Minnesota, Minneapolis, MN</i> <i>“The Psychological Impact of Living with a Genetic Condition”</i>
12:30 - 12:45 p.m.	Jenell Holstead, MS: <i>Center for Evaluation and Education Policy,</i> <i>“Introduction of Research Study”</i>
12:45 - 1:30 p.m.	Lunch and roundtable with speakers • Dr. Soraya Beiraghi (<i>Dentistry</i>), • Dr. Anna Petryk (<i>Endocrinology</i>), • Dr. Cheryl Cermin, (<i>Orthodontics</i>), • Anna Thurmes (<i>Craniofacial Clinic</i>)
1:30 - 5:00 p.m.	Open for participation in research studies and individual meetings with clinicians and scientists <i>(main and small conference rooms available)</i>

SATURDAY, JUNE 28th:**MORNING SCIENTIFIC SESSIONS**

8:00 - 9:00 a.m.	Continental breakfast provided
9:00 - 9:30 a.m.	Lisa Schimmenti <i>or</i> Soraya Beiraghi <i>“Clinical Findings in Dominant & Recessive Robinow Syndrome”</i>
9:30 - 10:00 a.m.	Hans van Bokhoven, Ph.D. <i>Radboud University, Nijmegen Medical Center, The Netherlands</i> <i>“Molecular Causes of Recessive Robinow Syndrome”</i>
10:00 - 10:30 a.m.	Stephen Ekker, Ph.D. <i>Professor of Biochemistry and Molecular Biology, Mayo Clinic, Rochester, MN</i> <i>“Animal Models of ROR2 Mutation”</i>
10:30 - 10:45 a.m.	Break
10:45 - 11:15 a.m.	Melissa Dempsey, M.S. <i>CETT Program, University of Chicago, Chicago, IL</i> <i>“Clinical Testing for Recessive Robinow Syndrome”</i>
11:15 - 11:45 p.m.	Aseem Shukla, M.D., <i>Assistant Professor of Urology, University of Minnesota, Minneapolis, MN</i> <i>“Genital Hypoplasia - Common Problems and Treatments</i>
11:45 - 12:30 p.m.	Karla Kruger, President & Founder, Andover, MN Kim Ortiz-Kremeier, Resource Manager, Shell Knob, MO <i>“The Robinow Syndrome Foundation”</i>
12:30 - 1:30 p.m.	Lunch and roundtable with speakers <ul style="list-style-type: none">• Dr. Soraya Beiraghi (<i>Dentistry</i>),• Dr. Hans Van Bokhoven, (<i>Genetics</i>),• Dr. Aseem Shukla (<i>Urology</i>),• Dr. Anne Biosclair-Fahey (<i>Urology</i>)• Dr. Steven Ekker (<i>Genetics</i>),• Anna Thurmes (<i>Craniofacial Clinic</i>)• Dr. Melissa Dempsey (<i>Genetics</i>),
1:30 - 4:00 p.m.	Open for participation in research studies and individual meetings with clinicians and scientists <i>(main and small conference rooms available)</i>

FRIDAY, JUNE 27th and SATURDAY, JUNE 28th

AFTERNOON CLINICAL SESSIONS

PLANNED RESEARCH STUDIES

One of the greatest benefits of this convention will be the fostering of research efforts in the genotype-phenotype correlation, clinical genetics, molecular biology and pathophysiology of Robinow Syndrome. The scientific sessions will serve to review current findings related to Robinow Syndrome and will foster collaboration and new studies among researchers.

The 2006 convention was attended by close to 20 families. 90-100 people gathered and included approximately 25 families, 90-100 people and approximately 25 affected individuals. At the 2008 convention we are striving to have 35 families and approximately 40 affected individuals who are willing to participate in research about their condition.

The following clinical investigations will be available for enrollment and testing by interested and qualified participants at the 2008 Robinow Syndrome Foundation Convention.

CURRENTLY I R B APPROVED:

- **Juliana Mazzeu, Ph.D.** *Department of Genetics, University of Sao Paulo, Brazil*
“Clinical Characteristics of Autosomal Recessive and Autosomal Dominant Forms of Robinow Syndrome”
Dr. Mazzeu will enroll patients and perform a detailed pedigree and physical examination for genotype/phenotype correlation.
 - *Dr. Mazzeu is unable to attend, Drs Lohr & Beiraghi will collect information on new patients.*
- **Jamie Lohr, M.D.** *University of Minnesota, Minneapolis, MN*
“The Molecular Causes of Autosomal Dominant Robinow Syndrome”
Dr. Lohr will enroll patients and obtain a blood sample for investigation of mutations in the Wnt5a-ROR2 signaling pathway.
- **Jenell L. Holstead,** *Graduate Student, Indiana University*
“The The Psychological Impact of Robinow Syndrome”
Ms. Holstead will meet with patients and families and provide them with questionnaires regarding the psychological aspects of Robinow Syndrome.

PENDING I R B APPROVAL:

- **Jamie Lohr, M.D.** *University of Minnesota, Minneapolis, MN*

“Cardiac Manifestations of Robinow Syndrome”

Dr. Lohr will perform EKG's and echocardiograms on patients after enrollment to screen for asymptomatic cardiac findings

- **Soraya Beiraghi, D.D.S.** *University of Minnesota, Minneapolis, MN*

“3D Craniofacial Imaging in Robinow Syndrome”

Dr. Beiraghi will perform 3D assessments of oro-dental and craniofacial abnormalities in patients.