

# ROBINOW SYNDROME FOUNDATION

## CONVENTION *9th Biennial*

**Minneapolis, Minnesota**

**June 25<sup>th</sup> - 29<sup>th</sup>**

**2008**

## INTRODUCTION:

*With this convention and our ongoing efforts at research and education, we hope to improve clinical and scientific awareness of this syndrome, improve the accuracy of diagnosis and apply current rapidly expanding genetic knowledge to understanding the development and treatment of this rare but increasingly recognized and somewhat disabling syndrome.*

*We are pleased you are joining us this year. We have many families attending! We hope your journey will be inspirational and your memories will last a lifetime. May you leave our convention with the comfort of knowing that you are not alone. We are all part of this "extended family" and we all support each other. May the next two years pass by quickly . . . until we can meet again in 2010.*

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### BACKGROUND

#### **The Robinow Syndrome Foundation and its Biennial Convention**

The Robinow Syndrome Foundation was established in the early 1990's by the parents of a child with the diagnosis of Recessive Robinow Syndrome. The group became a non-profit organization in 2002. The Foundation's main goal is to provide education and support to patients and their families, primarily through biennial conventions and their website. **The Robinow Syndrome Foundation** has planned "conventions" every other year where families can meet, socialize and share useful information and experiences. Over the last several years, there has been increasing participation from researchers and medical professionals. Prior "gatherings/conventions" have been held in Findlay, OH (1992), Tucson, AZ (1993), Mt. Lemmon, AZ (1994), Andover, MN (1993, 1995, 1997, 2002, 2008), Tucson, AZ (1996), Lee's Summit, MO (1999), Shell Knob, MO (2000), Hurley, NY (2004), and Dayton Ohio (2006).

**TO OUR NEW FAMILIES:** *2008 is already proving to be yet another spectacular year - all of us together are creating life-long memories of our "Robinow family". We hope you continue to remain in contact with us and return for years & years to come.*

**TO OUR RETURN FAMILIES:** *Thank you for being a part of this foundation. We wouldn't be . . . if it weren't for you.*

**TO ALL OF US:** *We are grateful to be **THE FOUNDATION:***

- *To share*
- *To learn*
- *To teach*
- *To celebrate our existence!*

For the 2008 meeting, we will maintain unstructured time for the families to meet and share experiences and socialize before and after the scientific sessions. The scientific portion of the meeting will be expanded to include a minimum of 4-6 scientific speakers with topics of interest to healthcare professionals and researchers as well as patients and families, and at least four clinical speakers on topics ranging from the diagnosis of Robinow Syndrome to oro-dental care, orthopedic care, gynecologic care and the psychosocial issues associated with genetic conditions in general and Robinow Syndrome in particular. In addition to patients, families, and healthcare professionals and researchers already interested in Robinow Syndrome, we hope to attract a small scientific audience of postdoctoral fellows and faculty from the MinnCRest Program at the University of Minnesota (*Minnesota Craniofacial Research Training Program*), the Craniofacial Clinic at the University of Minnesota, the Mayo Clinic, genetics and genetic counseling professionals from local and regional programs, and developmental biologists interested in craniofacial, genital and skeletal development. In addition to the scientific sessions, we will have several researchers present to enroll patients who desire to participate in IRB approved studies of the clinical features, genetic etiology, dental evaluation, growth hormone and cardiac evaluation of Robinow Syndrome patients. The presence of so many patients and families with this single diagnosis in one place represents a unique clinical and scientific opportunity.

Costs for the scientific sessions and research portions of the meeting will be borne by the sponsoring institutions, not by the families. **The Robinow Syndrome Foundation** will provide funding for social activities, travel awards and hotel subsidies for attendees as the budget and other fundraisers allow.

#### **CLINICAL REVIEW/SUMMARY** (*Submitted by U of MN*)

In 1969, Meinhard Robinow and colleagues described a human syndrome characterized by short stature, mesomelic limb shortening, hypertelorism, mandibular hypoplasia, irregular dental alignment and hypoplastic external genitalia<sup>1</sup>. Based on the initial pedigree, Robinow syndrome was recognized as an autosomal dominant inherited syndrome [*MIM 180700*] with high penetrance. Over 100 patients with Robinow syndrome have since been identified in families with both autosomal dominant and autosomal recessive inheritance patterns<sup>2</sup>.

The autosomal recessive form of Robinow Syndrome [*MIM 268310*], which is characterized by more severe skeletal, vertebral and craniofacial abnormalities,<sup>2,3</sup> is often caused by loss-of-function mutations in the gene encoding the tyrosine kinase-like orphan receptor<sup>2</sup>, ROR2<sup>4,5</sup>. Recently, Ror2 has been identified as a putative receptor for Wnt5a6. Wnt5a and Ror2 are expressed in adjacent and partially overlapping domains during mouse embryogenesis, and Wnt5a can directly bind to the extracellular domain of Ror27,<sup>8</sup>. In addition, Wnt5a<sup>-/-</sup> and ROR1/2<sup>-/-</sup> mice have similar phenotypes which are suggestive of many findings seen in patients with Robinow Syndrome<sup>8,9</sup>. Our work in autosomal dominant Robinow Syndrome suggests that decreased WNT5A activity may be associated with dominant Robinow Syndrome in a subset of patients. To date, however, a role for WNT5A/ROR2 signaling in human disease has not been identified. The goal of our ongoing work is to explore the role of the Wnt5a/ROR2 signaling pathway in human development and disease.

#### **CONFERENCE VENUE AND CHILD CARE**

The 2008 Robinow Syndrome Foundation Convention will be held at the Minneapolis Airport Marriott in Bloomington, Minnesota. The Marriott is within 5 miles of the Minneapolis-St. Paul International Airport and adjacent to the Mall of America. The hotel offers free shuttles to and from the airport and mall for all guests. The conference facilities include one large conference room for lectures and two smaller rooms for research projects or individual consultations. Catering and restaurants are onsite and are excellent. The rooms appear comfortable and there are adequate common areas, including a large pool and deck for socialization and impromptu meetings. Other amenities include wireless internet and smaller rooms adjacent to the conference rooms for activities for toddlers, school age children and adolescents who are patients, or siblings of patients, and do not want to attend the sessions. Supervision and age-appropriate activities will be arranged by the conference organizers.

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## INCLUSION OF WOMEN AND MINORITIES, PERSONS WITH DISABILITIES

The organizers have made a concerted effort to achieve representation of women, racial and ethnic minorities, persons with disabilities and other individuals who are under-represented in science in our speakers and audience. The conference facilities and organizers are prepared to meet the needs of conference attendees with disabilities. The registration form will include a question that will allow registrants to identify these needs and begin planning.

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## SUMMARY

The 2008 **The Robinow Syndrome Foundation** Convention will bring together patients, clinicians and researchers to learn about the clinical diagnosis, pathophysiology and genetic causes of Robinow Syndrome. The information shared at this conference will improve the health and well being of patients with Robinow Syndrome, will augment the clinical knowledge of their care providers, and will stimulate research on the role of a novel WNT5A-ROR2 signaling pathway in human development and disease.

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## REFERENCES

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