

5th SMS Research Roundtable Symposia held in conjunction with 6th International Family conference on Smith-Magenis Syndrome, “Building Bridges of Hope”, Sept 17-20, 2009

Report

Funding support received from the Office of Rare Disease Research (ORDR) was used to co-sponsor a one-day invited scientific symposium in collaboration with PRISMS¹, the internationally recognized support group for Smith-Magenis syndrome (SMS). SMS is a rare disorder affecting 1/25,000 births. In collaboration with PRISMS' Professional Advisory Board (PAB), Ann C.M. Smith, MA, DSc (hon) (OCD/NHGRI) and the NIH SMS Research

Team organized the 5th SMS Research Roundtable (SMS-RR) Symposium. The SMS-RR symposium promotes a *trans-disciplinary* approach to research that enhances and advances knowledge about the molecular genetic basis, clinical variability, natural history, and underlying pathophysiology of this rare syndrome. The goals of the symposia were to:

- 1) Increase awareness of the salient and sometimes subtle clinical and neurobehavioral features of SMS to promote early and accurate diagnosis;
- 2) Update current knowledge about the molecular basis for the syndrome and potential role of *RAI1* and/or other gene(s) that account for phenotypic variability;
- 3) Explore the biologic basis for paradoxical inverted circadian rhythm of melatonin that underlies disrupted sleep cycle;
- 4) Determine optimal intervention/management and treatment strategies to benefit individuals with SMS and families affected by this condition; and
- 5) Discuss the current status of animal models being used to study the pathophysiology of SMS.

The 5th SMS RR symposium was held at the Hyatt Reston (Reston, VA) Sept. 17th, 2009 in conjunction with PRISMS 6th International Conference on Smith-Magenis Syndrome, “Building Bridges of Hope” (Sept 17-20, 2009). Thirty-nine invited clinical and basic science researchers, professionals and clinicians currently involved with SMS in the US and worldwide (Canada, UK, France, Australia) attended the 5th SMS-RR. The SMS-RR program agenda included a morning session, *Lessons from the Bedside* moderated by PAB chair Ann C.M. Smith, MA, DSc(hon), that provided a clinical research update with 13 abstracts focused on expanding the SMS phenotype and family support issues and respite needs.

The afternoon session, *Lessons from the Bench*, moderated by PAB members Sarah Elsea, PhD, included six scientific presentations focused on the molecular genetic aspects of *RAI1* and other genes in SMS and current animal models (mouse, zebrafish). Researchers who attended the SMS-RR also spoke at PRISMS' 2009 conference, “Building Bridges of Hope” (Sept 18-20). This year's conference was the largest conference attended to date (over 330 attendees). Invited researchers/speakers spoke during the general plenary and/or workshop sessions and were available to all SMS families during “Curbstone Consults”. On Saturday, PRISMS' PAB is responsible for organizing and moderating the “*International SMS Research Update*”, a session that highlights cutting-edge research in this rare genetic syndrome stemming from the SMS-RR.

¹Parents and Researchers Interested in Smith-Magenis syndrome (PRISMS)

Recommendations/Outcome:

The Roundtable offers a unique collaborative forum for discussion that encourages strong cross-disciplinary exchange likely to enhance future research efforts. These efforts are vitally important in defining the natural history and clinical spectrum of SMS, ultimately leading to improved diagnostic, management, and therapeutic approaches likely to benefit persons with SMS and their families. *PRISMS* Professional Advisory Board (PAB) uses this forum to solicit expert input and consensus with respect to management guidelines and criteria for diagnostic confirmation of SMS. At the close of the SMS-RR, PRISMS PAB met in closed session to generate an action plan & priorities based on recommendations arising from SMS-RR discussions:

1. Approval of PAB revised SMS entry for GeneReviews for posting.
2. Need to develop a mechanism to record/track identified *RAI1* mutations/variants (database):
 - a. Commercial lab offering *RAI1* mutation analysis (GeneDX) and research labs (VCU, BCM, and NIH).
3. Develop a strategic research plan for 2010-2015 (SMS Research Alliance).
 - a. Need to encourage and foster collaboration/interest with other ‘non-SMS’ researchers/centers to consider SMS as a study population.
 - b. *SMS Research Alliance – Charting our Future for 2010-2015*:
 - i. Organize/plan a conference to identify the needs, explore the opportunities, and establish an action plan for research and collaboration for future SMS projects in target areas and/or other research “communities”
 - ii. Target areas of interest to SMS community: CAM (complementary & alternative medicine); behavioral research (impact of ABA); autism spectrum disorder;
4. Investigate the possibility of developing a student SMS research fellowship award – dependent on identifying funding source (PRISMS/Other grants)