



Fall 2006

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2006 Annual Meeting A Huge Success!!

170 people from all eight states in our region and two other regions attended the meeting in Denver on July 13th-15th. This made the 2006 annual meeting the most well attended to date. The presentations, committee meetings and social events were well received and based on the evaluations, this meeting was a huge success. If you were not able to attend this year, please visit the website at www.mostgene.org to download many of the presentations. Planning has begun for next year's meeting. Be sure to put it on your calendar. It will be held on July 12th -14th in the Denver metro area. The feedback from the evaluations is most helpful in planning for next year's meeting. Please check the website for updates on midyear meetings and all MSGRCC events.

Annual Meeting presentations available to view from the website include but not limited to:

- Issues in the Transition from Pediatric to Adult Genetics
- Surveillance Strategies for Genetic Disorders
- Clinical and Technical Aspects of Hematological Malignancies
- The Role of Geneticists in the Hurricane Katrina DNA Identification Effort
- Maternal Serum Screening
- Much More!!



[Committee Chair Election Results](#)

Please welcome the new committee chairs that will serve the MSGRCC through the 2008 annual meeting. They are appreciated for their willingness to give time and effort to MSGRCC. The outgoing chairs are also appreciated for all the time and expertise they contributed. Please assist your new chairs in their efforts to make each committee viable and productive. Don't hesitate to contact them with any questions or ideas you have. Remember, this is YOUR region, and the committees are here to support and enhance the services we provide.

The New Committee Chairs Are:

Clinical Services: Co-Chairs: Joanne Milisa-Draught and Marc Williams, M.D.

Consumer Advocacy: Co-Chairs: Rod Slaght and Joe Martinec

Education: Chair: Mary Esther Carlin, M.D.. Vice-Chair: Teresa Gehrke

Laboratory Practice: Co-Chairs: Sheila Dobin, Ph.D. and Linda Sheppard

Newborn Screening: Chair: Rebecca Anderson

Prenatal Diagnosis: Co-Chairs: Janice Rinsky and Valerie Rappaport, M.D.

Public Health: Chair: Lynn Martinez

Mostgene Website Has New Host

It is exciting news that the RCC has a new website host who is better equipped to meet our needs. The website has been updated and expanded. It includes a more comprehensive Genetic Services Directory, updated job listings, a link for each committee, updated and expanded information, and more. We have also implemented a listserv for each of our seven committees. If you have designated a primary committee, you should have received an invitation to join the listserv. If you have not, please contact Janice Rinsky and she will advise you on how to join. Also, if you have any ideas for the website, or know of a link you think should be included, please contact Janice at Janice.Rinsky@mostgene.org or 720-939-2505 to let her know.

E-Mail: Point of Diminishing Returns?

In the era of high technology and rapid communication, we have all come, to some extent, to rely on e-mail in our professional and personal lives. E-mail is a wonderful tool, but in recent years it has been used to send unwanted material, hence the spam blockers, firewalls, filters, etc. MSGRCC relies on e-mail for communications. However, due to the tools used to block unwanted e-mail, messages are often not delivered to the intended recipient. To further complicate this, the e-mail may be delivered to an institution, a confirmation of this is received, but it may never reach the intended recipients computer due to the e-mail protections in place. It is important for all members to be kept abreast of what is happening in their committees and the MSGRCC as a whole. The new website host has put some tools in place to assist in getting e-mail to its intended recipient. However, if you know your institution has filters that may prevent e-mail from reaching you, please let Janice Rinsky know so that the problem can be addressed. E-mail will be kept to a minimum. If you have any questions or know that you are having problems receiving e-mail from MSGRCC, please let Janice Rinsky know. She can be reached at 720-939-2505 or Janice.Rinsky@mostgene.org. Thanks so much.



Update of Year 2 Selected Grants for Funding

Selection Committee: *Dr. John Johnson (Clinical), Dr. Gurbax Sekhon (Laboratory), Lynn Martinez (Public Health). These individuals were chosen for their expertise and knowledge of related activities in their sphere of influence. Joyce Hooker assisted as Director of the Mountain States Regional Collaborative Center.*

Mile High Down Syndrome Association of Denver: The Mile High Down Syndrome Association (MHDSA) Health Care Partnership was developed in 2004 in response to the concerns of our members that they had not received the information and support they needed at the time of their children's diagnosis. Many also indicated that their children's health care providers did not have the knowledge, skills and resources needed to provide the level of care they required. The Health Care Partnership Coordinator is both a pediatric care provider and a parent of a child with Down syndrome, and is therefore able to apply both the parent and the professional perspectives to educate and offer resources to health care providers regarding Down syndrome. This project is providing education conducted through face-to-face meetings, in-services, presentations, guest lecturing at local universities, published articles, and the provision of very practical resource material designed for health care providers. It too is a model that can be adopted by others in the region. **PI: Sarah Hartway, Mile High Down Syndrome Association of Denver.**

University of Colorado Cancer Center: This project is providing educational outreach about genetic testing to individuals at risk for hereditary colon cancer. It is to educate individuals who are at-risk for hereditary colon cancer about the potential benefits of cancer risk assessment and to provide genetic services (risk assessment) to individuals without access. It is using the Bethesda Criteria for Hereditary Non-Polyposis Colon Cancer and the Colorado Central Cancer Registry (Colorado Department of Public Health and Environment). An objective is to determine how useful the registry data is and how it can best be used. Educational materials have been developed and include an introduction letter, educational brochure about hereditary colon cancer, and information on how to obtain a cancer risk assessment. Feedback on the material was obtained from focus groups. Currently a web-based application for telephone risk assessment is under development. This will be an interactive web site where the user and counselor can view visual aids simultaneously during the counseling session. Topics covered will include characteristics of hereditary cancer, cancer risks for hereditary cancer syndromes, review of family history and risk of hereditary cancer and the testing process. Again, this model project can be implemented in other states and the lessons learned will apply to other genetic disorders. **PI: Jan Lowry, M.D., University of Colorado Cancer Center.**

University of Utah/Utah Department of Health: The Mountain States Genetics Regional Collaborative Center is funding a planning project to develop a longitudinal regional/national surveillance system for children affected with conditions identified through newborn screening. Dr. Jeffrey Botkin at the University of Utah is the PI on the project and Rebecca Anderson at the University of Utah is the Project Director. While metabolic conditions will be the initial focus of the planning, the intent is to extend the surveillance system to include hematologic, endocrine, and other conditions on NBS panels. Data acquisition will occur through subspecialty clinics, the medical home, hospitals, and from family interviews. Independent variables and outcome domains include demographics, clinical and biochemical measures, developmental status, psychosocial measures, school performance, and economic impacts. Preliminary discussions about a tissue repository for selected conditions have occurred. The surveillance system will create a minimal data set and an interactive resource for clinicians and families, NBS programs, the NBS system and investigators. **PI: Jeffrey Botkin, M.D., MPH, University of Utah.**

University of Arizona: The goal of the project is to increase knowledge about genetics and genetic services with the ultimate aim of improving access to genetics information and services in the region. This will be accomplished through the development and implementation of consumer education programs for the general public, and separate presentations created specifically for and offered in locations accessible to the Native American population and supportive web-based resources for these programs. The methods employed by this project are lectures and forums on topical subjects in genetics for the lay public, public forums on genetics and genetic diseases in at least three Native American communities and invite members of the communities to attend and discuss the information presented. A web-based site is being created for the content of educational activities that will serve as a continuing education opportunity for consumers in our region and beyond, as well as genetics students, faculty, and professionals. **PI: Dr. Murray Brilliant, University of Arizona**

The Children's Hospital of Denver: Dr. Marci Sontag is assessing the current variability in experiences with receiving genetic information (presence/absence of any form of 'genetic counseling', which type of healthcare provider delivered, quality of and retention of information) of individuals identified as being heterozygotes for cystic fibrosis mutations. A website has been developed, (www.cfcarriers.org). The methods implemented by this project are an electronic survey, postcards, and webcasts. The instruments and material developed for this project are available on the website. **PI: Dr. Marci Sontag, The Children's Hospital of Denver.**

University of Colorado Sickle Cell Treatment and Research Center: This project is creating a system to reduce financial and access barriers to these services in an effort to determine the actual community interest in sickle cell trait counseling in the Mountain States Region. By piloting programs through the public health community clinics, educating and integrating local public health professionals and trained lay community members, a self-sustaining model of genetic counseling for sickle cell trait and other genetic carrier states will be developed that may be applicable to other states in the region. Specific accomplishments toward the creation of outreach genetic counseling services that have; The identification of, initial contact with and visit to community health clinics; Completion of training of two lay community members as sickle cell trait counselors at the most recent national training course offered in New Britain, Connecticut in May, 2006; Advertisement to families of infants recognized to have sickle cell trait by newborn screening with letters to providers with notification of positive screens. Notification of outreach programs will occur by direct phone call from the newborn screening follow-up programs during routine follow-up contact. **PI: Kathryn Hassell MD, Colorado Sickle Cell Treatment and Research Center.**

Texas Tech University Health Sciences Center: GENE-ARM is a pilot project to inform and empower nurses to recognize, refer, and guide management of genetic disease. The project has established a GEN-ARM website and central email/phone. A brochure has been designed in consultation with MSGN personnel and nurse consultants to alert nurses in various specialties to key genetic issues and solicit their participation in the GEN-ARM network. Nurse facilitators at major hospitals or centers who have some interest in genetics have been identified. These facilitators will be encouraged to give in-service presentations and distribute the brochures to nurse attendees. Local meetings and in-service presentations through OB genetics nurses and a pediatric nurse are underway. A manual is being developed with input from MSGRCC administrators and GEN-ARM nurse consultants and will emphasize case presentations that nurses should recognize, based on realization that assessment is a key part of the nursing role. The manual will list available genetics courses for nurses, from particular medical centers, our own CEU video being produced at Texas Tech HealthNet, and on the ISONG website. **PI: Vijay Tonk MD, Texas Tech University Health Sciences Center.**

The Children's Hospital of Denver: This project is providing a neurodevelopmental review of long-term outcomes for Recombinant 8 syndrome children. It is a model for replication in the region to understand other genetic disorders. This syndrome occurs with greater frequency in children in the Mountain States Region. This study is expected to reach its targeted enrollment by Feb 2007. The data from the project will then be ready for publication and presentation few weeks after the targeted enrollment has been met. **PI: Anne Chun-Tsai MD, The Children's Hospital of Denver.**

Identification of Possible Shifting Communities at Risk for Sickle Cell Trait: This project examined the shift in the at-risk population (Latinos) for sickle cell trait and disease. Latinos are the fastest growing population in most of the states in this region. Knowing which populations are more at risk for this condition will aid in more focused outreach and education in these communities. A Retrospective cohort of infants with an initial positive test for hemoglobinopathy trait was identified using the Colorado Sickle Cell Treatment and Research Center data from June to July for the years 1984-1985, 1994-1995, 2000-2001 and 2004-2005. An increasing trend of Hispanics identified with sickle cell trait from 1985 to 2005 was evident, this is significant at the $p=0.01$ level. The increase in the Hispanic population is not just within this specific population but throughout the entire state of Colorado. According to CDPHE Colorado Health Information Dataset (COHID) there were 11,522 babies born to Hispanic mothers in 1995 as compared to 18,231 in 2000 (a 37% increase) and 21,738 in 2004 (a 16% increase from 2000). According to these numbers babies born of Hispanic mothers make up a larger proportion of the general birth cohorts from those years similar to this cohort of babies (21% in 1995, 28% in 2000, 32% in 2004). The increase of the number of babies identified with sickle cell trait and all hemoglobinopathy trait can be noted by the percent increase of the Hispanic population year to year and not necessarily by an increase rate of Hispanic babies born with sickle cell trait in the general population.

The realization that about 14% of the population is not confirmed brought about another concern including which populations are not coming in for the follow-up confirmatory test. Further investigations have shown that not only is the Hispanic population with 15.43% not coming in for a follow-up, confirmatory test but also the Caucasian population with 14% not coming in for a follow-up, confirmatory test. Dr. Hassel has concluded that perhaps the spread of education past the African American population would cause these percentages to come closer to that of the African American population (11% not confirmed). **PI: Kathryn Hassell MD, University of Colorado Sickle Cell Treatment and Research Center.**

Analyte Stability: Dr. Pasquali's project is evaluating the stability of analytes and enzymes measured in blood spots by tandem mass spectrometry (MS/MS) and by traditional methods of screening (biotinidase deficiency, congenital adrenal hyperplasia (CAH), cystic fibrosis (CF), congenital hypothyroidism (CH), and galactosemia). To date analytes have been measured at 11 time points from day 0 to 9 months. The preliminary conclusion is that Acylcarnitines and amino acids in blood spotted on filter paper are stable at least up to 9 months in most storage conditions. The storage conditions with the best rank are below zero degrees. Enzymes (Ex. GALT) levels rapidly decline if blood spots are stored at RT or refrigerated. Storage of blood spots at -0 °C preserve analytes and enzyme activity. **PI: Marzia Pasquali PhD and Jeffery Botkin MD MPH**