

Celia Kaye, MD, PhD

The possession of knowledge does not kill the sense of wonder and mystery.
There is always more mystery. ~ Anais Nin

Wonder and mystery. Perhaps the very things which drew Dr. Celia Kaye to the study of genetics; “When you see disease at the cellular level, the biochemistry is right there. It is pathology at the biochemical level. It is where understanding begins.”

Dr. Kaye, M.D., Ph.D. serves as Principal Investigator of the Mountain States Genetics Regional Collaborative Center (MSGRCC), one of seven regional collaborative centers in the nation.

The MSGRCC is federally funded by the US. Department of Health and Human Services, Health Resources and Services Administration (HRSA) Genetic Services Branch and administered by Texas Health Institute. The MSGRCC provides infrastructure for state genetics programs and capacity building for regional genetic and newborn screening collaboratives which strive to improve early identification of infants. The regional center includes the states of Arizona, Colorado, Montana, New Mexico, Nevada, Texas, Utah and Wyoming.

Dr. Kaye is a pediatrician, geneticist and educator with faculty appointments as clinical professor at the University of Colorado Denver School of Medicine and the University of Texas Health Science Center at San Antonio. She is also Senior Genetics Advisor at the National Newborn Screening and Genetics Resource Center.

Dr. Kaye began her career earning her M.S. and M.D. from Wayne State University in Detroit, Michigan and her Ph.D. from Northwestern University in Evanston, Ill. She has practiced as a clinical and metabolic geneticist, and is the author of numerous original articles, chapters and educational manuals. She is the mother of two children and grandmother of three. Her husband is a full-time faculty anesthesiologist at the University of Colorado Denver School of Medicine.

Balancing her personal life with her academic career and the numerous clinical studies which she has become involved in, Dr. Kaye took a few moments to discuss her involvement in the MSGRCC:

Why is MSGRCC important? “MSGRCC plays the important role of taking new knowledge and information to where the people are. While genetic research is funded and digested at the national level, it takes organizations like MSGRCC to get that information to people at the local level, to physicians and consumers.”

Regional Collaborative Centers operate on small budgets but function as the intermediary, transferring information to the physicians and people at the local level.

“In addition to making sure that good information gets to real people, MSGRCC also functions to ensure that no one needing services gets lost in the search for quality healthcare,” says Dr. Kaye. In many cases, Regional Collaborative Centers help families needing services find the right provider of those services. By providing resource information and education, MSGRCC helps make connections possible.

More than information. MSGRCC also provides funding for genetics projects based upon HRSA’s national focus criteria. MSGRCC selects projects for funding with two things in mind: talent and need. Is there sufficient talent and expertise to perform the proposed project, and is there significant need to pursue the project?

One project MSGRCC funds is a hot topic on the national genetics front: shared data sets, which are a key factor in the Newborn Screening Outcomes Project headed by Dr. Janet Thomas. Dr. Kaye says that the shared data sets in this project help determine treatment paths which may lead to healthier and more productive long-term outcomes for infants with inborn errors of metabolism diagnosed through newborn screening.

To further those health and productive outcomes, Dr. Kaye notes the work of another MSGRCC project, headed by Dr. Laura Pickler. The project is working to identify and connect provider resources to genetics patients as they move from pediatric care to adult treatment needs. Because many of the metabolic diseases are rare, identifying physicians with training in family medicine or inherited metabolic disease who feel qualified and are willing to treat them is difficult.

“Our region is blessed with some outstanding primary specialty care providers – our issue is geography,” says Dr. Kaye. “We have huge areas of dispersed populations within the region which makes access to care all the more difficult and may cause families already struggling with the economics of care to face the burden of ongoing travel expenses.”

The eight states in MSGRCC region have a combined area of 1,081,813 square miles and a population of more than 38 million people. While there are major urban areas in the region, the overall population density is about 37 people per square mile, approximately one half of the national population density per square mile.

Genetics today and beyond. While the focus of many of the MSGRCC projects is overcoming obstacles in the early identification and treatment of genetic

disorders, Dr. Kaye is encouraged by opportunities she sees on the horizon for improving the health and health care of persons with genetic health challenges. She says personalized medicine has shown clear promise of health and economic benefits. “When we have a better understanding of a person’s genome, we may have a better understanding of how to treat them. Medication metabolism, for example: We may be able to tailor the use or dose of some medications if we understand the genetic factors that influence metabolism.” Kaye says treatment with the right drugs at the right time will save lives, improve overall health and will reduce unnecessary treatment and diagnostic costs.

The idea of personalized medicine, combined with a medical home environment, brings a smile to Dr. Celia Kaye’s face, energizing her in the love and challenge of her career, understanding how cell biology and genetics can become tools to improving health and healing.