MUSC Center for Genomic Medicine

Mission:
The mission of the Center for Genomic Medicine will be to promote and support basic and clinical research and training in genetic and genomic influences on human health and disease, and to translate these discoveries into clinical practice.

Goals:
The role of genetics in the practice of medicine today ranges from diagnostic testing for hereditary disorders to the recognition that the development, diagnosis, and treatment of many diseases are influenced by an individual's genotype. In addition, it is becoming increasingly clear that environmental factors, ranging from chemical pollutants and drugs to sensory stimuli, influence genetic processes. The rapid development of molecular technologies has enabled the screening of tens of thousands of genes for their variants, expressions, interactions, and assembly into complex functional networks. Recent advances demonstrate that modifications of gene transcription and translation are important for health and disease. Finally, it is recognized that the translation and back translation of the human condition, to animal models utilizing genetic background and modifications, will be necessary to fulfill the promise of truly understanding differences in human disease and treatments.

The MUSC Center for Genomic Medicine will provide the leadership, expertise and core resources to facilitate the acquisition and application of genetic and genomic information to research and to the practice of medicine across the MUSC campus and throughout South Carolina.

Center Leadership and Faculty:
The identification and recruitment of a Center Director with the experience and vision to lead a multi-disciplinary program that provides access to Center resources across the MUSC campus is essential for the success of the Center. The Center Director should be a senior, well-funded, internationally recognized investigator with a research program that complements strategic research areas within the College of Medicine. To ensure that Center resources are accessible to all faculty, the Director must report directly to the Dean of the College of Medicine.

The complexity of genetic/genomic research today also requires specialized leadership within key core resource areas. Hence, the identification of core directors for Informatics/Statistical Genetics and Molecular Genetics is also essential for the ultimate success of the Center.

A final component necessary for the Center’s success is the recruitment of a critical mass of researchers with a genetic/genomic focus across the College. These recruitments should be considered a priority by Departments, the College of Medicine and the University. Efforts in this area should include identification and prioritization of faculty with research programs identified as essential for the Center’s growth, and coordinated recruitment and start-up packages.

Center Space:
A central requirement for the development of a Center for Genomic Medicine is the assignment of dedicated space to house the Center’s core facilities. Ideally all core facilities and administrative offices should be housed in adjoining areas. Center space should have reasonable access to appropriate vivarium and clinical facilities.
Core Resources:
The establishment of core facilities within the Center is an essential element for meeting the genetic research needs of the faculty within the College of Medicine and across the MUSC campus. These core facilities should include: Administrative, Informatics/Statistical Genetics and Molecular Genetics. To provide needed services to College faculty and avoid conflicts of interest, core staff and scientists will report to the Center Director. Other associated core facilities that would strengthen genomic research include: biorepository, proteomics, lipidomics, clinical data warehouse, and transgenic mouse and zebra-fish facilities.

Administrative Core
The operation of genomic core facilities will require administrative assistance for general operation of the Center and its Cores. In addition, the administrative core should serve as a resource for study design and execution as well as faculty and student education.

Informatics/Statistical Genetics Core
The informatics/statistical genetics core is an essential element for meeting the genetic and genomic research needs of the faculty within the College of Medicine and across the MUSC campus. The bioinformatics and statistical genetics groups within the core are tightly related in supporting genetic/genomic research. The infrastructure required for this core should include hardware with large storage capacity making it possible to develop and implement computational tools necessary for analyzing data generated from genetic/genomic studies, such as genome-wide association studies and Next-Gen sequencing studies. The core should consist of faculty trained in computer science, computational biology, statistics, and epidemiology, whose research efforts will broadly support discovery of genes implicated in disease pathways. Obtaining faculty and qualified supporting staff with appropriate expertise and track records for the bioinformatics group as well as the statistical genetics/genetic epidemiology group within the core, is a critical and early step in the development of the Center. Each group within the core may require its own leader, if a director for the core with expertise in both bioinformatics and statistical genetics cannot be identified.

Molecular Genetics Core
The molecular genetics core should provide open access to genetic/genomic technologies and technical expertise in the areas of genome services, functional genomics, and diagnostic and customized medicine. Genome services could include: high throughput DNA sequencing, such as whole genome sequencing, exomic sequencing, chromatin immunoprecipitation (ChIP-Seq), whole transcriptome analysis (RNA-Seq, exome sequencing), genotyping, genome-wide methylation analysis, microarray analysis for DNA copy number variation, mRNA expression, microRNA, and DNA-SNP. Functional genomics could include: RNAi screens and synthetic lethal screens using shRNA expression libraries. Diagnostics and customized medicine could include: the ability to do single or small numbers of SNPs and variable nucleotide tandem repeat assays both for diagnosis or prediction. There will also be a need to evaluate post-transcriptional modifications for discovery, diagnosis and treatment.

Clinical Medical Genetics
Medical genetic and genomic research begins with access to information on patients and families. The Center should support Clinical Geneticists specializing in evaluation, diagnosis and counseling for individuals with genetic disorders and their family members; Biochemical Geneticists who describe the underlying biochemical defects associated with
human conditions; and Cytogeneticists and Molecular Geneticists who oversee clinical
genetic testing to identify sequence variations (genotypes) associated with disease. As the
Center matures, it should aid faculty in the development and coordination of individual and
family-based genetic studies, and provide consultative services to investigators and
assistance with patient recruitment, preparation of IRB protocols and informed consent
documents, gene mapping and identification, and development of genetic testing for
genotype-phenotype studies. The Center could also provide for the translation of genetic
and genomic discoveries to the clinic by assisting investigators with access to clinical
laboratory test development services.

**Education and Training:**
The Education and Training program should promote the use of the most current molecular
biology tools in biomedical research, in part by holding numerous workshops and short courses
throughout the year. Workshops would target graduate and medical students, fellows and
faculty members who are interested in applying genetic and genomic methodologies to their
research but may not have the necessary experience or those who may want to explore the
latest techniques. Instruction would be provided by Center staff or faculty and invited experts in
a semi-formal, hands-on setting. Workshop topics could include: Basic Recombinant DNA
methods, Genomics, Molecular Markers, DNA Forensics, Differential Gene Expression,
Genomic Experimental Design and Analysis (Microarrays, Next-Gen Sequencing), In Situ
Hybridization, Statistical Genetics and Bioinformatics. Additional courses that could be
produced in partnership with other units on campus might include: Protein Chemistry/
Proteomics, Glycobiology, Lipidomics, Laser Capture Microscopy, and Advanced Imaging.

The Center could also host a genetics training/mentoring program to link young faculty/fellows
with established senior faculty to promote successful genetic research programs. This would
promote cross-disciplinary and cross-department/university-wide collaborations.

A final component of the educational program could be to expand training opportunities in
genetics by working with the Colleges to determine the feasibility of establishing graduate
program(s) in genetics and clinical residencies/fellowships in Clinical Medical genetics.

**Technology Transfer and Innovation:**
The Center for Genomic Medicine is a research-based center. Hence, its ultimate success will
be judged by its faculty’s ability to discover new therapeutic targets and diagnostics and
integrate these findings into the practice of medicine. This technology transfer will occur in two
ways. First, in partnership with MUSC’s Center for Medical Innovation and Entrepreneurialism,
the Center will work to bring the ideas and technology of its faculty into public use by supporting
the patenting, and licensing of discoveries. Second, in partnership with the CTSA, the Center
will support the translation of basic science discoveries into the practice of medicine.

**Coordination with Regional Partners:**
No single center has the resources to develop expertise in every facet of genetics and
genomics. Therefore, the Center will develop partnerships and liaisons with both regional and
remote facilities with extant expertise to deepen the resources available to MUSC investigators.
The following extramural resources have been consulted and are enthusiastic about forming
stronger alliances with MUSC researchers: University of South Carolina, Clemson University,
Greenwood Genetic Center, and the David H. Murdock Research Institute.