

# SC Rare Disease Symposium



**February 29, 2024**  
**Program Agenda**

**9:30** Check in / Registration

**10:00** Welcome & Opening Remarks

*Patrick Flume, MD*, Endowed Chair, Power-Huggins Endowed Chair for Cystic Fibrosis & Chair for the Rare Disease Advisory Council

**10:10** Clinical Trials & Patient Engagement

*Patrick Flume, MD*

**10:30** Research Presentations: Innovation and Discovery within South Carolina

**The Evolving Role of Genetic Testing for Rare Disorders**

*Dan Judge, MD*, Director of the Cardiovascular Genetics Program & the Fellowship Director for Cardiovascular Disease, MUSC

**The Changing Landscape of Newborn Screening: What's New and What's on the Horizon**

*Neena Champaigne, MD*, Division Chief of Pediatric Genetics, MUSC

**A Genetic Counselor's Role in Rare Disease**

*Maysen Mesaros, MS, CGC*, Genetics Counselor for Neurology, MUSC & *Olivia Thompson, MS, CGC*, Genetics Counselor for Pediatrics, MUSC

**Transforming Care and Research for Those with Rare Diseases: Start by Listening to Patients**

*Russell "Chip" Norris, PhD*, Professor of Regenerative Medicine and Cell Biology, MUSC

**12:00** Q&A Research Presentations

**12:15** Break / Pick Up Lunch

**12:30** Advocacy Panel

**Moderator: Karen Kemper, PhD**, Advocate for Scleroderma & Associate Professor of Department of Health Services, Clemson University

**Yvonne Donald**, Advocate for Sickle Cell Disease & Executive Director of the James R. Clark Memorial Sickle Cell Foundation

**Miranda McManus**, Advocate for Neurofibromatosis & Member of the Directors Council for the Response Evaluation in Neurofibromatosis and Schwannomatosis International Collaboration

**Kerri Nelson**, Advocate for Mosaic Trisomy 22 & Former Ambassador of the National Organization of Rare Diseases South Carolina Rare Action Network

**LaKesha Williams**, Advocate for Scleroderma & Support Group Facilitator for the South Carolina Piedmont Chapter of the National Scleroderma Foundation

**1:30** Q&A Advocacy Panel

**1:50** Wrap Up/Closing Remarks

