



Mark Hughes, MD, PhD
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Professor Mark Hughes graduated in Biology and Chemistry from **St. Johns University** followed by a Masters in Biophysics at **Stanford University** and a Ph.D. in Molecular Biochemistry at the **University of Arizona** Medical Center. He continued his training at the **Baylor College of Medicine** in Houston as a postdoctoral fellow with Bert O'Malley, where his pivotal work was published in *Science and Nature* and involved the cloning of the vitamin D and progesterone receptors and characterization of the first mutations found in human gene transcription factors. Mutations in the "tips" of zinc fingers of the vitamin D receptor were identified in the DNA of patients with rickets. These were the first mutations identified in any human gene transcription factor. Following this training Hughes completed his M.D. at Baylor, followed by house staff training in Internal Medicine and clinical subspecialty training at **Duke University**. He then returned as junior faculty to Baylor's newly formed Genetics Institute led by Thomas Caskey and Arthur Beaudet. Among his accomplishments was the realization that single cells could be molecularly data mined for diagnostic advantage: This led to a multi-year collaboration with reproductive endocrinologists and embryologists at the Hammersmith and UCLondon; the field of Preimplantation Genetic Diagnosis was born. *In 1993 Hughes' research was recognized by Science magazine as being one of the "ten most significant advances" in all of science that year; spanning all the physical, biological and mathematical sciences for that year.*

It was then that Professor Hughes was recruited to be one of the first 11 members of the **Human Genome Institute at NIH**. The Genome Project was getting underway and Hughes was recruited to lead the section on Translational Genomic Diagnostics. He also was named chair of Human Genetics at **Georgetown University**. Doctor Hughes then moved to Michigan to take a position as Professor and Director of Molecular Medicine and Genetics, Professor of OB-Gyn, and Professor of Pathology at Wayne State. He was named as the Director of the State of Michigan's 'Life Sciences Genomics Hub', a joint state-wide project with the University of Michigan and Michigan State University.

Hughes' work has centered on understanding gene expression in the early human embryo. His work on embryonic stem cells was acknowledged in 2001 when, along with Ian Wilmut (of Dolly the sheep fame) Hughes was awarded the "Pioneer in Stem Cell Biology" award. Professor Hughes, along with Professor Lord Robert Winston and Dr. Alan Handyside developed and performed the world's first cases of has PGD. As we know, this field is now practiced world wide – today's speaker continues to push the frontiers of this technology and guide it in all its ethical ramifications, while he has expanded this work to systems-wide molecular understanding of early embryo development. In January he co-authored the first proven somatic cell nuclear transfer involving human somatic cells and oocytes (in other words, these scientists actually did what the South Korean group lied about). His clinical/scientific goal has been to better understand, and hopefully prevent, many inherited birth defects of children. You may have seen him on the two hour BBC special last month. He has appeared on "Good Morning America", the "Today show", "CBS Evening News", and the subject of television newsmagazine segments for 60 Minutes and 20/20, and full hour programs on the Discovery Channel. His PGD work to assist couples avoid serious disease in their children and, at the same time, obtain a stem cell cure for a sick child already in the family, has gained world-wide attention. Most recently, his group has begun a program for patients with inherited neoplasia. Patients (250) undergo fertility preservation with oocyte/embryo genetic analysis prior to cancer treatment. Because of federal funding limitations on embryonic stem cell science, he moved the clinical PGD aspects of his work into the Genesis Genetics Institute where this technology is provided in concert with some 470 human reproductive centres in North and South America, Europe, Africa and Asia.