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Mark Hughes graduated in Biology and Chemistry from St. Johns University and then a Ph.D. in Molecular Biochemistry at the University of Arizona Medical Center. He continued his training at Houston's Baylor College of Medicine where, as a postdoctoral fellow in the laboratories of Bert O'Malley, his pivotal work published in Science and Nature, involved the cloning of the vitamin D and progesterone receptors and characterization of the first mutations found in a human gene transcription factor. This work began his career in molecular endocrinology and genetics. Following this training, Professor Hughes completed his M.D. at Baylor, followed by housestaff training in Internal Medicine and a clinical subspeciality training at Duke University. He then returned as junior faculty to Baylor's newly formed Genetics Institute led by Thomas Caskey. 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 IVF clinicians and embryologists at the Hammersmith Hospital in London; the field of Preimplantation Genetics 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.

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. Human He also headed 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. He was named as the Director of the state's 'Life Sciences Genomics Hub', where genomics, proteomics and bioinformatics merge in the field of molecular medicine.

Hughes' work has centered on understanding gene expression in the early human embryo. His pioneering work the field of PGD has expanded to systems-wide molecular understanding of early embryo development. Two years ago, he moved the PGD aspects of his work into an Institute where the diagnostic aspects of PGD are provided to 132 human reproductive centers in North America and Europe.

Preimplantation Genetic Diagnosis 2004: The Technology, The Medicine & The Future Promise

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The completion of the Human Genome Project heralds a new era of Functional Genomics. Raw DNA information that comprises the blueprint of human life is being data-mined, taken apart, spliced together, and injected into cells, embryos and animals in ways we could barely imagine just a decade ago. The promise is for new medicines, predictive diagnostic tests, and stem cell therapies. The potential for societal, legal, and ethical uses/abuses of this powerful information is especially strong in reproductive genetics involving human embryos and the developmentally totipotent cells derived from embryos. Most every American has a visceral and reflexive response regarding these promising yet troubling technologies. One such new technology is called "Preimplantation Genetic Diagnosis" (PGD) of the human embryo, which combines the technologies of IVF, embryo culture and biopsy, and single-cell molecular and cytogenetics. It provides couples at high genetic risk the opportunity to begin their pregnancy on day-one, with the knowledge that their fetus will not have the inherited disorder that afflicts their family. No longer do they need to throw the genetic dice, take a chance, and hope for the best. This presentation will describe the science and medical application behind this technology.

This "hope" of a decade ago is now in daily clinical practice around the world. As we learn more about human genome 'function,' and as we increasingly identify gene mutations causing inherited disease, there will be more utilization and acceptance of this technology. It is now possible to test single cells derived from cleavage-stage embryos for over 1,200 different genotypes. It is now routine to test simultaneoulsy for dozens of single nucleotide polymorphisms (SNPs). Gene Chip technology currently provides the ability to examine numerous DNA alterations in a single haploid or diploid cell, and this technology is moving from the research bench to the clinical bedside. In addition, cytogenetic aneuploidy and chromosome translocation testing is widely in use in the fertility center. These technologies will improve, but the biology of multi-gene segregation will not. A reasonable prediction for the future will allow women to test her oocytes when she is young, identify those without disease and cryopreserve them even before she has a male partner, and well before the inevitable decline of ovarian function. This would alter society like nothing since the release of oral contraception. Technology will remain the fuel that propels this science. Science will always be the engine driving medical advances, and medicine will continue, as it has through history, to steer us into the bioethical corner of new "opportunities." As we translate the secrets of the human genome, and as we move this information into proteomics and the story of embryonic human life, this newfound knowledge will dazzle & trouble, amaze & alarm. It will offer irrational hyperbole to some, and the "miracle" of a healthy family to many others.