THE GENETICS OF MALE INFERTILITY
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Edited by

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Male infertility is a common and severe health problem. Infertility not only affects one’s ability to have children, but also has emotional, psychological, family, and societal effects. Despite the prevalence and significance of this health problem, resources and attention have not been sufficiently focused on this important issue.

Approximately 7% of men suffer from infertility, and the incidence may be increasing. Of those affected, roughly 40% have idiopathic infertility. It is likely that the majority of those patients have genetic abnormalities that are the cause of their infertility. However, it is important to remember that there are genetic ramifications for essentially all infertile male patients. For example, it is likely that there are genetic predispositions to pathologies such as varicoceles, and environmental factors almost certainly modulate the underlying condition. The understanding of the genes involved in spermatogenesis, sperm maturation, and normal sperm function is key, but we must also focus on better methods of accelerating advances into meaningful clinical diagnostic tests and therapies.

During the past 20 years, significant improvements in technology have advanced the treatment of male infertility. The primary advance has been intracytoplasmic sperm injection (ICSI) in conjunction with in vitro fertilization. Although this technological leap has allowed thousands of men to father a child who otherwise would have been unable to do so, the scientific study of the causes of male infertility has not kept pace. In fact, the clinical application of ICSI proceeded without sufficient scientific study of its safety to the offspring, or the future genetic ramifications.

We currently stand at a point in history in which new tools are available to evaluate genetic diseases. The completion of the Human Genome Project has ushered in an era of unprecedented momentum and ability to tackle the complex issues in the genetics of male infertility. New tools include in vitro methodologies, *in silico* technologies, and new model organisms. Together these advances portend great possibilities.

In January 2006, an international symposium was held at the University of Utah Campus in Salt Lake City to address the genetic causes of male infertility and the translation of the knowledge to the clinical realm. Twenty-one researchers and clinicians, and an international audience of
experts in the field, reviewed the study of the genetics of male infertility, the tools available in the laboratory and clinic, the current state of knowledge, and the future of research and translation into clinical diagnostics and treatments. This book is the result of the symposium. The book is intended as a review of our current understanding of genetic causes of male infertility, a guide to evidence-based clinical applications, and a preview of future possibilities.

Douglas T. Carrell, PhD
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