Textbook of Human Reproductive Genetics

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Preface

Genetic advances in the reproductive sciences are arguably occurring with greater rapidity than those in any other organ system. Not only have the dazzling technological advances of molecular genetics become applicable in reproduction, but meteoric advances are occurring in diagnosis and treatment. Assisted reproductive technology (ART) has rendered infertility far less daunting. Yet treating those couples, that once would not have conceived, may generate offspring who differ from the general population.

Textbook of Human Reproductive Genetics addresses pivotal topics of clinical and scientific interest. There is much for the student, practicing physician, and laboratory scientist alike. Crisply edited by renowned geneticists of international repute – Stéphane Viville and Karen Sermon – the book begins with a précis of genetic principles – molecular, single gene, and cytogenetic. The novice quickly gets up to speed.

The basic science landscape targets fields of most immediate relevance to reproduction. Techniques suitable for analyzing a single cell are explained, one cell's (6 picograms) DNA obviously necessitates different approaches than if larger amounts of DNA were available. Thus, Kumar *et al.* explain where we are now and where we will soon be (microarrays and next generation sequencing). Cell division and the consequences of its perturbation are framed, respectively, by Eichenlaub-Ritter in her chapter on meiosis and by Baart and Van Opstal in their chapter on the role of aneuploidy in human embryonic development. Yatsenko and Rajkovic extend the dialogue to cytogenetic disorders affecting infertility. Monogenic causes are not neglected, Liebaers and colleagues covering infertility in pleiotropic presentation. This is especially relevant because it has become clear that common conditions like premature ovarian failure or polycystic ovarian syndrome are heterogeneous. Thus, looking at rare genes whose perturbations cause syndromes could be a fruitful strategy in identifying more common disorders like premature ovarian failure.

Van Monfoort covers epigenetic phenomena as related specifically to ART. The basic science component of this text also lets us know that genes must be *expressed*, without which it matters little whether they are present or absent. To this end, Balakrishnan and Chaillet discuss transgenerational effects mediated by epigenetic alteration. Fauque and Bourc'his shed light on transposons as newly appreciated determinants of male reproductive fitness.

The scientific framework having been established, specific clinical aspects of human reproduction are then systematically addressed. Traeger-Synodinos and Staessen cover clinical preimplantation genetic diagnosis. Individualized ART tests are discussed by Verpoest. Lashwood and Bagshawe review genetic counseling, providing not only traditional "how to" checklists but also discussing the emotional impact experienced by client families. Defense mechanisms like denial and anger impede patients from gaining requisite knowledge, and must be overcome. Dondorp and de Wert extend this theme by their treatise on ethical considerations.

Our authors have thus provided us with a text broad in coverage. Contributions by an international spectrum of authors – European and American – assure us provincial views are eschewed. Here we have a text that students, practitioners, and scientists involved in reproduction genetics should have on their shelves, or readily accessible in their computer.