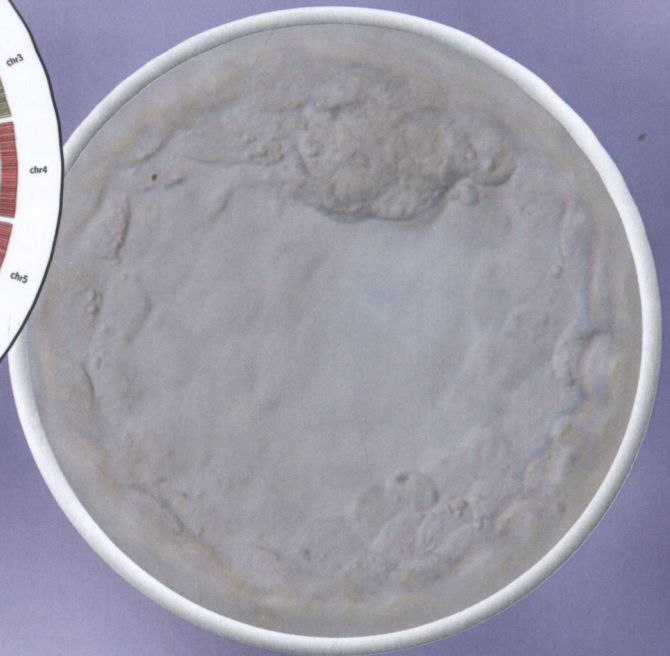
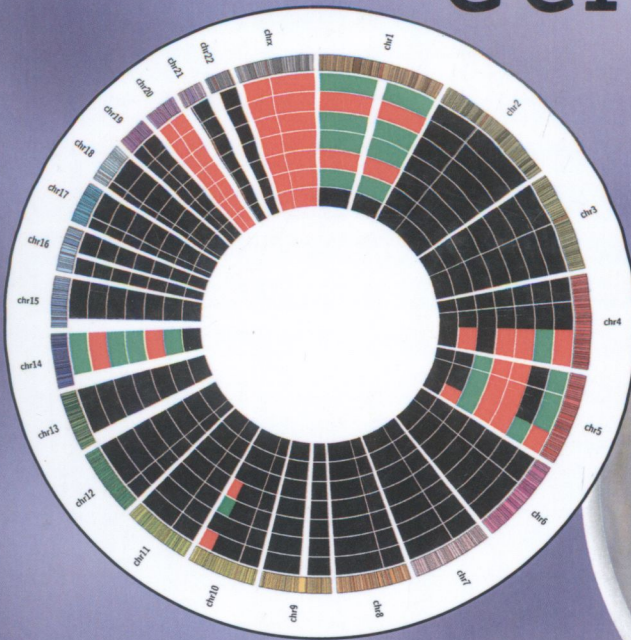


Textbook of **Human
Reproductive
Genetics** **SECOND
EDITION**



Edited by **Stéphane Viville**
Karen Sermon

CAMBRIDGE

Medicine

Textbook of Human Reproductive Genetics

SECOND EDITION

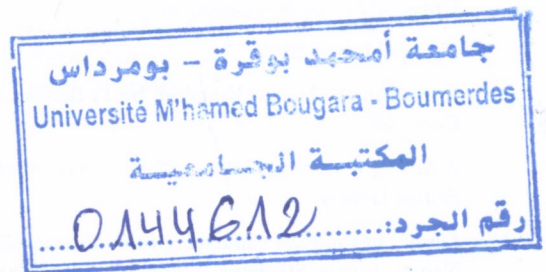
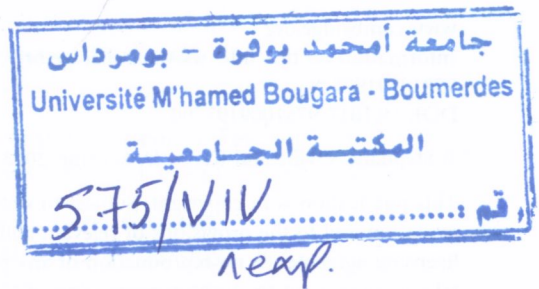
Edited by

Stéphane Viville

Institut de Génétique et de Biologie Moléculaire et Cellulaire, Strasbourg

Karen D. Sermon

Research Group Reproduction and Genetics, Vrije Universiteit Brussel



CAMBRIDGE
UNIVERSITY PRESS

Contents

List of Contributors vii

-
- | | |
|---|--|
| <p>1 Basic Genetics and Cytogenetics:
A Brief Reminder 1
Karen D. Sermon</p> <p>2 Application of Whole-Genome
Technologies to Assisted
Reproductive Treatment 16
Elia Fernandez Gallardo, Thomas Lefevre,
Koen Theunis, and Thierry Voet</p> <p>3 Meiosis: How to Get a Good Start in Life 31
Ursula Eichenlaub-Ritter</p> <p>4 Chromosomes in Early Human Embryo
Development: Incidence of Chromosomal
Abnormalities, Underlying Mechanisms,
and Consequences for Diagnosis
and Development 48
Esther B. Baart, Effrosyni A. Chavli, and
Diane Van Opstal</p> <p>5 DNA Is Not the Whole Story:
Transgenerational Epigenesis
and Imprinting 68
Ashwini Balakrishnan and J. Richard Chaillet</p> <p>6 Genes Are Not the Whole Story:
Retrotransposons as New Determinants
of Male Fertility 83
Patricia Fauque and Déborah Bourc'h</p> <p>7 Chromosomal Causes of Infertility 99
Svetlana A. Yatsenko and Aleksandar Rajkovic</p> | <p>8 Genetics of Human Male Infertility: The Quest
for Diagnosis and Treatment 116
Stéphane Viville and Özlem Okutman</p> <p>9 Genetics of Human Female Infertility 132
Svetlana A. Yatsenko and Aleksandar Rajkovic</p> <p>10 Preconception Genetics Analysis/Screening
in IVF 142
Juan José Guillén and Rita Vassena</p> <p>11 Genetic Counseling in Assisted
Reproductive Treatment 155
Christine de Die-Smulders and Ron van Golde</p> <p>12 Mitochondrial Genetics in
Reproductive Medicine 169
Claudia Spits and Filippo Zambelli</p> <p>13 Preimplantation Genetic Testing 185
Jan Traeger-Synodinos and Carmen Rubio</p> <p>14 Epigenetics and Assisted
Reproductive Technology 202
Aafke P.A. van Montfoort</p> <p>15 Human Reproductive Genetics in Medically
Assisted Reproduction:
Ethical Considerations 219
Guido de Wert and Wybo Dondorp</p> |
|---|--|
-
- Index* 237

A basic understanding of human genetics is vital for all those working in the field of assisted human reproduction. Genetic makeup can hamper reproduction, and insight into this is making genetic diagnosis and counselling increasingly important. This fully updated textbook continues the clear structure of the original edition, beginning with a chapter on the basics of genetics and cytogenetics. Genetic causes of infertility and the effect of epigenetics and transposons on fertility are discussed in detail. Several new chapters are included in this edition, reflecting the advances of the field, including preconception genetic analysis and screening in IVF and mitochondrial genetics. Combining genetics, reproductive biology, and medicine, this is an essential text for practitioners in reproductive medicine and geneticists involved in the field looking to improve their knowledge of the subject and provide outstanding patient care.

Stéphane Viville is a professor at the University of Strasbourg. He leads a laboratory of fundamental research at the Laboratoire de génétique médicale (UMRS-1112) Strasbourg. He is a former chair of the ESHRE Reproductive Genetics Special Interest Group.

Karen D. Sermon is Head of the Reproduction & Genetics Research Group, Vrije Universiteit Brussel, Brussels. She is a former chair of the ESHRE Reproductive Genetics Special Interest Group and is currently chair-elect for ESHRE (2021–2023) and chair (2023–2025).

Cover designed by James F. Brisson

Printed in the United Kingdom



CAMBRIDGE
UNIVERSITY PRESS

ISBN 978-1-00-919772-4



9 781009 197724 >