1. Record Nr. UNINA9910383821003321 Autore Arafa Mohamed. **Titolo** Genetics of male infertility: a case-based guide for clinicians / / Mohamed Arafa [and three others] Cham, Switzerland: ,: Springer, , [2020] Pubbl/distr/stampa **ISBN** 3-030-37972-8 Edizione [First edition.] Descrizione fisica 1 online resource (XVII, 375 pages 54 illustration, 51 illustration in color.) 612.6 Disciplina Soggetti Reproductive health Human genetics Infertility - Genetic aspects Lingua di pubblicazione Inglese **Formato** Materiale a stampa Livello bibliografico Monografia Note generali Includes index. Part I: The Genetic Basis of Reproduction -- The Molecular Genetics of Nota di contenuto Testis Determination -- Molecular Regulation of Sperm Production Cascade -- Spermatozoal Chromatin Structure: Role in Sperm Functions and Fertilization -- Genetic Basis of Endocrine Regulation of Spermatogenesis -- Epidemiology of Genetic Disorders in Male Infertility -- Genetic Evaluation of Male Infertility -- Part II: Impact of Genetics on the Sperm Cell -- Genetic Basis of Sperm Morphologic Defects: Head Defects. Body and Tail Defects -- Mitochondrial Function and Male Infertility -- Sperm DNA Fragmentation and Male Infertility --The Sperm Epigenome and Potential Implications for the Developing Embryo -- Part III: Clinical Case Scenarios -- Klinefelter Syndrome --Chromosomal Translocations and Inversion in Male Infertility --Genetics of Vas Aplasia -- Anomalies of the Y Chromosome --Infertility and Cryptorchidism -- Kartagener and Immotile Cilia Syndrome -- Persistent Müllerian Duct Syndrome -- Disorders of Sex Determination -- Endocrine Genetic Defects -- Sperm Aneuploidy --Part IV: Genetic Infertility: Is There Any Hope?- Sperm DNA Fragmentation -- The Potential of CRISPR/Cas Gene Editing to Correct

This unique, case-based guide provides a thoughtful and

comprehensive overview of the genetic basis of male infertility for the

Male Infertility.

Sommario/riassunto

practicing clinician. In addition to discussing the molecular foundations of sperm production and the consequences of genetic abnormalities on various stages of sperm development, it examines the clinical aspects of acknowledged genetic disorders and their implications on male fertility. In so doing, it offers the necessary tools required by the clinician for the diagnosis and treatment of infertile men with genetic abnormalities. Moreover, it provides essential algorithms that may aid in counseling patients in the clinic. The text is arranged in four thematic sections for easy reference. The genetic foundation of male reproduction is presented in part 1, including regulation of sperm production, the structure of sperm chromatin, and spermatogenesis. The impact of genetic abnormalities on male infertility is the subject of part 2, covering sperm defects, mitochondrial function and DNA fragmentation. The clinical case material in part 3 illustrates real-world examples of genetic etiologies and the current diagnostic and therapeutic strategies for conditions such as vas asplasia, cryptorchidism, immotile cilia syndrome, sperm aneuploidy and other challenging scenarios. Casting forward, the fourth and final section presents an overview of future possibilities for management of genetic causes of male infertility, including gene editing. Fully exploring the clinical context of these genetic conditions in a practical manner that appeals to the practicing clinician, Genetics of Male Infertility is an exciting and essential text for reproductive medicine specialists, andrologists, urologists, researchers and all other clinicians treating infertile patients.