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Nota di contenuto	Dedication; Title page; Copyright; Preface; Acknowledgements; List of Contributors; 1 Genetic Counseling: Preconception, Prenatal, and Perinatal; Incidence, prevalence and burden of genetic disorders and congenital malformations; The goal and purpose of prenatal diagnosis; Prerequisites for genetic counseling; Guiding principles for genetic counseling; Preconception genetic counseling; Genetic disorders that threaten maternal health; Maternal genetic disorders that may threaten fetal health and survival; Genetic disorders that pregnancy may aggravate; A history of infertility Parental carrier of a genetic disorder A family history of a genetic disorder; Consanguinity; Environmental exposures that threaten fetal health; Identification of preconception options; Genetic counseling as a prelude to prenatal diagnosis; Genetic counseling when the fetus is affected; Perinatal genetic counseling; References; 2 Amniocentesis, Chorionic Villus Sampling, and Fetal Blood Sampling; Introduction; Amniocentesis; Chorionic villus sampling; Fetal blood sampling;

References; 3 Amniotic Fluid Constituents, Cell Culture, and Neural Tube Defects; Introduction; Amniotic fluid
Amniotic fluid cell culture Prenatal diagnosis of neural tube defects (NTDs); References; Additional References; 4 Prenatal Diagnosis of Chromosomal Abnormalities through Chorionic Villus Sampling and Amniocentesis; The incidence of chromosomal abnormalities detected by conventional cytogenetics; Indications for prenatal cytogenetic diagnosis; Interpretation issues: chromosome mosaicism and pseudomosaicism; Interpretation issues: chromosome rearrangements; Interpretation issues: chromosome polymorphisms, common inversions, and other structural variations
Interpretation issues: maternal cell contamination Factors affecting diagnostic success rate and accuracy; Technical standards for prenatal cytogenetics laboratories; Conclusion; Acknowledgments; References; 5 Prenatal Diagnosis of Sex Chromosome Abnormalities; Incidence; Patterns of inheritance; Prenatal diagnosis; Turner syndrome; Klinefelter syndrome; Triple X and poly-X syndromes; 47,XYY males; Structural abnormalities of the X chromosome; Structural abnormalities of the Y chromosome; Disorders of sex development; Ovotesticular disorders of sex development; Conclusion; References
6 Molecular Cytogenetics and Prenatal Diagnosis Microdeletions; Subtle/cryptic rearrangements; Identification of marker chromosomes; Structural rearrangements: duplications; Prenatal diagnosis: interphase analysis; Chorionic villus samples; Interphase studies: fetal cells in maternal blood; Interphase analysis: transcervical and uterine cavity samples; Interphase analysis: preimplantation genetic diagnosis; Conclusion; References; 7 Prenatal Diagnosis and the Spectrum of Involvement from Fragile X Mutations; Introduction; Epidemiology; Clinical involvement in those with the full mutation
Clinical phenotype in the premutation