1. Record Nr. UNINA990005795230403321

Autore	De Sanctis, Gaetano <1870-1957>
Titolo	1.1.: La conquista del primato in Italia / Gaetano De Sanctis
Pubbl/distr/stampa	Milano : F.lli Bocca, 1907
Descrizione fisica	458 p. ; 22 cm
Collana	Biblioteca di scienze moderne ; 32
Lingua di pubblicazione	Italiano
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	La bibl. possiede il vol. 1

2. Record Nr. UNINA990006003100403321

Autore	Stevens, Edward F.
Titolo	Shipping practice with a consideration ot the law relating thereto / Edward F. Stevens ; with a Foreword by Essendon
Pubbl/distr/stampa	London, : Isaac Pitman & Sons, 1957
Edizione	[8th ed.]
Descrizione fisica	X, 172 p. ; 22 cm
Disciplina	343.096
Locazione	FGBC
Collocazione	VIII N 237
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia

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Autore	Bourn David
Titolo	Diagnostic genetic testing : core concepts and the wider context for human DNA analysis / / David Bourn
Pubbl/distr/stampa	Cham, Switzerland : , : Springer, , [2022] ©2022
ISBN	9783030855109 9783030855093
Descrizione fisica	1 online resource (145 pages)
Disciplina	573.21
Soggetti	Human genetics Medicine Bioethics Genetic Testing Bioethical Issues Molecular Diagnostic Techniques
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di contenuto	Intro -- Foreword -- Preface -- Further Reading -- Laboratory Techniques -- Genetic Disorders -- General Overviews of Genomic Testing in Healthcare -- Patient Support Groups -- Acknowledgements -- Contents -- About the Author -- Abbreviations -- 1 Genetic Testing, Some Themes and Some Basics -- Genetic Testing -- Complexity: Genes and Environment -- Risk and Uncertainty -- DNA and Categorisation -- Future Promises and Concerns -- Genetics in Other Areas of Medicine -- Basic Concepts in Genetics -- DNA Stores Information that Can Be Copied -- Genomic Architecture -- Gene Function and Organisation Within Genomes -- Transcription, Translation and the Genetic Code -- Mutation -- Patterns of Inheritance: Autosomal Dominant and Autosomal Recessive -- X-linked Inheritance -- Some Basics of Genetic Testing -- Isolation of DNA -- Finding Mutations -- Finding a Complementary Sequence -- The Polymerase Chain Reaction and DNA Amplification -- Electrophoresis -- DNA Sequencing -- 2 Autosomal Dominant

Inheritance and Huntington Disease -- Huntington Disease -- A Very Specific Genetic Error -- A Gain of Function -- Why Expansions? -- Determinism, but with Complications -- Anticipation -- Genetic Testing for HD -- Test Sensitivity and Specificity -- The Value of Genetic Testing for HD -- Laboratory Errors -- Genetic Information and Families -- 3 Autosomal Recessive Inheritance and Cystic Fibrosis -- Contrasting Dominant and Recessive Conditions -- Cystic Fibrosis -- Many Different Genetic Errors: Some with Variable Effects -- Common Recessive Disorders -- Genetic Testing for CF -- The Value of Genetic Testing in CF -- Prenatal Diagnosis -- Therapies for CF and Genetic Testing -- Calculating Risks -- Scenario 1 -- Scenario 2 -- 4 X-linked Inheritance: A Question of Gender -- A Fundamental Imbalance -- Switching Off Genes on the Inactive X Chromosome. Inheritance of X-linked Genetic Disorders -- Three X-linked Genes Associated with Genetic Disorders -- X-linked Example 1: The DMD Gene and Duchenne Muscular Dystrophy/Becker Muscular Dystrophy -- The Spectrum and Significance of Mutations in the DMD Gene -- Testing for DMD Gene Mutations -- X-linked Example 2: The FMR1 Gene, Fragile X Syndrome and Other Phenotypes -- Multiple Conditions Are Associated with FMR1 Gene Mutations -- Transmission of Fragile X Syndrome -- Testing for FMR1 Gene Mutations -- X-linked Example 3: The Androgen Receptor (AR) Gene, Spinal and Bulbar Muscular Atrophy and Androgen Insensitivity Syndrome -- SBMA: A Trinucleotide Expansion Disorder -- AIS: Loss of Function Mutations in the AR Gene -- Genetics and Gender -- 5 Genetic Testing in Cancer -- Cancer as a Genetic Disease -- Inherited Cancer Predisposition -- Tumor Suppressor Genes -- BRCA1 and BRCA2 as Tumor Suppressor Genes -- Oncogenes -- Cytogenetics and Cancer Testing -- Chromosome Analysis -- The Philadelphia Chromosome -- Rapid Detection of Specific Gene Fusions and Other Chromosomal Rearrangements in Cancers by FISH -- Genetic Testing in Cancer Diagnosis and Treatment -- 6 DNA Testing, Genetics and Identity -- Identity Testing in the Diagnostic Genetic Laboratory -- Diagnostic Applications for Genetic Identity Testing -- Direct Testing of Identity -- Family Relationships -- Identity in Twins -- Avoiding Errors in Prenatal Diagnosis -- Monitoring Bone Marrow Transplants -- DNA as a Marker of Unique Personal Identity -- Widening Circles -- Identity as a Member of Humanity -- 7 Out of Sequence: Genome-Scale Testing -- Whole Genome Analyses -- Sanger Sequencing -- Diagnostic Sanger Sequencing Applications -- Next-Generation Sequencing (NGS) -- Diagnostic Applications of New Sequencing Technologies -- Trio Analysis and New Mutations -- Genomic Analysis in Cancer. NGS, Clonal Sequencing and Finding a Needle in a Haystack -- Finding New Disease Associations -- Comparative Genomics -- Third-Generation (Long-Read) Sequencing -- Limitations to the Utility of Genome-Scale Sequencing -- Confounding Factors: Complexity of Common Disease -- Confounding Factors: Lots of Variation, Many Rare Variants -- Confounding Factors: Complex Metabolic Networks -- Epigenetic Regulation: A Further Level of Complexity -- The Risk of False Positives -- Will WGS Improve Outcomes for Common Disorders? -- Genomic Testing in Mainstream Medicine: Because We Can Rather Than Because We Should? -- 8 DNA Testing: Pulling the Strands Together -- Diagnostic Genetics and Ethical Principles -- Consent in the Genomic Era -- Making Genetic Choices -- Compartmentalisation on Genetic Grounds -- Commercial Access to Genetic Testing -- Acknowledging Uncertainties and Avoiding Error -- The Value of Genetic Testing -- The Language of Genetics: Uses and Misuses -- Genetics and Society.