Record Nr. UNINA9910817394203321 Rosenberg's molecular and genetic basis of neurological and psychiatric **Titolo** disease / / edited by Roger N. Rosenberg, Juan M. Pascual Pubbl/distr/stampa London, England:,: Academic Press,, 2015 ©2015 **ISBN** 0-12-410549-1 Edizione [Fifth edition.] 1 online resource (1465 p.) Descrizione fisica 616.80442 Disciplina Soggetti Nervous system - Diseases - Molecular aspects Nervous system - Diseases - Genetic aspects Lingua di pubblicazione Inglese **Formato** Materiale a stampa Livello bibliografico Monografia Note generali Description based upon print version of record. Nota di bibliografia Includes bibliographical references at the end of each chapters and index. Nota di contenuto Front Cover; Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease; Copyright; Dedications; Contents; Preface to the Fifth Edition; Contributors; Section I: General Concepts and Tools; Chapter 1: Mendelian, Non-Mendelian, Multigenic Inheritance, and Epigenetics; Introduction; Mendelian traits; Mendel's Laws; Chromosomes and Genes; Mendelian Inheritance; Molecular Pathomechanisms of Mutations: Factors That Modify Classic Mendelian Inheritance Patterns: New Mutations, Mosaicism, and Somatic Mutations; Penetrance and Expressivity; Repeat expansion disorders Non-mendelian inheritanceMitochondrial Inheritance; Imprinting; Uniparental Disomy; Imprinting, UPD, and Genetic Disorders: Chromosomal and genomic disorders; Aneuploidy; Isochromosomes; Translocations; Intrachromosomal Rearrangements; Mechanisms for Formation of Chromosomal Rearrangements: Nonallelic homologous recombination (NAHR); Nonhomologous end joining (NHEJ); Fork stalling and template switching/microhomology-mediated breakinduced replication (FoSTeS/MMBIR); How Chromosomal Rearrangements Confer Phenotypes; Assays for Chromosomal and Genomic Disorders; Multigenic inheritance Digenic InheritanceModifier Genes; Complex traits; Genetic Features of

Complex Traits; Assessing Variation in the Human Genome; Genetic

Variation and Complex Traits; Examples of Susceptibility Genes for Complex Traits: Epigenetics: DNA Methylation and Hydroxymethylation: Histone Modifications and Higher Order Chromatin Remodeling: Noncoding RNA Regulation; The human genome: High-throughput technologies; Conclusions; References; Chapter 2: Genotype-Phenotype Correlations; Introduction; Single phenotype: Multiple genes; Single gene: Multiple phenotypes; Neuronal/cellular selective vulnerability Highly variable systemic phenotypesPenetrance and age of onset; Conclusion and future directions; References; Chapter 3: Immunogenetics of Neurological Disease; Introduction; Epidemiological evidence for genetic susceptibility; Genetics of MS: Family-based investigations: The role of major histocompatibility complex genes: Other immune-related genes: The environment and immune-related genes; Conclusion; References; Chapter 4: Pharmacogenomic Approaches to the Treatment of Sporadic Alzheimer Disease using Cholinomimetic Agents; Introduction; Genetic risk factors and sporadic alzheimer disease

Genetic risk factors, cholinergic dysfunction, and alzheimer diseaseApoE4 and cholinomimetic drugs in alzheimer disease; Experimental drugs and their relationship to the apoE4 allele; Acetylcholinesterase and butyrylcholinesterase genetic variants in dementia; Acknowledgements; References; Chapter 5: Application of Mouse Genetics to Human Disease: Generation and Analysis of Mouse Models; Generation and Analysis of Mouse Models; Introduction; Creating mouse models; Transgenesis; Gene Targeting; Random Mutagenesis; Phenotypic analysis of mouse models; Summary; References

Chapter 6: DNA Sequencing and Other Methods of Exonic and Genomic Analyses

Sommario/riassunto

Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease, Fifth Edition provides a comprehensive introduction and reference to the foundations and key practical aspects relevant to the majority of neurologic and psychiatric disease. A favorite of over three generations of students, clinicians and scholars, this new edition retains and expands the informative, concise and critical tone of the first edition. This is an essential reference for general medical practitioners, neurologists, psychiatrists, geneticists, and related professionals, and for the neuroscience and neur