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Titolo	Assessing Rare Variation in Complex Traits : Design and Analysis of Genetic Studies / / edited by Eleftheria Zeggini, Andrew Morris
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Nota di bibliografia	Includes bibliographical references.
Nota di contenuto	Calling Rare Variants from Genotype Data -- Calling Variants from Sequence Data -- Rare Variant Quality Control -- Rare Structural Variants -- Functional Annotation of Rare Genetic Variants -- The 1000 Genomes Project -- The UK10K Project -- Population Isolates -- Natural Selection at Rare Variants -- Collapsing Approaches for the Association Analysis of Rare Variants -- Rare Variant Association Analysis: Beyond Collapsing Approaches -- Significance Thresholds for Rare Variant Signals -- Power of Rare Variant Aggregate Tests -- Replicating Sequence-based Association Studies of Rare Variants -- Meta-analysis of Rare Variants -- Population Stratification of Rare Variants -- Use of Appropriate Controls in Rare Variant Studies -- Trans-ethnic Fine-mapping of Rare Causal Variants.
Sommario/riassunto	This unique volume is the first to cover a wide range of design and analysis issues in genetic studies of rare variants, with contributions from experts in the field, through large-scale international consortia including the UK10K Project, GO-T2D and T2D-GENES. The book presents state-of-the-art methodology for rare variant detection and

calling, imputation, and analysis in samples of unrelated individuals and families. It also covers analytical issues associated with the study of rare variants, such as the impact of fine-scale population structure, and rare variants studies in a meta-analysis framework. This book covers multiple aspects of study design, analysis and interpretation for complex trait studies focusing on rare sequence variation. In many areas of genomic research, including complex trait association studies, technology is in danger of outstripping our capacity to analyze and interpret the vast amounts of data generated. The field of statistical genetics in the whole-genome sequencing era is still in its infancy, but as this book illustrates, powerful methods to analyze the aggregation of low-frequency and rare variants are now starting to emerge. The chapter Functional Annotation of Rare Genetic Variants is available open access under a Creative Commons Attribution 4.0 International License via link.springer.com.