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Sommario/riassunto

This eBook contains the 19 articles that were part of a Special Topic in Frontiers in Genetics entitled "Genetics Research in Electronic Health Records Linked to DNA Biobanks". The Special Issue was published online in 2014-2015 and contained papers representing the diverse research ongoing in the integration of electronic health records (EHR) with genomics through basic, clinical, and translational research. We have divided the eBook into four Chapters. Chapter 1 describes the Electronic Medical Records and Genomics (eMERGE) network and its contri-bution to genomics. It highlights methodological questions related to large data sets such as imputation and population stratification. Chapter 2 describes the results of genetic studies on different diseases for which all the phenotypic information was extracted from the EHR with highly specific ePhenotyping algorithms. Chapter 3 focuses on more complex analyses of the genome including copy number variants (CNV), pleiotropy com-bined with phenome-wide association studies (PheWAS), and epistasis (gene-gene interactions). Chapter 4 discusses the use of genetic data together with EHR-derived clinical data in clinical settings, and how to return genetic results to patients and providers. It also contains a comprehensive review on genetic risk scores. We have included mostly Original Research Articles in the eBook, but also Reviews and Methods papers on the relevant topics of analyzing and integrating genomic data. The release of this

eBook is timely, since several countries are launching Precision Medicine initiatives. Precision Medicine is a new concept in patient care taking into account individual variability in genetic, environmental and lifestyle factors, when treating diseases or trying to prevent them from developing. It has become an important focus for biomedical, clinical and translational informatics. The papers presented in this eBook are well positioned to educate the readers about Precision Medicine and to demonstrate the potential study designs, methods, strategies, and applications where this type of research can be performed successfully. The ultimate goal is to improve diagnostics and provide better, more targeted care to the patient.