

The electronic health record as a primary source of clinical phenotype for genetic epidemiological studies

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The electronic health record (EHR) is a longitudinal electronic record of a patient's health information generated by one or more encounters in any care delivery setting. Included in this information are patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, laboratory data, and radiology reports. The EHR automates and streamlines the clinician's work flow. It has the ability to generate a complete record of a clinical patient encounter, including evidence-based decision support, quality management, and outcomes reporting. The EHR system is thus thought to increase physician efficiency and to reduce costs, as well as to promote standardization of care. The main advantages of the EHR include the following: (1) Information can be continuously updated. (2) Data from the EHR system can be used anonymously in statistical evaluation for purposes of quality improvement, outcome reporting, resource management, and public health surveillance of communicable diseases. (3) Information can be exchanged between different EHR systems, facilitating the coordination of health care delivery in nonaffiliated facilities. Through use of the EHR, potential participants of clinical trials may be more readily identified, administrative overhead costs may be lessened, data errors may be reduced, and adverse outcomes may be more rapidly detected. Clinicians and researchers may thus benefit from use of the EHR for data collection and analysis in clinical trials.

In this issue of *Genomic Medicine*, Wood et al. (2008) make use of the EHR as a potential rich source of clinical phenotype data in a genetic epidemiological study. Accurate clinical phenotype data are essential for such studies, and the authors were successful in implementing and integrating data collection and analysis. The results presented in the paper support the feasibility of this approach and thus provide an important example of an EHR-based genetic epidemiological study. As the authors point out, a potential limitation to utilization of EHR-derived data for genetic epidemiology is the quality of the data, which is influenced by different practices among staff and clinicians. Data collection may also vary among different institutions. Despite this limitation, EHR-based data are likely to prove useful as the primary source of clinical phenotype information for genetic epidemiological studies.

Reference

Wood GC, Still CD, Chu X, Susek M, Erdman R, Hartman C, Yeager S, Blosky MA, Krum W, Carey DJ, Skelding KA, Benotti P, Stewart WF, Gerhard GS (2008) Association of chromosome 9p21 SNPs with cardiovascular phenotypes in morbid obesity using electronic health record data. *Genomic Med* doi:[10.1007/s11568-008-9023-z](https://doi.org/10.1007/s11568-008-9023-z)

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