RARE VISIBLE DISORDERS/ DISEASES AS INDIVIDUALLY IDENTIFIABLE HEALTH INFORMATION

Tewodros Eguale, M.D, MSc; Gillian Bartlett, PhD; Robyn Tamblyn, PhD
Clinical and Health Informatics Research Group, McGill University, Montreal, Quebec, Canada

Abstract

Individually identifiable health information needs to be masked or deleted in the secondary use of data for research. We defined and identified rare visible disorders as part of de-identification of individually identifiable health information. The prevalence of these disorders was evaluated using an administrative database and conformed to our definition. Due emphasis should be given to rare visible disorders in the de-identification process of health information.

Introduction

Privacy is a fundamental right and needs to be protected. Electronic health information, which is originally collected in activities of provision of medical care, is increasingly available for research. This secondary use of data for research has desirable features including 1) more timely clinical and epidemiological data to assess treatment quality and economics of health care and 2) substantial reduction in cost and time to the researcher. However, these databases and records usually contain individually identifiable health information that can be used to identify the individual. These identifiers need to be masked or removed when data are used for research purpose without explicit informed consent. Individually identifiable health information has been defined by legislation in the USA - Health Insurance Portability and Privacy Act (HIPPA), UK - Medical Research Council (MRC), the European Union and others. In addition, MRC identified rare disease or condition of disease, 9th edition, Clinical Modification (ICD-9-CM) list for rareness and visibility. An extensive literature search was conducted using medical text books, published literatures, online materials, governmental and non-governmental rare disease sites to identify diseases which qualify for rareness and visibility. An administrative database from the province of Quebec (Canada) with universal health coverage was used to estimate the prevalence of the identified rare visible diseases.

Results

A total of 410 ICD-9-CM codes were identified as rare visible diseases representing 5.9% of ICD-9-CM codes. These diseases include tumors, endocrine disorders, orofacial abnormalities, infectious diseases, gait, posture and stature disorders, disorders of the extremities, developmental malformations, metabolic disorders, central nervous system abnormalities, peripheral nervous system disorders, and late effects of trauma. Majority of the codes (43.6%) are in the category of congenital anomalies.

Overall, 87129 patients with rare visible disorder were identified from the database containing 4.25 million people representing 2.04% of the population. The majority of the disorders (88.5%) have a prevalence of 1 or less per 10,000 populations, while 9.5% of the disorders have rate from 1 to 5 per 10,000 populations. Approximately, 0.5% of the rare visible disorders have prevalence of 10 - 11 per 10,000 populations.

Conclusion: Two percent of the population had rare visible disorder. Almost all (99.5%) of the rare visible disorders had a prevalence of less than 10 per 10,000 of the study populations.

Recommendation: Rare visible diseases need to be identified in the context of a study population and should be treated as any individually identifiable health information in the de-identification of health information to protect the privacy of individuals.

Contact: tewodros.eguale@mail.mcgill.ca