USE OF ELECTRONIC HEALTH RECORDS TO DEVELOP A MORE ROBUST PHENOTYPE OF PEDIATRIC OVERWEIGHT AND OBESITY

USO DE HISTORIAS CLÍNICAS ELECTRÓNICAS PARA DESARROLLAR UN FENOTIPO MÁS ROBUSTO DE SOBREPESO Y OBESIDAD PEDIÁTRICOS

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ABSTRACT

Clinical EHR data can be used to better develop a more robust phenotype of pediatric overweight and obesity.

Keywords: Overweight; obesity; electronic health record; eHealth; transdisciplinary; phenotype.

RESUMEN

Los datos clínicos de EHR se pueden utilizar para desarrollar mejor un fenotipo más sólido de sobrepeso y obesidad pediátricos.

Palabras clave: Sobrepeso; obesidad; historia clínica electrónica; eSalud; transdisciplinario; fenotipo.

Developing phenotypes, a term that classically refers to the "observable characteristics in an individual resulting from the expression of genes" or "the clinical presentation of an individual with a particular genotype," is an important pillar of science research. Recently, different fields, especially precision medicine, have endeavored to expand the classical understanding of a phenotype to focus not only on disease etiology but also treatment.

Deep phenotyping, the precise and comprehensive analysis of phenotypic abnormalities in which the individual components of the phenotype are observed and described, is an important pillar of precision medicine that seeks to both uncover subclasses of diseases and improve the translation of this knowledge into clinical care through more robust data collection and analysis. In fact, big data and such analyses are transforming all types of disease research (not just etiological research),[1] including in reducing the extreme lag that has been documented in translation research. This type of research typically has been conducted through genome-wide association studies (GWAS) research. With respect to overweight and obesity (OW/OB), such data has identified genes associated with obesity, the limitations of body mass index (BMI) as an indicator to detect overweight and obesity[2] and recent research has described how BMI may mask etiological differences between different types of obesity.[3] Importantly, electronic health record (EHR) data has not been included in existing deep phenotyping for pediatric OW/OB.

Digital phenotyping has been growing since the term was first introduced in 2015, as it may improve our ability to diagnose, treat, and manage an array of diseases, including OW/OB.[4] Digital phenotyping generally involves constant mobile sensing and collection of data through smartphones or other technology such as wearable smartwatches.[5] In a recent review, authors found 46 studies using digital phenotyping in the past three years, and 7 that were focused on OW/OB.[6] These studies focused on identifying phenotypes that could predict engagement in and health outcomes of various types of interventions.[7] Across the disease spectrum, researchers looked at a variety of wearable, digital and clinical variables, including sleep, movement, screen time, blood pressure, weight, and food intake. However, clinical EHR data such as international classification of disease (ICD) codes for visit diagnoses and active issues on the EHR problem list were not included. In fact, an immediate opportunity in digital phenotyping is to look at other sources for digital health data, especially using insights from commonly used technologies.[8]

As evidenced, deep phenotyping is more focused on genetic associations and digital phenotyping is more focused on measuring behavior from smartphone sensors, keyboard interaction, and various features of voice and speech in the status quo.[8] Given the nascency of digital phenotyping and the shared goal of using big data to develop machine-learning algorithms that better characterize disease etiology and treatment,[6] it is necessary to assess what role clinical patient data from the EHR may play in developing more accurate digital phenotypes.

Currently, such EHR data is employed in electronic phenotyping, or the process by which patients with a medical condition or characteristic are identified, to create computable phenotypes. In other words, EHR data is currently used to conduct queries and identify patients that facilitate building registries, conducting research trials, and conducting surveillance for harms and complications of rare disease.[9] Moreover, published OW/OB phenotypes have been limited to behavioral and genetic components. While it is documented that individuals with OW/OB likely have subtypes and are

phenotypically heterogenous, these are relatively new findings in a formative research stage that have yet to impact clinical practice.(10)

Transdisciplinary research practices could guide these distinct fields to synergistically create a more robust phenotype of pediatric OW/OB, both in terms of etiology and treatment. Specifically, there is an opportunity to consider how existing EHR data like ICD codes for annual well child visit diagnoses and active issues on the EHR problem list could better identify OW/OB subtypes as well as predict enrollment and participation in weight management programs to improve health outcomes and equity. EHR data sources could serve as objective clinical proxies for access to healthcare or family history of disease; they might also serve as proxies for the quality of care a patient receives. Future research could conduct sensitivity analyses of multivariable regression models with both clinical and sociodemographic data that is captured in the EHR. Most importantly, researchers should note that clinical EHR data, such as the list of ICD codes from well child visits and issues in the problem list, may serve as important sources of data, which have not been included in building a more robust phenotype of pediatric OW/OB.

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