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Feasibility of Using Cerner Health Facts to Characterize Comorbid Down Syndrome and Autism Spectrum Disorder

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☐**Resident/Psychology Intern (≤ 1 month of dedicated research time)**
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****Fellow (DBP Fellow)**

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IRB Number: 17080492 (Non-Human Subjects Determination for Cerner Health Facts projects)

Describe role of Submitting/Presenting Trainee in this project (limit 150 words):

The presenting trainee serves as the primary investigator for this project. Initial tasks as a part of that role included meeting with faculty and subject matter experts to determine any knowledge gaps and to identify a focus for the project. This was followed by a background literature review and identifying Cerner Health Facts as a possible source after attending a presentation on the database. I then led an initial meeting with a data scientist to determine project feasibility and several subsequent meetings to review data and project updates. Along with the data scientist, I identified the phenotype code technique from the literature review and consolidated the data into systems-based categories. I also coordinated and led initial meetings with a biostatistician.

Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words

Background: Up to 19% of individuals with Down syndrome (DS) may also have Autism Spectrum Disorder (ASD), and it is unknown if the DS-ASD population faces unique medical and psychological comorbidities (Channell, et al., 2019). Research on DS-ASD is challenging given the low rates of occurrence and diagnostic overshadowing. Large healthcare databases present an opportunity to study individuals with rare conditions like DS-ASD, if coding in the electronic health record captures that phenotype. This project explores the feasibility of using Cerner Health Facts, a data warehouse comprised of 69 million patients spread over more than 150 medical centers, to characterize the DS-ASD population.

Objectives/Goal: The objective of the project is to determine whether a DS-ASD sample can be derived from Cerner Health Facts. Exploratory objectives also include initial phenotypic characterization as a proof-of-concept for using large databases to investigate DS-ASD and other rare conditions. Our working hypothesis is that Cerner Health Facts will allow us to identify patients with DS-ASD, from which basic phenotypic data can be derived, providing the basis for future comparisons with other populations.

Methods/Design: Queries were developed to extract all patient encounters (e.g., visits) in Cerner Health Facts that included ICD-9/10 codes related to DS for patients between the ages of 0 and 18. Unique identifiers were used to group encounters together for individual patients. All additional ICD codes present for individuals in the sample were extracted and subsequently aggregated into Compound Phenotype codes based on the biological system (Denny et al., 2013). Similar processes were completed for patients with ICD codes associated with ASD, Intellectual Disability (ID), and Attention-Deficit/Hyperactivity Disorder (ADHD) to establish rates of co-occurrence and develop approaches to phenotypic characterization.

Results: A total of 120,616 encounters meeting inclusion criteria were derived, representing the health care of 22,862 individual patients with DS. Of these, 1,087 (4.8%) also had ASD. Figure 1 displays rates of overlap between individuals with codes for DS, ASD, ADHD, and ID in their records. A total of 6,425 unique ICD codes were associated with patients with DS-ASD, which consolidated to 28 Compound Phenotype codes.

Conclusions: Fewer individuals with DS had an identified comorbid ASD than expected in Cerner Health Facts, but the size of the database and population identified demonstrates that it is still a viable source for investigating the health and psychological outcomes of this population. Methodological challenges and data quality questions should be considered when using resources like Cerner Health Facts for the study of rare conditions.

Figure 1. Venn diagram of diagnostic combinations for Down syndrome, Autism Spectrum Disorder, ADHD, and Intellectual Disability from the Cerner Health Facts population.